myriad WOMEN'S HEALTH

African American Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 190	>99%	1 in 18,000
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 410	>99%	1 in 41,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 8	>99%	1 in 700
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 310	>99%	1 in 38,000

* For X-linked diseases, female carrier frequencies are presented.

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 97	99%	1 in 7,400
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 140	92%	1 in 1,700
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 140	>99%	1 in 14,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 62	>99%	1 in 6,100
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 6	>99%	1 in 530
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 420	>99%	1 in 42,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 71	>99%	1 in 7,000
Gamma-sarcoglycanopathy (SGCG)	1 in 340	87%	1 in 2,600
Gaucher Disease (GBA1)	1 in 120	60%	1 in 310
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 7	90%	1 in 61
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000
Hereditary Fructose Intolerance (ALDOB)	1 in 230	>99%	1 in 23,000

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Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 240	98%	1 in 17,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 260	>99%	1 in 26,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 400	97%	1 in 13,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 180	>99%	1 in 18,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 38	>99%	1 in 3,700
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	82%	1 in 430
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 110	>99%	1 in 11,000
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 60	>99%	1 in 5,900
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 200	>99%	1 in 20,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	< 1 in 500	>99%	< 1 in 50,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 180	>99%	1 in 18,000
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 66	71%	1 in 120
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000

* For X-linked diseases, female carrier frequencies are presented.

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Ashkenazi Jewish Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 55	>99%	1 in 5,400
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 190	94%	1 in 3,100
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 390	>99%	1 in 39,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	1 in 140	>99%	1 in 14,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 24	>99%	1 in 2,300
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 440	>99%	1 in 60,000
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	1 in 110	>99%	1 in 11,000
Calpainopathy (CAPN3)	< 1 in 500	99%	< 1 in 38,000
Canavan Disease (ASPA)	1 in 55	98%	1 in 2,700
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 47	>99%	1 in 4,600
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 62	>99%	1 in 6,100
Congenital Amegakaryocytic Thrombocytopenia (MPL)	1 in 76	>99%	1 in 7,500
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 62	>99%	1 in 6,100
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 24	>99%	1 in 2,300
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	1 in 94	>99%	1 in 9,300
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	1 in 250	92%	1 in 2,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	1 in 10	>99%	1 in 870
Familial Dysautonomia (ELP1)	1 in 31	>99%	1 in 3,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 45	>99%	1 in 4,400
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	1 in 230	>99%	1 in 23,000
Familial Mediterranean Fever (MEFV)	1 in 98	>99%	1 in 9,700
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	1 in 94	>99%	1 in 9,300
FKRP-related Disorders (FKRP)	< 1 in 500	>99%	< 1 in 50,000
FKTN-related Disorders (FKTN)	1 in 64	>99%	1 in 6,300
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 160	>99%	1 in 16,000
Gamma-sarcoglycanopathy (SGCG)	1 in 340	87%	1 in 2,600
Gaucher Disease (GBA1)	1 in 14	95%	1 in 250
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	50%	1 in 20
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 36	>99%	1 in 3,500
Glycogen Storage Disease, PFKM-related (PFKM)	1 in 250	>99%	1 in 25,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 68	>99%	1 in 6,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000

* For X-linked diseases, female carrier frequencies are presented.

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myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	85%	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	1 in 240	>99%	1 in 23,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 26	>99%	1 in 2,500
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	1 in 97	>99%	1 in 9,600
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 470	98%	1 in 32,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 320	>99%	1 in 32,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 98	>99%	1 in 9,700
Maple Syrup Urine Disease Type II (DBT)	1 in 180	97%	1 in 5,900
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 110	>99%	1 in 11,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	>99%	< 1 in 50,000
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000

* For X-linked diseases, female carrier frequencies are presented.

Ashkenazi Jewish

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	1 in 90	>99%	1 in 8,900
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 54,000	89%	1 in 480,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	1 in 300	>99%	1 in 30,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 110	>99%	1 in 11,000
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 100	>99%	1 in 10,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 23	>99%	1 in 2,200
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	1 in 180	>99%	1 in 18,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 78	93%	1 in 1,200
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	1 in 120	>99%	1 in 12,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 220	>99%	1 in 22,000
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 57	>99%	1 in 5,600
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	1 in 250	>99%	1 in 25,000
Retinitis Pigmentosa, EYS-related (EYS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	1 in 100	99%	1 in 7,300
Sandhoff Disease (HEXB)	< 1 in 500	98%	< 1 in 29,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 95	>99%	1 in 9,400
Spastic Paraplegia, TECPR2-related (TECPR2)	1 in 310	>99%	1 in 31,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 41	94%	1 in 350
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 130	>99%	1 in 13,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000

* For X-linked diseases, female carrier frequencies are presented.

Ashkenazi Jewish

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Eastern Asia Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	1 in 240	>99%	1 in 24,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 190	>99%	1 in 19,000
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	< 1 in 500	>99%	< 1 in 50,000
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 170	97%	1 in 6,300
Argininosuccinic Aciduria (ASL)	1 in 450	>99%	1 in 45,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 100,000	90%	< 1 in 1,000,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 10	>99%	1 in 900
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotinidase Deficiency (BTD)	1 in 460	>99%	1 in 67,000
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	1 in 450	>99%	1 in 45,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 320	>99%	1 in 31,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	60%	1 in 500
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 130	>99%	1 in 13,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 97	86%	1 in 700
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 67	88%	1 in 550
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 450	>99%	1 in 45,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	1 in 440	>99%	1 in 44,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 91	>99%	1 in 9,000
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 370	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	< 1 in 500	78%	< 1 in 2,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 140	>99%	1 in 14,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	1 in 420	>99%	1 in 42,000
Familial Mediterranean Fever (MEFV)	< 1 in 500	>99%	< 1 in 50,000
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 320	>99%	1 in 32,000
FKTN-related Disorders (FKTN)	1 in 95	10%	1 in 110
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	< 1 in 500	>99%	< 1 in 50,000
Galactosemia (GALT)	1 in 320	>99%	1 in 32,000
Gamma-sarcoglycanopathy (SGCG)	< 1 in 500	87%	< 1 in 3,800
Gaucher Disease (GBA1)	1 in 180	60%	1 in 450
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	30%	1 in 15
Glutaric Acidemia, GCDH-related (GCDH)	1 in 140	>99%	1 in 13,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 260	94%	1 in 4,200
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	92%	1 in 2,600
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000

* For X-linked diseases, female carrier frequencies are presented.

Eastern Asia

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 160	>99%	1 in 16,000
Homocystinuria, CBS-related (CBS)	< 1 in 500	>99%	< 1 in 50,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 240	>99%	1 in 24,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 390	>99%	1 in 39,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	1 in 400	>99%	1 in 40,000
Lysosomal Acid Lipase Deficiency (LIPA)	< 1 in 500	98%	< 1 in 34,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 430	>99%	1 in 43,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 240	>99%	1 in 23,000
Maple Syrup Urine Disease Type II (DBT)	1 in 470	97%	1 in 15,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 110	>99%	1 in 11,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 120	>99%	1 in 12,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 330	>99%	1 in 33,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	Not calculated due to rarity of disease in this individual's reported ethnicity	< 1 in 500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700

* For X-linked diseases, female carrier frequencies are presented.

Eastern Asia

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 29,000	89%	1 in 260,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 420	>99%	1 in 42,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 33	>99%	1 in 3,200
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 66	>99%	1 in 6,500
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	< 1 in 500	>99%	< 1 in 50,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 78	>99%	1 in 7,700

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 100	>99%	1 in 10,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 200	>99%	1 in 20,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 53	93%	1 in 630
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 450	>99%	1 in 44,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900

* For X-linked diseases, female carrier frequencies are presented.

Eastern Asia

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 130	>99%	1 in 12,000
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	97%	< 1 in 16,000
Wilson Disease (ATP7B)	1 in 66	>99%	1 in 6,500
Xeroderma Pigmentosum Group A (XPA)	1 in 100	>99%	1 in 10,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 19,000	77%	1 in 81,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

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WOMEN'S HEALTH

Finland Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 28	>99%	1 in 2,700
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 150	>99%	1 in 15,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 370	94%	1 in 6,000
Alport Syndrome, COL4A4-related (COL4A4)	1 in 370	>99%	1 in 36,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 18,000
Argininosuccinic Aciduria (ASL)	1 in 190	>99%	1 in 19,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	1 in 71	>99%	1 in 7,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 80	>99%	1 in 7,900
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 52	>99%	1 in 5,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 390	>99%	1 in 39,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	1 in 120	>99%	1 in 12,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 80	>99%	1 in 7,900
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	< 1 in 500	99%	< 1 in 38,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	1 in 370	>99%	1 in 37,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	1 in 76	>99%	1 in 7,500
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	60%	1 in 170
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 71	>99%	1 in 7,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	1 in 110	>99%	1 in 11,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	1 in 140	>99%	1 in 13,000
Cohen Syndrome (VPS13B)	1 in 160	97%	1 in 4,800
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 62	89%	1 in 560
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 71	>99%	1 in 7,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 80	>99%	1 in 7,900
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	1 in 450	>99%	1 in 45,000
Familial Mediterranean Fever (MEFV)	1 in 29	>99%	1 in 2,800
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 180	>99%	1 in 18,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	1 in 51	>99%	1 in 5,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 340	87%	1 in 2,600
Gaucher Disease (GBA1)	1 in 110	60%	1 in 260
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	50%	1 in 20
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 130	94%	1 in 2,100
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 130	>99%	1 in 12,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	96%	1 in 350,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	85%	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	1 in 51	>99%	1 in 5,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	< 1 in 500	98%	< 1 in 34,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 320	>99%	1 in 32,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 400	97%	1 in 13,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 110	>99%	1 in 11,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	1 in 48	>99%	1 in 4,700
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 210	>99%	1 in 21,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	1 in 46	>99%	1 in 4,500
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 71	>99%	1 in 7,000
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 23	>99%	1 in 2,200
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	1 in 130	>99%	1 in 13,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 180	>99%	1 in 18,000
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000
POMGNT1-related Disorders (POMGNT1)	1 in 110	98%	1 in 5,600
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000

* For X-linked diseases, female carrier frequencies are presented.

Finland

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	1 in 270	>99%	1 in 27,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 130	>99%	1 in 13,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 87	>99%	1 in 8,600
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 95	>99%	1 in 9,400
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 35	94%	1 in 560
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 120	>99%	1 in 12,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 130	>99%	1 in 13,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000

* For X-linked diseases, female carrier frequencies are presented.

Finland

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 8,500	98%	1 in 570,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

French Canadian or Cajun Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 28	>99%	1 in 2,700
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 23	>99%	1 in 2,200
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	1 in 24	>99%	1 in 2,300
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	1 in 22	99%	1 in 1,800
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 390	>99%	1 in 39,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 24	>99%	1 in 2,300
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	60%	1 in 170
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 62	96%	1 in 1,400
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 71	>99%	1 in 7,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 16	>99%	1 in 1,500
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 29	>99%	1 in 2,800
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 220	>99%	1 in 21,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 340	87%	1 in 2,600
Gaucher Disease (GBA1)	1 in 110	60%	1 in 260
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	50%	1 in 20
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 40	>99%	1 in 3,900
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000

* For X-linked diseases, female carrier frequencies are presented.

French Canadian or Cajun

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Fructose Intolerance (ALDOB)	1 in 81	>99%	1 in 8,000
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	85%	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 62	>99%	1 in 6,100
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 310	>99%	1 in 31,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	1 in 23	>99%	1 in 2,200
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 150	98%	1 in 10,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 290	>99%	1 in 29,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 400	97%	1 in 13,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	1 in 470	>99%	1 in 47,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 660	>99%	< 1 in 66,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 380	>99%	1 in 38,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 230	>99%	1 in 23,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000

* For X-linked diseases, female carrier frequencies are presented.

French Canadian or Cajun

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 35	>99%	1 in 3,400
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 83	>99%	1 in 8,200
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 56	97%	1 in 1,600
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 75	>99%	1 in 7,400
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 170	>99%	1 in 17,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 130	>99%	1 in 13,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 120	98%	1 in 6,500
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 95	>99%	1 in 9,400
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 35	94%	1 in 570
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 64	>99%	1 in 6,300
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 100	>99%	1 in 10,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000

* For X-linked diseases, female carrier frequencies are presented.

French Canadian or Cajun

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Hispanic Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 130	>99%	1 in 12,000
Alpha-mannosidosis (MAN2B1)	1 in 190	98%	1 in 13,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 290	>99%	1 in 29,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 390	>99%	1 in 39,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 26	>99%	1 in 2,500
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	70%	1 in 230
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 62	95%	1 in 1,200
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 140	>99%	1 in 14,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 53	>99%	1 in 5,200
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	1 in 320	96%	1 in 8,400
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

Hispanic

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 29	>99%	1 in 2,800
Fanconi Anemia Complementation Group A (FANCA)	1 in 250	92%	1 in 2,900
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 380	>99%	1 in 38,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 430	87%	1 in 3,300
Gaucher Disease (GBA1)	1 in 120	60%	1 in 310
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 15	90%	1 in 140
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	1 in 86	94%	1 in 1,400
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 100	>99%	1 in 10,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 160	98%	1 in 11,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 180	>99%	1 in 18,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 350	97%	1 in 12,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 120	>99%	1 in 12,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 27	>99%	1 in 2,600
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 72	>99%	1 in 7,100
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000

* For X-linked diseases, female carrier frequencies are presented.

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myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	98%	1 in 6,500
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 200	>99%	1 in 20,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 140	>99%	1 in 14,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 170	>99%	1 in 17,000
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 120	91%	1 in 1,100
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 180	98%	1 in 7,200
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000

* For X-linked diseases, female carrier frequencies are presented.

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myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

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myriad WOMEN'S HEALTH

Middle East Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	1 in 61	>99%	1 in 6,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 56	>99%	1 in 5,500
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	>99%	< 1 in 50,000
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 130	>99%	1 in 13,000
Alpha-mannosidosis (MAN2B1)	1 in 130	98%	1 in 8,700
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	1 in 160	>99%	1 in 16,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 110	96%	1 in 2,800
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 470	>99%	1 in 47,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	1 in 210	>99%	1 in 20,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 200	>99%	1 in 20,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	1 in 180	>99%	1 in 18,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 27	>99%	1 in 2,600
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 110	>99%	1 in 11,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 43	97%	1 in 1,300
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 140	>99%	1 in 14,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	1 in 51	>99%	1 in 5,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 30	>99%	1 in 2,900
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	1 in 350	96%	1 in 9,300
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

Middle East

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	1 in 110	>99%	1 in 11,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 67	>99%	1 in 6,600
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	1 in 410	>99%	1 in 40,000
Familial Mediterranean Fever (MEFV)	1 in 6	>99%	1 in 460
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 240	>99%	1 in 24,000
FKTN-related Disorders (FKTN)	1 in 460	>99%	1 in 46,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 95	87%	1 in 710
Gaucher Disease (GBA1)	1 in 120	60%	1 in 310
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 9	75%	1 in 31
Glutaric Acidemia, GCDH-related (GCDH)	1 in 66	>99%	1 in 6,500
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 97	94%	1 in 1,600
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 140	>99%	1 in 14,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000
Hereditary Fructose Intolerance (ALDOB)	1 in 98	>99%	1 in 9,700

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	1 in 180	>99%	1 in 18,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 41	>99%	1 in 4,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 84	>99%	1 in 8,300
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 160	>99%	1 in 16,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 33	98%	1 in 2,200
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 99	>99%	1 in 9,800
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 44	>99%	1 in 4,300
Maple Syrup Urine Disease Type II (DBT)	1 in 110	97%	1 in 3,500
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 68	>99%	1 in 6,700
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 76	>99%	1 in 7,500
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 48,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 80	97%	1 in 2,800
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 150	>99%	1 in 14,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 110	>99%	1 in 11,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 350	>99%	1 in 35,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	94%	1 in 3,100
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 27	>99%	1 in 2,600
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 91	95%	1 in 1,700
PCCB-related Propionic Acidemia (PCCB)	1 in 100	>99%	1 in 10,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 37	>99%	1 in 3,600
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000

* For X-linked diseases, female carrier frequencies are presented.

Middle East

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	1 in 44	>99%	1 in 4,300
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 200	>99%	1 in 20,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 170	96%	1 in 4,500
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 130	>99%	1 in 13,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 50	92%	1 in 560
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 110	98%	1 in 4,400
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000

* For X-linked diseases, female carrier frequencies are presented.

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	1 in 280	>99%	1 in 28,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Native American Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 80	>99%	1 in 7,900
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 390	>99%	1 in 39,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 24	>99%	1 in 2,300
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600

* For X-linked diseases, female carrier frequencies are presented.

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 62	90%	1 in 610
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 140	>99%	1 in 14,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 53	>99%	1 in 5,200
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

Native American

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 29	>99%	1 in 2,800
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 220	>99%	1 in 21,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 340	87%	1 in 2,600
Gaucher Disease (GBA1)	1 in 120	60%	1 in 310
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	50%	1 in 20
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000

* For X-linked diseases, female carrier frequencies are presented.

Native American

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 150	98%	1 in 10,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 320	>99%	1 in 32,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 400	97%	1 in 13,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 41	>99%	1 in 4,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 110	>99%	1 in 11,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 35	>99%	1 in 3,400
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	85%	1 in 500
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 56	>99%	1 in 5,500
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000

* For X-linked diseases, female carrier frequencies are presented.

Native American

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 200	>99%	1 in 20,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 95	>99%	1 in 9,400
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 50	93%	1 in 690
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000

* For X-linked diseases, female carrier frequencies are presented.

Native American

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Northwestern Europe Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 92	>99%	1 in 9,100
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 28	>99%	1 in 2,700
Alpha-mannosidosis (MAN2B1)	1 in 330	98%	1 in 22,000
Alpha-sarcoglycanopathy (SGCA)	< 1 in 500	>99%	< 1 in 50,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 210	94%	1 in 3,400
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 160	>99%	1 in 15,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 64,000	90%	1 in 660,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 150	>99%	1 in 15,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 360	>99%	1 in 36,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 330	>99%	1 in 32,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 38	>99%	1 in 3,700
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600

* For X-linked diseases, female carrier frequencies are presented.

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 130	>99%	1 in 13,000
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 160	99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 250	>99%	1 in 25,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	60%	1 in 170
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 140	>99%	1 in 14,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 87	>99%	1 in 8,600
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 55	96%	1 in 1,300
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 71	>99%	1 in 7,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 31	>99%	1 in 3,000
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

Northwestern Europe

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,500
ERCC8-related Disorders (ERCC8)	< 1 in 500	97%	< 1 in 16,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 55,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 110	>99%	1 in 11,000
Fanconi Anemia Complementation Group A (FANCA)	1 in 240	92%	1 in 2,800
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 160	>99%	1 in 16,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 370	>99%	1 in 37,000
Galactosemia (GALT)	1 in 87	>99%	1 in 8,600
Gamma-sarcoglycanopathy (SGCG)	1 in 440	87%	1 in 3,300
Gaucher Disease (GBA1)	1 in 110	60%	1 in 260
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 14	50%	1 in 26
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	1 in 230	>99%	1 in 23,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 200	>99%	1 in 20,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	96%	1 in 350,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	85%	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 230	>99%	1 in 23,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 95	>99%	1 in 9,400
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 300	>99%	1 in 30,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 330	>99%	1 in 32,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 32,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 140	>99%	1 in 14,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 200	98%	1 in 14,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 390	>99%	1 in 39,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 390	>99%	1 in 39,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	97%	1 in 16,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 45	>99%	1 in 4,400
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	1 in 480	>99%	1 in 48,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 270	>99%	1 in 26,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000

* For X-linked diseases, female carrier frequencies are presented.

Northwestern Europe

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 190	>99%	1 in 19,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 270	>99%	1 in 27,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	1 in 200	96%	1 in 4,400
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 190	>99%	1 in 19,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	1 in 160	>99%	1 in 16,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 36	>99%	1 in 3,500
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 83	>99%	1 in 8,200
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 49	>99%	1 in 4,800
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	98%	1 in 4,000

* For X-linked diseases, female carrier frequencies are presented.

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 110	>99%	1 in 11,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 170	>99%	1 in 17,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 130	>99%	1 in 13,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 26	>99%	1 in 2,500
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 320	98%	1 in 17,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	98%	1 in 11,000
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 300	>99%	1 in 30,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 110	>99%	1 in 11,000
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 95	>99%	1 in 9,400
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 35	95%	1 in 630
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 88	98%	1 in 4,100
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 180	>99%	1 in 18,000
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000

* For X-linked diseases, female carrier frequencies are presented.

Northwestern Europe

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Wilson Disease (ATP7B)	1 in 66	>99%	1 in 6,500
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 16,000	77%	1 in 71,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

South Asia Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 24	>99%	1 in 2,300
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	< 1 in 500	>99%	< 1 in 50,000
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 25	>99%	1 in 2,400
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 62	88%	1 in 530
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 450	>99%	1 in 45,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 100	>99%	1 in 10,000
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	< 1 in 500	>99%	< 1 in 50,000
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 320	>99%	1 in 32,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	< 1 in 500	87%	< 1 in 3,800
Gaucher Disease (GBA1)	1 in 120	60%	1 in 310
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	50%	1 in 21
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	< 1 in 500	98%	< 1 in 34,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 95	>99%	1 in 9,400
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 130	97%	1 in 4,100
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 100	>99%	1 in 10,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 26	>99%	1 in 2,500
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	92%	1 in 1,000
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	93%	1 in 270
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 87	>99%	1 in 8,600
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000

* For X-linked diseases, female carrier frequencies are presented.

South Asia

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 200	>99%	1 in 20,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 50	93%	1 in 630
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000

* For X-linked diseases, female carrier frequencies are presented.

South Asia

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	1 in 280	>99%	1 in 28,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Southeast Asia Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	1 in 330	>99%	1 in 33,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 250	>99%	1 in 25,000
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 390	>99%	1 in 39,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 18	>99%	1 in 1,700
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 18,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 50	88%	1 in 410
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 450	>99%	1 in 45,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 91	>99%	1 in 9,000
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

Southeast Asia

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	< 1 in 500	>99%	< 1 in 50,000
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 320	>99%	1 in 32,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 340	87%	1 in 2,600
Gaucher Disease (GBA1)	1 in 120	60%	1 in 310
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	80%	1 in 51
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 250	94%	1 in 4,100
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	< 1 in 500	98%	< 1 in 34,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 190	>99%	1 in 19,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 180	97%	1 in 5,700
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 64	>99%	1 in 6,300
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 56	>99%	1 in 5,500
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	< 1 in 500	>99%	< 1 in 50,000
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 200	>99%	1 in 20,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 53	93%	1 in 630
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000

* For X-linked diseases, female carrier frequencies are presented.

Southeast Asia

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 66	>99%	1 in 6,500
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.

Southeast Asia

myriad WOMEN'S HEALTH

Southern Europe Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 28	>99%	1 in 2,700
Alpha-mannosidosis (MAN2B1)	1 in 460	98%	1 in 31,000
Alpha-sarcoglycanopathy (SGCA)	1 in 290	>99%	1 in 29,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 210	94%	1 in 3,400
Alport Syndrome, COL4A4-related (COL4A4)	1 in 210	>99%	1 in 21,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 440	>99%	1 in 43,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 8	>99%	1 in 710
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 120	99%	1 in 9,400
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 200	>99%	1 in 20,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	60%	1 in 170
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 280	>99%	1 in 28,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 68	96%	1 in 1,600
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 71	>99%	1 in 7,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 28	>99%	1 in 2,700
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

Southern Europe

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,500
ERCC8-related Disorders (ERCC8)	< 1 in 500	97%	< 1 in 16,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 24	>99%	1 in 2,300
Fanconi Anemia Complementation Group A (FANCA)	1 in 240	92%	1 in 2,800
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 280	>99%	1 in 28,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 310	>99%	1 in 30,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 260	87%	1 in 2,000
Gaucher Disease (GBA1)	1 in 110	60%	1 in 260
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 14	50%	1 in 26
Glutaric Acidemia, GCDH-related (GCDH)	1 in 140	>99%	1 in 14,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 130	94%	1 in 2,100
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	85%	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	1 in 180	>99%	1 in 18,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 180	>99%	1 in 17,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 270	>99%	1 in 27,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 220	>99%	1 in 22,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 310	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 14,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 150	98%	1 in 10,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 130	>99%	1 in 13,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 210	>99%	1 in 21,000
Maple Syrup Urine Disease Type II (DBT)	1 in 220	97%	1 in 7,300
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 62	>99%	1 in 6,100
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 97	>99%	1 in 9,600
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000
Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 210	>99%	1 in 21,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 470	>99%	1 in 47,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 96	98%	1 in 4,500
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	1 in 390	96%	1 in 8,600
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 110	>99%	1 in 11,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	1 in 160	>99%	1 in 16,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 22	>99%	1 in 2,100
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	98%	1 in 940
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 42	>99%	1 in 4,100
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000
POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 380	98%	1 in 15,000
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590

* For X-linked diseases, female carrier frequencies are presented.

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 170	>99%	1 in 17,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 130	>99%	1 in 13,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 35	>99%	1 in 3,400
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 220	>99%	1 in 22,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	>99%	1 in 20,000
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 95	>99%	1 in 9,400
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 57	94%	1 in 890
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	< 1 in 500	>99%	< 1 in 50,000
USH2A-related Disorders (USH2A)	1 in 140	98%	1 in 6,500
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 200	>99%	1 in 20,000
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000

* For X-linked diseases, female carrier frequencies are presented.

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 14,000	77%	1 in 60,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

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myriad WOMEN'S HEALTH

Worldwide Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 9/25/2025

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
3-methylcrotonyl-CoA Carboxylase Deficiency, MCCC2-related (MCCC2)	1 in 190	>99%	1 in 19,000
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
Abetalipoproteinemia (MTTP)	< 1 in 500	99%	< 1 in 40,000
Achromatopsia, CNGB3-related (CNGB3)	1 in 98	>99%	1 in 9,700
Acute Liver Failure, TRMU-related (TRMU)	< 1 in 500	>99%	< 1 in 50,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	98%	1 in 22,000
Aicardi-Goutières Syndrome, RNASEH2B-related (RNASEH2B)	1 in 220	>99%	1 in 22,000
Aldosterone Synthase Deficiency (CYP11B2)	< 1 in 500	32%	< 1 in 740
Alkaptonuria (HGD)	1 in 400	>99%	1 in 39,000
Alpha-1 Antitrypsin Deficiency (SERPINA1)	1 in 80	>99%	1 in 7,900
Alpha-mannosidosis (MAN2B1)	1 in 220	98%	1 in 15,000
Alpha-sarcoglycanopathy (SGCA)	1 in 340	>99%	1 in 34,000
Alpha Thalassemia, HBA1/HBA2-related (HBA2, HBA1)	Not calculated	>99%	Not calculated
Alport Syndrome, COL4A3-related (COL4A3)	1 in 350	94%	1 in 5,800
Alport Syndrome, COL4A4-related (COL4A4)	1 in 350	>99%	1 in 35,000
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	1 in 330	97%	1 in 12,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
Arthrogryposis, Impaired Intellectual Development, and Seizures (SLC35A3)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
*ARX-related Disorders (ARX)	1 in 8,700	31%	1 in 12,000
Asparagine Synthetase Deficiency (ASNS)	< 1 in 500	>99%	< 1 in 50,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 160	96%	1 in 4,200
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 38,000	90%	1 in 400,000
Atransferrinemia (TF)	1 in 120	>99%	1 in 12,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	96%	1 in 8,900
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	< 1 in 500	99%	< 1 in 44,000
Bardet-Biedl Syndrome, BBS10-related (BBS10)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 390	>99%	1 in 39,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
BCS1L-related Disorders (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
Beta Globin-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 24	>99%	1 in 2,300
Beta-ketothiolase Deficiency (ACAT1)	1 in 200	98%	1 in 9,600
Beta-sarcoglycanopathy (SGCB)	1 in 400	>99%	1 in 39,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000

* For X-linked diseases, female carrier frequencies are presented.

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Biotin-thiamine–responsive Basal Ganglia Disease (SLC19A3)	1 in 110	98%	1 in 4,500
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 140	99%	1 in 11,000
Canavan Disease (ASPA)	1 in 160	89%	1 in 1,400
Carbamoylphosphate Synthetase I Deficiency (CPS1)	< 1 in 570	>99%	< 1 in 57,000
Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	< 1 in 500	>99%	< 1 in 50,000
Carnitine Palmitoyltransferase II Deficiency (CPT2)	1 in 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
CC2D2A-related Disorders (CC2D2A)	1 in 200	>99%	1 in 20,000
CEP290-related Disorders (CEP290)	1 in 69	>99%	1 in 6,800
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Chronic Granulomatous Disease, CYBA-related (CYBA)	< 1 in 500	>99%	< 1 in 50,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
Classical-like Ehlers-Danlos Syndrome, TNXB-related (TNXB)	1 in 28	36%	1 in 44
CLN3-related Disorders (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN8-related Neuronal Ceroid Lipofuscinosis (CLN8)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Adrenal Hyperplasia, CYP11A1-related (CYP11A1)	1 in 110	>99%	1 in 11,000
Congenital Adrenal Hyperplasia, CYP11B1-related (CYP11B1)	1 in 220	97%	1 in 8,400
Congenital Adrenal Hyperplasia, CYP21A2-related (CYP21A2)	1 in 62	97%	1 in 1,900
Congenital Amegakaryocytic Thrombocytopenia (MPL)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, ALG6-related (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, MPI-related (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation, PMM2-related (PMM2)	1 in 140	>99%	1 in 14,000
Congenital Hydrocephalus, CCDC88C-related (CCDC88C)	1 in 140	98%	1 in 6,700
Congenital Insensitivity to Pain with Anhidrosis, NTRK1-related (NTRK1)	< 1 in 500	>99%	< 1 in 50,000
Congenital Myasthenic Syndrome, CHRNE-related (CHRNE)	1 in 74	98%	1 in 3,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
*Creatine Transporter Deficiency (SLC6A8)	Not calculated	95%	Not calculated
Cystic Fibrosis (CFTR)	1 in 29	>99%	1 in 2,800
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	96%	< 1 in 13,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dihydropyrimidine Dehydrogenase Deficiency (DPYD)	< 1 in 500	98%	< 1 in 29,000
Distal Renal Tubular Acidosis with Deafness, ATP6V1B1-related (ATP6V1B1)	< 1 in 500	>99%	< 1 in 50,000
Donnai–Barrow Syndrome (LRP2)	1 in 210	>99%	1 in 21,000
DYNC2H1-related Disorders (DYNC2H1)	1 in 67	>99%	1 in 6,700
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
Dystrophic Epidermolysis Bullosa (COL7A1)	Not calculated	>99%	Not calculated
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	99%	Not calculated

* For X-linked diseases, female carrier frequencies are presented.

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Ehlers-Danlos Syndrome, ADAMTS2-related (ADAMTS2)	< 1 in 500	92%	< 1 in 5,900
Enhanced S-cone Syndrome (NR2E3)	< 1 in 500	>99%	< 1 in 50,000
ERCC2-related Disorders (ERCC2)	1 in 66	98%	1 in 3,900
ERCC6-related Disorders (ERCC6)	1 in 380	96%	1 in 8,400
ERCC8-related Disorders (ERCC8)	1 in 380	97%	1 in 12,000
EVC2-related Ellis-van Creveld Syndrome (EVC2)	1 in 250	98%	1 in 9,800
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 280	96%	1 in 7,800
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Factor V Leiden Thrombophilia (F5)	Not calculated	>99%	Not calculated
Factor XI Deficiency (F11)	< 1 in 500	>99%	< 1 in 50,000
Familial Dysautonomia (ELP1)	< 1 in 500	>99%	< 1 in 50,000
Familial Hemophagocytic Lymphohistiocytosis, PRF1-related (PRF1)	1 in 150	>99%	1 in 15,000
Familial Hyperinsulinism, ABCC8-related (ABCC8)	1 in 170	>99%	1 in 17,000
Familial Hyperinsulinism, KCNJ11-related (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 29	>99%	1 in 2,800
Fanconi Anemia Complementation Group A (FANCA)	1 in 260	92%	1 in 3,100
Fanconi Anemia, FANCC-related (FANCC)	< 1 in 500	>99%	< 1 in 50,000
FKRP-related Disorders (FKRP)	1 in 220	>99%	1 in 21,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile XE Syndrome (AFF2)	1 in 25,000	31%	1 in 36,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Fraser Syndrome, GRIP1-related (GRIP1)	1 in 83	98%	1 in 4,000
Free Sialic Acid Storage Disorders (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Friedreich Ataxia (FXN)	Not calculated	96%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 440	>99%	1 in 44,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 340	87%	1 in 2,600
Gaucher Disease (GBA1)	1 in 120	60%	1 in 310
GLB1-related Disorders (GLB1)	1 in 170	>99%	1 in 17,000
*Glucose-6-phosphate Dehydrogenase Deficiency (G6PD)	1 in 11	50%	1 in 20
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycine Encephalopathy, AMT-related (AMT)	1 in 260	>99%	1 in 26,000
Glycine Encephalopathy, GLDC-related (GLDC)	1 in 160	94%	1 in 2,500
Glycogen Storage Disease, GBE1-related (GBE1)	1 in 420	97%	1 in 12,000
Glycogen Storage Disease, PFKM-related (PFKM)	< 1 in 500	>99%	< 1 in 50,000
Glycogen Storage Disease, PYGM-related (PYGM)	1 in 110	>99%	1 in 11,000
Glycogen Storage Disease Type Ia (G6PC1)	1 in 180	98%	1 in 8,700
Glycogen Storage Disease Type Ib (SLC37A4)	1 in 350	>99%	1 in 35,000
Glycogen Storage Disease Type III (AGL)	1 in 160	>99%	1 in 16,000
GNE Myopathy (GNE)	< 1 in 500	>99%	< 1 in 50,000
GNPTAB-related Disorders (GNPTAB)	1 in 200	>99%	1 in 20,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
*Hemophilia A (F8)	Not calculated	49%	Not calculated
*Hemophilia B (F9)	1 in 13,000	97%	1 in 490,000
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900

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Hereditary Hemochromatosis, HFE-related (HFE)	Not calculated	Not calculated due to rarity of disease in this individual's reported ethnicity	Not calculated
Hermansky-Pudlak Syndrome, HPS1-related (HPS1)	1 in 59	94%	1 in 1,100
Hermansky-Pudlak Syndrome, HPS3-related (HPS3)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 280	>99%	1 in 28,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	>99%	< 1 in 50,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria, CBS-related (CBS)	1 in 270	>99%	1 in 27,000
Homocystinuria, MTHFR-related (MTHFR)	< 1 in 500	>99%	< 1 in 50,000
HSD17B4-related Disorders (HSD17B4)	1 in 160	98%	1 in 9,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 230	>99%	1 in 23,000
Inherited Retinal Dystrophy, RPE65-related (RPE65)	1 in 450	>99%	1 in 45,000
Isovaleric Acidemia (IVD)	1 in 260	>99%	1 in 26,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
Joubert Syndrome, AHI1-related (AHI1)	1 in 100	98%	1 in 4,500
Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	1 in 320	>99%	1 in 31,000
Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 180	>99%	1 in 17,000
*L1 Syndrome (L1CAM)	1 in 15,000	98%	1 in 640,000
Leigh Syndrome, French-Canadian Type (LRPPRC)	< 1 in 500	>99%	< 1 in 50,000
Lipoid Congenital Adrenal Hyperplasia (STAR)	< 1 in 500	>99%	< 1 in 50,000
Lysosomal Acid Lipase Deficiency (LIPA)	1 in 150	98%	1 in 10,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 320	>99%	1 in 32,000
Maple Syrup Urine Disease Type Ib (BCKDHB)	1 in 360	>99%	1 in 36,000
Maple Syrup Urine Disease Type II (DBT)	1 in 400	97%	1 in 13,000
Medium-chain Acyl-CoA Dehydrogenase Deficiency (ACADM)	1 in 61	>99%	1 in 6,000
Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)	< 1 in 500	>99%	< 1 in 50,000
Metachromatic Leukodystrophy (ARSA)	1 in 160	>99%	1 in 16,000
Methylmalonic Acidemia, cblA Type (MMAA)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, cblB Type (MMAB)	< 1 in 500	>99%	< 1 in 50,000
Methylmalonic Acidemia, MMUT-related (MMUT)	1 in 110	>99%	1 in 11,000
Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
Mevalonate Kinase Deficiency (MVK)	1 in 170	99%	1 in 15,000
Microcephaly with Seizures and Brain Atrophy, MED17-related (MED17)	< 1 in 500	89%	< 1 in 4,500
Microphthalmia, Anophthalmia, and Coloboma, VSX2-related (VSX2)	< 1 in 500	>99%	< 1 in 50,000
Mild MTHFR Deficiency (MTHFR)	Not calculated	>99%	Not calculated
Mitochondrial Complex I Deficiency, NDUFAF5-related (NDUFAF5)	< 1 in 500	29%	< 1 in 700
Mitochondrial Complex I Deficiency, NDUFS4-related (NDUFS4)	< 1 in 500	97%	< 1 in 17,000

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Mitochondrial Complex I Deficiency, NDUFS6-related (NDUFS6)	< 1 in 500	94%	< 1 in 8,000
Mitochondrial Complex IV Deficiency, SCO2-related (SCO2)	1 in 150	>99%	1 in 15,000
Mitochondrial Neurogastrointestinal Encephalopathy Disease (TYMP)	< 1 in 500	>99%	< 1 in 50,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	98%	< 1 in 20,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	97%	1 in 5,600
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	89%	1 in 670,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 260	>99%	1 in 26,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Multiple Sulfatase Deficiency (SUMF1)	< 1 in 500	>99%	< 1 in 50,000
Muscular Dystrophy, LAMA2-related (LAMA2)	1 in 120	98%	1 in 5,700
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
Myopathy, Lactic Acidosis, and Sideroblastic Anemia 1 (PUS1)	< 1 in 500	>99%	< 1 in 50,000
Myotonia Congenita (CLCN1)	1 in 180	98%	1 in 11,000
NAGA-related Disorders (NAGA)	1 in 94	>99%	1 in 9,300
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS1-related (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Neuronal Ceroid Lipofuscinosis, CLN6-related (CLN6)	< 1 in 500	96%	< 1 in 11,000
Neuronal Ceroid Lipofuscinosis, PPT1-related (PPT1)	1 in 78	>99%	1 in 7,700
Niemann-Pick Disease, SMPD1-related (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C1 (NPC1)	1 in 170	>99%	1 in 17,000
Niemann-Pick Disease Type C2 (NPC2)	< 1 in 500	>99%	< 1 in 50,000
Nijmegen Breakage Syndrome (NBN)	< 1 in 500	>99%	< 1 in 50,000
Nonsyndromic Hearing Loss, GJB2-related (GJB2)	1 in 35	>99%	1 in 3,400
Nonsyndromic Hearing Loss, LOXHD1-related (LOXHD1)	< 1 in 500	>99%	< 1 in 50,000
Normophosphatemic Familial Tumoral Calcinosis (SAMD9)	< 1 in 500	>99%	< 1 in 50,000
Oculocutaneous Albinism, OCA2-related (OCA2)	1 in 76	96%	1 in 1,700
Oculocutaneous Albinism, TYR-related (TYR)	1 in 20	99%	1 in 1,600
*Opitz G/BBB Syndrome, MID1-related (MID1)	1 in 25,000	87%	1 in 190,000
Ornithine Aminotransferase Deficiency (OAT)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 16,000	97%	1 in 530,000
PCCA-related Propionic Acidemia (PCCA)	1 in 220	95%	1 in 4,200
PCCB-related Propionic Acidemia (PCCB)	1 in 220	>99%	1 in 22,000
PCDH15-related Disorders (PCDH15)	1 in 220	93%	1 in 3,300
Pendred Syndrome (SLC26A4)	1 in 65	>99%	1 in 6,400
Peroxisome Biogenesis Disorder Type 1 (PEX1)	1 in 160	>99%	1 in 16,000
Peroxisome Biogenesis Disorder Type 3 (PEX12)	1 in 440	>99%	1 in 44,000
Peroxisome Biogenesis Disorder Type 4 (PEX6)	1 in 310	97%	1 in 9,300
Peroxisome Biogenesis Disorder Type 5 (PEX2)	< 1 in 710	>99%	< 1 in 71,000
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX7-related Disorders (PEX7)	1 in 160	>99%	1 in 16,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 56	>99%	1 in 5,500
*PLP1-related Disorders (PLP1)	1 in 100,000	32%	1 in 150,000
POLG-related Disorders (POLG)	1 in 190	>99%	1 in 19,000

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POMGNT1-related Disorders (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
Pompe Disease (GAA)	1 in 100	>99%	1 in 10,000
Pontocerebellar Hypoplasia, RARS2-related (RARS2)	< 1 in 500	16%	< 1 in 590
Pontocerebellar Hypoplasia, SEPSECS-related (SEPSECS)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VPS53-related (VPS53)	< 1 in 500	>99%	< 1 in 50,000
Pontocerebellar Hypoplasia, VRK1-related (VRK1)	< 1 in 500	>99%	< 1 in 50,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Ciliary Dyskinesia, DNAH5-related (DNAH5)	1 in 140	99%	1 in 13,000
Primary Ciliary Dyskinesia, DNAI1-related (DNAI1)	1 in 230	>99%	1 in 23,000
Primary Ciliary Dyskinesia, DNAI2-related (DNAI2)	1 in 450	>99%	1 in 45,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 140	>99%	1 in 13,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 200	>99%	1 in 20,000
Primary Microcephaly, MCPH1-related (MCPH1)	1 in 150	88%	1 in 1,300
Primary Trimethylaminuria (FMO3)	1 in 140	>99%	1 in 14,000
Prothrombin Thrombophilia (F2)	Not calculated	>99%	Not calculated
Pseudocholinesterase Deficiency (BCHE)	1 in 32	>99%	1 in 3,100
Pycnodysostosis (CTSK)	1 in 430	>99%	1 in 43,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
RAPSN-related Disorders (RAPSN)	1 in 480	98%	1 in 26,000
Refsum Disease, PHYH-related (PHYH)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, CERKL-related (CERKL)	1 in 440	>99%	1 in 44,000
Retinitis Pigmentosa, DHDDS-related (DHDDS)	< 1 in 500	>99%	< 1 in 50,000
Retinitis Pigmentosa, EYS-related (EYS)	1 in 200	96%	1 in 5,100
Retinitis Pigmentosa, FAM161A-related (FAM161A)	1 in 440	>99%	1 in 44,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	99%	< 1 in 37,000
Sandhoff Disease (HEXB)	1 in 320	98%	1 in 18,000
Serine Deficiency Disorder, PHGDH-related (PHGDH)	< 1 in 500	>99%	< 1 in 50,000
Severe Combined Immunodeficiency, RAG2-related (RAG2)	< 1 in 500	>99%	< 1 in 50,000
Short-chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	1 in 98	>99%	1 in 9,700
Sjogren-Larsson Syndrome (ALDH3A2)	< 1 in 500	96%	< 1 in 12,000
SLC26A2-related Disorders (SLC26A2)	1 in 160	>99%	1 in 16,000
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 95	>99%	1 in 9,400
Spastic Paraplegia, TECPR2-related (TECPR2)	< 1 in 500	>99%	< 1 in 50,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 54	91%	1 in 510
Spinocerebellar Ataxia, ANO10-related (ANO10)	1 in 94	97%	1 in 2,800
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	93%	< 1 in 6,800
Surfactant Deficiency, ABCA3-related (ABCA3)	1 in 120	>99%	1 in 12,000
TGM1-related Autosomal Recessive Congenital Ichthyosis (TGM1)	1 in 220	>99%	1 in 22,000
TPP1-related Neuronal Ceroid Lipofuscinosis (TPP1)	1 in 300	>99%	1 in 30,000
Tyrosine Hydroxylase Deficiency (TH)	< 1 in 500	>99%	< 1 in 50,000
Tyrosinemia Type I (FAH)	1 in 160	>99%	1 in 16,000
Tyrosinemia Type II (TAT)	1 in 250	>99%	1 in 25,000
USH1C-related Disorders (USH1C)	1 in 300	>99%	1 in 30,000
USH2A-related Disorders (USH2A)	1 in 150	98%	1 in 5,900
Usher Syndrome Type 3 (CLRN1)	1 in 410	>99%	1 in 41,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000

* For X-linked diseases, female carrier frequencies are presented.

Worldwide

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Vitamin D–dependent Rickets, CYP27B1-related (CYP27B1)	1 in 180	>99%	1 in 18,000
VPS13A Disease (VPS13A)	< 1 in 500	99%	< 1 in 40,000
Wilson Disease (ATP7B)	1 in 91	>99%	1 in 9,000
Xeroderma Pigmentosum Group A (XPA)	< 1 in 500	>99%	< 1 in 50,000
Xeroderma Pigmentosum Group C (XPC)	1 in 240	97%	1 in 7,300
*X-linked Adrenal Hypoplasia Congenita (NR0B1)	1 in 300,000	97%	< 1 in 1,000,000
*X-linked Adrenoleukodystrophy (ABCD1)	1 in 8,400	77%	1 in 36,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	96%	Not calculated
*X-linked Choroideremia (CHM)	1 in 25,000	97%	1 in 780,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	96%	Not calculated
*X-linked Retinal Dystrophy, RPGR-related (RPGR)	1 in 17,000	38%	1 in 28,000
*X-linked Severe Combined Immunodeficiency (IL2RG)	1 in 50,000	>99%	< 1 in 1,000,000

* For X-linked diseases, female carrier frequencies are presented.