myriad WOMEN'S HEALTH

African American Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 120	92%	1 in 1,400
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	90%	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 120	>99%	1 in 12,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 440	>99%	1 in 44,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 310	>99%	1 in 38,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000

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

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 66	>99%	1 in 6,500
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 71	>99%	1 in 7,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 120	60%	1 in 310
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 48	>99%	1 in 4,700
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 11	>99%	1 in 950
Hereditary Fructose Intolerance (ALDOB)	1 in 230	>99%	1 in 23,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000

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

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 260	>99%	1 in 26,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	96%	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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 180	>99%	1 in 18,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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 71	>99%	1 in 7,000
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

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

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 160	>99%	1 in 16,000
Pompe Disease (GAA)	1 in 60	>99%	1 in 5,900
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	< 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
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 58	>99%	1 in 5,700
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 45	>99%	1 in 4,400
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	Unknown due to rarity of disease	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 200	>99%	1 in 20,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 420	>99%	1 in 42,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	1 in 140	>99%	1 in 14,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 440	>99%	1 in 60,000
Bloom Syndrome (BLM)	1 in 110	>99%	1 in 11,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 55	98%	1 in 3,300
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
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 180	97%	1 in 5,500
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000

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

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Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 28	>99%	1 in 2,700
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	1 in 94	>99%	1 in 9,300
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	1 in 31	>99%	1 in 3,000
Familial Mediterranean Fever (MEFV)	1 in 11	>99%	1 in 1,000
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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	1 in 150	>99%	1 in 15,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 160	>99%	1 in 16,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 14	95%	1 in 270
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 21	>99%	1 in 2,000
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 71	>99%	1 in 7,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 67	>99%	1 in 6,600
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000

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

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 31	>99%	1 in 3,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	1 in 97	>99%	1 in 9,600
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	1 in 200	>99%	1 in 20,000
Krabbe Disease (GALC)	< 1 in 500	>99%	< 1 in 50,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 97	>99%	1 in 9,600
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 320	>99%	1 in 32,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	96%	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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	1 in 90	>99%	1 in 8,900
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 180	>99%	1 in 18,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 110	>99%	1 in 11,000
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 100	>99%	1 in 10,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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

* 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
Pendred Syndrome (SLC26A4)	1 in 71	>99%	1 in 7,000
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
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 220	>99%	1 in 22,000
Pompe Disease (GAA)	1 in 160	>99%	1 in 16,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
Primary Hyperoxaluria Type 2 (GRHPR)	< 1 in 500	>99%	< 1 in 50,000
Primary Hyperoxaluria Type 3 (HOGA1)	1 in 87	>99%	1 in 8,600
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	1 in 100	>99%	1 in 10,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	< 1 in 500	99%	< 1 in 47,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 100	>99%	1 in 10,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 41	94%	1 in 350
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	1 in 120	>99%	1 in 12,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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

Eastern Asia Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 72	88%	1 in 590
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	1 in 350	>99%	1 in 35,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 140	>99%	1 in 14,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	90%	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 120	>99%	1 in 12,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 180,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
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 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 460	>99%	1 in 67,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 97	86%	1 in 700
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 87	>99%	1 in 8,600
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 370	99%	1 in 26,000
ERCC8-related Disorders (ERCC8)	< 1 in 510	95%	< 1 in 9,800
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 500	>99%	< 1 in 50,000
FKTN-related Disorders (FKTN)	1 in 190	10%	1 in 210
*Fragile X Syndrome (FMR1)	Not calculated	>99%	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 380	88%	1 in 3,200
Gaucher Disease (GBA)	1 in 220	60%	1 in 560
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 100	>99%	1 in 10,000
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 140	>99%	1 in 13,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	98%	1 in 17,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 51	>99%	1 in 5,000
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 160	>99%	1 in 16,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 190	>99%	1 in 19,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	1 in 420	>99%	1 in 42,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	< 1 in 610	>99%	< 1 in 61,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 490	>99%	1 in 49,000
Maple Syrup Urine Disease Type II (DBT)	< 1 in 500	96%	< 1 in 13,000
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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 330	>99%	1 in 33,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 24,000	88%	1 in 200,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 300	>99%	1 in 30,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 110	>99%	1 in 11,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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 71	>99%	1 in 7,000
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

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

Eastern Asia

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 350	>99%	1 in 35,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 51	>99%	1 in 5,000
Pompe Disease (GAA)	1 in 110	>99%	1 in 11,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 100	>99%	1 in 10,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	< 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
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 130	>99%	1 in 12,000
Wilson Disease (ATP7B)	1 in 51	>99%	1 in 5,000
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 Adrenoleukodystrophy (ABCD1)	1 in 20,000	77%	1 in 86,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	99%	Not calculated
*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

Finland Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 58	89%	1 in 530
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 100	>99%	1 in 10,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	Unknown due to rarity of disease	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 120	>99%	1 in 12,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 190	>99%	1 in 19,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	1 in 71	>99%	1 in 7,000
Ataxia-telangiectasia (ATM)	1 in 200	>99%	1 in 20,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 80	>99%	1 in 7,900
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 52	>99%	1 in 5,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 420	>99%	1 in 42,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 71	>99%	1 in 7,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	1 in 24	>99%	1 in 2,300
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	1 in 160	97%	1 in 4,800
COL4A3-related Alport Syndrome (COL4A3)	1 in 370	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 370	98%	1 in 22,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	1 in 45	>99%	1 in 4,400
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 80	>99%	1 in 7,900
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 110	60%	1 in 280
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 42	>99%	1 in 4,100
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 120	94%	1 in 2,100
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	1 in 110	>99%	1 in 11,000
HADHA-related Disorders (HADHA)	1 in 130	>99%	1 in 12,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 80	>99%	1 in 7,900
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	1 in 71	>99%	1 in 7,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	1 in 450	>99%	1 in 45,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 320	>99%	1 in 32,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	96%	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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	1 in 48	>99%	1 in 4,700
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	98%	< 1 in 25,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 180	>99%	1 in 18,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	1 in 140	>99%	1 in 13,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 31,000	97%	< 1 in 1,000,000
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

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pendred Syndrome (SLC26A4)	1 in 71	>99%	1 in 7,000
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
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 200	>99%	1 in 20,000
Pompe Disease (GAA)	1 in 160	>99%	1 in 16,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 71	>99%	1 in 7,000
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	1 in 100	>99%	1 in 10,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 100	>99%	1 in 10,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 35	94%	1 in 560
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 75	>99%	1 in 7,400
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 8,500	98%	1 in 570,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 58	96%	1 in 1,400
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	Unknown due to rarity of disease	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	1 in 23	>99%	1 in 2,200
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	1 in 22	99%	1 in 1,900
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 200	>99%	1 in 20,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 150	>99%	1 in 15,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	1 in 430	>99%	1 in 43,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 16	>99%	1 in 1,500
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 110	60%	1 in 280
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 42	>99%	1 in 4,100
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 40	>99%	1 in 3,900
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 67	>99%	1 in 6,600
Hereditary Fructose Intolerance (ALDOB)	1 in 81	>99%	1 in 8,000
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000

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

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 51	>99%	1 in 5,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 290	>99%	1 in 29,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	95%	1 in 9,600
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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 230	>99%	1 in 23,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 380	>99%	1 in 38,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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

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

myriad®
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pendred Syndrome (SLC26A4)	1 in 71	>99%	1 in 7,000
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
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 51	>99%	1 in 5,000
Pompe Disease (GAA)	1 in 160	>99%	1 in 16,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 120	99%	1 in 11,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 100	>99%	1 in 10,000
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 35	94%	1 in 570
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 230	>99%	1 in 23,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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.

French Canadian or Cajun

myriad WOMEN'S HEALTH

Hispanic Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 56	95%	1 in 1,100
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	Unknown due to rarity of disease	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 290	>99%	1 in 29,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 120	97%	1 in 3,700
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 420	>99%	1 in 42,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 46	>99%	1 in 4,500
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 120	60%	1 in 310
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 100	>99%	1 in 10,000
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 25	>99%	1 in 2,400
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000

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

myriad
WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 180	>99%	1 in 18,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 150	>99%	1 in 14,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	96%	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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 170	>99%	1 in 17,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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

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

Hispanic

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pendred Syndrome (SLC26A4)	1 in 71	>99%	1 in 7,000
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
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 51	>99%	1 in 5,000
Pompe Disease (GAA)	1 in 160	98%	1 in 10,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 140	>99%	1 in 13,000
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
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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

Middle East Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 42	97%	1 in 1,200
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	1 in 45	>99%	1 in 4,400
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 140	>99%	1 in 14,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	90%	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 120	>99%	1 in 12,000
Ataxia with Vitamin E Deficiency (TTPA)	1 in 160	>99%	1 in 16,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 97	>99%	1 in 9,600
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 110	>99%	1 in 11,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	1 in 210	>99%	1 in 20,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	1 in 180	>99%	1 in 18,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 110	>99%	1 in 11,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	1 in 51	>99%	1 in 5,000
Cystic Fibrosis (CFTR)	1 in 87	>99%	1 in 8,600
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 16	>99%	1 in 1,500
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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 120	60%	1 in 310
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 100	>99%	1 in 10,000
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 66	>99%	1 in 6,500
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 140	>99%	1 in 14,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 23	>99%	1 in 2,200
Hereditary Fructose Intolerance (ALDOB)	1 in 98	>99%	1 in 9,700
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	1 in 160	98%	1 in 10,000
Holocarboxylase Synthetase Deficiency (HLCS)	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
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	1 in 130	>99%	1 in 12,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	1 in 410	>99%	1 in 41,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 110	>99%	1 in 11,000
Maple Syrup Urine Disease Type II (DBT)	1 in 120	96%	1 in 3,300
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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 80	>99%	1 in 7,900
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 150	>99%	1 in 14,000
Mucopolysaccharidosis Type IIIB (NAGLU)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	< 1 in 500	>99%	< 1 in 50,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 53	>99%	1 in 5,200
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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 71	>99%	1 in 7,000
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

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 26	>99%	1 in 2,500
Pompe Disease (GAA)	1 in 160	>99%	1 in 16,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	< 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
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 56	90%	1 in 550
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	Unknown due to rarity of disease	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 120	>99%	1 in 12,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 420	>99%	1 in 42,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 87	>99%	1 in 8,600
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 120	60%	1 in 310
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 100	>99%	1 in 10,000
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 67	>99%	1 in 6,600
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000

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

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WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 320	>99%	1 in 32,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	96%	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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 180	>99%	1 in 18,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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

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

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WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pendred Syndrome (SLC26A4)	1 in 71	>99%	1 in 7,000
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
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 350	>99%	1 in 35,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 220	>99%	1 in 22,000
Pompe Disease (GAA)	1 in 160	>99%	1 in 16,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	< 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
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 220	94%	1 in 3,800
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 58	96%	1 in 1,400
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 220	>99%	1 in 22,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	Unknown due to rarity of disease	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 200	98%	1 in 11,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	96%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 150	>99%	1 in 15,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 420	>99%	1 in 42,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 130	>99%	1 in 13,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 250	>99%	1 in 25,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 140	>99%	1 in 14,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 87	>99%	1 in 8,600
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	1 in 430	>99%	1 in 43,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 210	97%	1 in 6,200
COL4A4-related Alport Syndrome (COL4A4)	1 in 210	98%	1 in 12,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 28	>99%	1 in 2,700
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 380	99%	1 in 26,000
ERCC8-related Disorders (ERCC8)	< 1 in 520	95%	< 1 in 9,900
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 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 X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 100	>99%	1 in 10,000
Galactosemia (GALT)	1 in 87	>99%	1 in 8,600
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 110	60%	1 in 280
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 33	>99%	1 in 3,200
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 200	>99%	1 in 20,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 32	>99%	1 in 3,100
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000

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

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WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	1 in 150	>99%	1 in 15,000
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 270	>99%	1 in 27,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 340	>99%	1 in 34,000
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 180	>99%	1 in 18,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 420	>99%	1 in 42,000
Maple Syrup Urine Disease Type II (DBT)	1 in 480	96%	1 in 13,000
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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 38,000	88%	1 in 300,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 120	>99%	1 in 12,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 250	>99%	1 in 25,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 370	>99%	1 in 37,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 260	>99%	1 in 26,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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

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

Northwestern Europe

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Pendred Syndrome (SLC26A4)	1 in 71	>99%	1 in 7,000
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
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 51	>99%	1 in 5,000
Pompe Disease (GAA)	1 in 160	98%	1 in 6,300
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 110	>99%	1 in 11,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	>99%	1 in 32,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 50	>99%	1 in 4,900
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 35	95%	1 in 630
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 180	>99%	1 in 18,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 42	88%	1 in 360
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	90%	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 120	>99%	1 in 12,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
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 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 87	>99%	1 in 8,600
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 500	>99%	< 1 in 50,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 120	60%	1 in 310
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 100	>99%	1 in 10,000
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 23	>99%	1 in 2,200
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	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
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 95	>99%	1 in 9,400
Maple Syrup Urine Disease Type II (DBT)	1 in 480	96%	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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 72	>99%	1 in 7,100
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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 71	>99%	1 in 7,000
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

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 350	>99%	1 in 35,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 51	>99%	1 in 5,000
Pompe Disease (GAA)	1 in 160	>99%	1 in 16,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	< 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
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 190	94%	1 in 3,300
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 59	88%	1 in 480
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	90%	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 120	>99%	1 in 12,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	92%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
Bardet-Biedl Syndrome, BBS10-related (BBS10)	1 in 420	>99%	1 in 42,000
Bardet-Biedl Syndrome, BBS12-related (BBS12)	< 1 in 500	>99%	< 1 in 50,000
Bardet-Biedl Syndrome, BBS1-related (BBS1)	1 in 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 18,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 180	>99%	1 in 18,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 130	>99%	1 in 13,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	< 1 in 500	>99%	< 1 in 50,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 350	97%	1 in 11,000
COL4A4-related Alport Syndrome (COL4A4)	1 in 350	98%	1 in 21,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 87	>99%	1 in 8,600
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 280	99%	1 in 19,000
ERCC8-related Disorders (ERCC8)	1 in 380	95%	1 in 7,300
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 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 500	>99%	< 1 in 50,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 350	>99%	1 in 35,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 120	60%	1 in 310
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 100	>99%	1 in 10,000
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 160	>99%	1 in 16,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 23	>99%	1 in 2,200
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	< 1 in 500	98%	< 1 in 33,000
Holocarboxylase Synthetase Deficiency (HLCS)	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
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 220	>99%	1 in 22,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 170	>99%	1 in 17,000
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 300	>99%	1 in 30,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 190	>99%	1 in 19,000
Maple Syrup Urine Disease Type II (DBT)	1 in 280	96%	1 in 7,600
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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 310	>99%	1 in 31,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 54	>99%	1 in 5,300
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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 71	>99%	1 in 7,000
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

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 350	>99%	1 in 35,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 51	>99%	1 in 5,000
Pompe Disease (GAA)	1 in 110	>99%	1 in 11,000
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	99%	1 in 30,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	< 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
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 140	>99%	1 in 14,000
Wilson Disease (ATP7B)	1 in 51	>99%	1 in 5,000
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 Adrenoleukodystrophy (ABCD1)	1 in 11,000	77%	1 in 45,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 13,000	98%	1 in 840,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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

Southern Europe Fact Sheet

Myriad Foresight® Carrier Screen - Last Updated: 1/15/2019

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
11-beta-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP11B1)	1 in 220	94%	1 in 3,800
21-hydroxylase-deficient Congenital Adrenal Hyperplasia (CYP21A2)	1 in 58	96%	1 in 1,300
6-pyruvoyl-tetrahydropterin Synthase Deficiency (PTS)	< 1 in 500	>99%	< 1 in 50,000
ABCC8-related Familial Hyperinsulinism (ABCC8)	1 in 170	>99%	1 in 17,000
Adenosine Deaminase Deficiency (ADA)	1 in 390	>99%	1 in 39,000
Alpha-mannosidosis (MAN2B1)	1 in 350	>99%	1 in 35,000
Alpha-sarcoglycanopathy (SGCA)	1 in 450	>99%	1 in 45,000
Alpha Thalassemia (HBA2, HBA1)	Not calculated	90%	Not calculated
Alstrom Syndrome (ALMS1)	< 1 in 500	>99%	< 1 in 50,000
AMT-related Glycine Encephalopathy (AMT)	1 in 220	>99%	1 in 22,000
Andermann Syndrome (SLC12A6)	< 1 in 500	>99%	< 1 in 50,000
Argininemia (ARG1)	< 1 in 500	97%	< 1 in 17,000
Argininosuccinic Aciduria (ASL)	1 in 130	>99%	1 in 13,000
ARSACS (SACS)	< 1 in 500	99%	< 1 in 44,000
Aspartylglucosaminuria (AGA)	< 1 in 500	>99%	< 1 in 50,000
Ataxia-telangiectasia (ATM)	1 in 200	99%	1 in 18,000
Ataxia with Vitamin E Deficiency (TTPA)	< 1 in 500	>99%	< 1 in 50,000
*ATP7A-related Disorders (ATP7A)	1 in 150,000	96%	< 1 in 1,000,000
Autoimmune Polyglandular Syndrome Type 1 (AIRE)	1 in 180	>99%	1 in 18,000
Autosomal Recessive Osteopetrosis Type 1 (TCIRG1)	1 in 350	>99%	1 in 35,000
Autosomal Recessive Polycystic Kidney Disease, PKHD1-related (PKHD1)	1 in 82	>99%	1 in 8,100
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 160	>99%	1 in 16,000
Bardet-Biedl Syndrome, BBS2-related (BBS2)	< 1 in 500	>99%	< 1 in 50,000
Beta-sarcoglycanopathy (SGCB)	< 1 in 500	>99%	< 1 in 50,000
Biotinidase Deficiency (BTD)	1 in 160	>99%	1 in 17,000
Bloom Syndrome (BLM)	< 1 in 500	>99%	< 1 in 50,000
Calpainopathy (CAPN3)	1 in 130	>99%	1 in 13,000
Canavan Disease (ASPA)	1 in 160	98%	1 in 9,700
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 200	>99%	1 in 20,000
Cartilage-hair Hypoplasia (RMRP)	< 1 in 500	>99%	< 1 in 50,000
Cerebrotendinous Xanthomatosis (CYP27A1)	1 in 110	>99%	1 in 11,000
Citrullinemia Type 1 (ASS1)	1 in 120	>99%	1 in 12,000
CLN3-related Neuronal Ceroid Lipofuscinosis (CLN3)	1 in 280	>99%	1 in 28,000
CLN5-related Neuronal Ceroid Lipofuscinosis (CLN5)	< 1 in 500	>99%	< 1 in 50,000
CLN6-related Neuronal Ceroid Lipofuscinosis (CLN6)	1 in 430	>99%	1 in 43,000
Cohen Syndrome (VPS13B)	< 1 in 500	97%	< 1 in 15,000
COL4A3-related Alport Syndrome (COL4A3)	1 in 210	97%	1 in 6,200
COL4A4-related Alport Syndrome (COL4A4)	1 in 210	98%	1 in 13,000
Combined Pituitary Hormone Deficiency, PROP1-related (PROP1)	1 in 62	>99%	1 in 6,100
Congenital Disorder of Glycosylation Type Ia (PMM2)	1 in 160	>99%	1 in 16,000

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Congenital Disorder of Glycosylation Type Ib (MPI)	< 1 in 500	>99%	< 1 in 50,000
Congenital Disorder of Glycosylation Type Ic (ALG6)	< 1 in 500	>99%	< 1 in 50,000
Congenital Finnish Nephrosis (NPHS1)	< 1 in 500	>99%	< 1 in 50,000
Costeff Optic Atrophy Syndrome (OPA3)	< 1 in 500	>99%	< 1 in 50,000
Cystic Fibrosis (CFTR)	1 in 28	>99%	1 in 2,700
Cystinosis (CTNS)	1 in 220	>99%	1 in 22,000
D-bifunctional Protein Deficiency (HSD17B4)	1 in 160	98%	1 in 9,000
Delta-sarcoglycanopathy (SGCD)	< 1 in 500	99%	< 1 in 40,000
Dihydrolipoamide Dehydrogenase Deficiency (DLD)	< 1 in 500	>99%	< 1 in 50,000
Dysferlinopathy (DYSF)	1 in 190	98%	1 in 11,000
*Dystrophinopathy (Including Duchenne/Becker Muscular Dystrophy) (DMD)	Not calculated	>99%	Not calculated
ERCC6-related Disorders (ERCC6)	1 in 380	99%	1 in 26,000
ERCC8-related Disorders (ERCC8)	< 1 in 520	95%	< 1 in 9,900
EVC2-related Ellis-van Creveld Syndrome (EVC2)	< 1 in 500	>99%	< 1 in 50,000
EVC-related Ellis-van Creveld Syndrome (EVC)	1 in 330	96%	1 in 7,500
*Fabry Disease (GLA)	1 in 20,000	98%	< 1 in 1,000,000
Familial Dysautonomia (IKBKAP)	< 1 in 500	>99%	< 1 in 50,000
Familial Mediterranean Fever (MEFV)	1 in 110	>99%	1 in 10,000
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 190	>99%	1 in 19,000
FKTN-related Disorders (FKTN)	< 1 in 500	>99%	< 1 in 50,000
*Fragile X Syndrome (FMR1)	Not calculated	>99%	Not calculated
Galactokinase Deficiency (GALK1)	1 in 310	>99%	1 in 31,000
Galactosemia (GALT)	1 in 110	>99%	1 in 11,000
Gamma-sarcoglycanopathy (SGCG)	1 in 350	88%	1 in 3,000
Gaucher Disease (GBA)	1 in 110	60%	1 in 280
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness (GJB2)	1 in 42	>99%	1 in 4,100
GLB1-related Disorders (GLB1)	1 in 190	>99%	1 in 19,000
GLDC-related Glycine Encephalopathy (GLDC)	1 in 160	94%	1 in 2,800
Glutaric Acidemia, GCDH-related (GCDH)	1 in 140	>99%	1 in 14,000
Glycogen Storage Disease Type Ia (G6PC)	1 in 180	>99%	1 in 18,000
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
GNPTAB-related Disorders (GNPTAB)	1 in 320	>99%	1 in 32,000
GRACILE Syndrome (BCS1L)	< 1 in 500	>99%	< 1 in 50,000
HADHA-related Disorders (HADHA)	1 in 250	>99%	1 in 25,000
Hb Beta Chain-related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (HBB)	1 in 14	>99%	1 in 1,300
Hereditary Fructose Intolerance (ALDOB)	1 in 80	>99%	1 in 7,900
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related (LAMA3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related (LAMB3)	< 1 in 500	>99%	< 1 in 50,000
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)	< 1 in 500	>99%	< 1 in 50,000
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (HEXA)	1 in 300	>99%	1 in 30,000
HMG-CoA Lyase Deficiency (HMGCL)	1 in 160	98%	1 in 10,000
Holocarboxylase Synthetase Deficiency (HLCS)	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
Homocystinuria Caused by Cystathionine Beta-synthase Deficiency (CBS)	1 in 250	>99%	1 in 25,000
Hydrolethalus Syndrome (HYLS1)	< 1 in 500	>99%	< 1 in 50,000
Hypophosphatasia (ALPL)	1 in 270	>99%	1 in 27,000
Inclusion Body Myopathy 2 (GNE)	< 1 in 500	>99%	< 1 in 50,000
Isovaleric Acidemia (IVD)	1 in 250	>99%	1 in 25,000
Joubert Syndrome 2 (TMEM216)	< 1 in 500	>99%	< 1 in 50,000
KCNJ11-related Familial Hyperinsulinism (KCNJ11)	< 1 in 500	>99%	< 1 in 50,000
Krabbe Disease (GALC)	1 in 150	>99%	1 in 15,000
LAMA2-related Muscular Dystrophy (LAMA2)	1 in 340	>99%	1 in 34,000
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 180	>99%	1 in 18,000
Maple Syrup Urine Disease Type 1B (BCKDHB)	1 in 250	>99%	1 in 25,000
Maple Syrup Urine Disease Type Ia (BCKDHA)	1 in 160	>99%	1 in 16,000
Maple Syrup Urine Disease Type II (DBT)	1 in 410	96%	1 in 11,000
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 Aciduria and Homocystinuria, cblC Type (MMACHC)	1 in 160	>99%	1 in 16,000
MKS1-related Disorders (MKS1)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis III Gamma (GNPTG)	< 1 in 500	>99%	< 1 in 50,000
Mucolipidosis IV (MCOLN1)	< 1 in 500	>99%	< 1 in 50,000
Mucopolysaccharidosis Type I (IDUA)	1 in 160	>99%	1 in 16,000
*Mucopolysaccharidosis Type II (IDS)	1 in 75,000	88%	1 in 600,000
Mucopolysaccharidosis Type IIIA (SGSH)	1 in 160	>99%	1 in 16,000
Mucopolysaccharidosis Type IIIB (NAGLU)	1 in 180	>99%	1 in 18,000
Mucopolysaccharidosis Type IIIC (HGSNAT)	1 in 430	>99%	1 in 43,000
Muscle-eye-brain Disease (POMGNT1)	< 1 in 500	96%	< 1 in 12,000
MUT-related Methylmalonic Acidemia (MUT)	1 in 180	>99%	1 in 18,000
MYO7A-related Disorders (MYO7A)	1 in 150	>99%	1 in 15,000
NEB-related Nemaline Myopathy (NEB)	1 in 87	92%	1 in 1,200
Nephrotic Syndrome, NPHS2-related (NPHS2)	1 in 360	>99%	1 in 35,000
Niemann-Pick Disease, SMPD1-associated (SMPD1)	1 in 250	>99%	1 in 25,000
Niemann-Pick Disease Type C (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
Northern Epilepsy (CLN8)	< 1 in 500	>99%	< 1 in 50,000
*Ornithine Transcarbamylase Deficiency (OTC)	1 in 34,000	97%	< 1 in 1,000,000
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 71	>99%	1 in 7,000
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

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

myriad WOMEN'S HEALTH

Disease	Carrier Frequency	Detection Rate	Residual Carrier Risk
Peroxisome Biogenesis Disorder Type 6 (PEX10)	< 1 in 500	>99%	< 1 in 50,000
PEX1-related Zellweger Syndrome Spectrum (PEX1)	1 in 110	>99%	1 in 11,000
Phenylalanine Hydroxylase Deficiency (PAH)	1 in 51	>99%	1 in 5,000
Pompe Disease (GAA)	1 in 160	98%	1 in 6,300
PPT1-related Neuronal Ceroid Lipofuscinosis (PPT1)	1 in 78	>99%	1 in 7,700
Primary Carnitine Deficiency (SLC22A5)	1 in 160	>99%	1 in 16,000
Primary Hyperoxaluria Type 1 (AGXT)	1 in 350	>99%	1 in 35,000
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
Pycnodysostosis (CTSK)	< 1 in 500	>99%	< 1 in 50,000
Pyruvate Carboxylase Deficiency (PC)	1 in 250	>99%	1 in 25,000
Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7)	1 in 160	>99%	1 in 16,000
RTEL1-related Disorders (RTEL1)	< 1 in 500	>99%	< 1 in 50,000
Salla Disease (SLC17A5)	< 1 in 500	98%	< 1 in 30,000
Sandhoff Disease (HEXB)	1 in 320	>99%	1 in 32,000
Segawa Syndrome (TH)	< 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 250	97%	1 in 9,100
Smith-Lemli-Opitz Syndrome (DHCR7)	1 in 83	>99%	1 in 8,200
Spastic Paraplegia Type 15 (ZFYVE26)	< 1 in 500	>99%	< 1 in 50,000
Spinal Muscular Atrophy (SMN1)	1 in 57	94%	1 in 890
Spondylothoracic Dysostosis (MESP2)	< 1 in 500	>99%	< 1 in 50,000
Sulfate Transporter-related Osteochondrodysplasia (SLC26A2)	1 in 110	>99%	1 in 11,000
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
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 350	>99%	1 in 35,000
USH2A-related Disorders (USH2A)	1 in 130	94%	1 in 2,200
Usher Syndrome Type 3 (CLRN1)	< 1 in 500	>99%	< 1 in 50,000
Very-long-chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)	1 in 200	>99%	1 in 20,000
Wilson Disease (ATP7B)	1 in 87	>99%	1 in 8,600
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 Adrenoleukodystrophy (ABCD1)	1 in 14,000	77%	1 in 60,000
*X-linked Alport Syndrome (COL4A5)	Not calculated	95%	Not calculated
*X-linked Congenital Adrenal Hypoplasia (NR0B1)	1 in 300,000	99%	< 1 in 1,000,000
*X-linked Juvenile Retinoschisis (RS1)	1 in 10,000	98%	1 in 670,000
*X-linked Myotubular Myopathy (MTM1)	Not calculated	98%	Not calculated
*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.