myriad WOMEN'S HEALTH

Genomic Diagnostics
LEADING THE WAY TO IMPROVE HEALTH

Myriad Foresight™ Carrier Screen Disease List

The Myriad Foresight Carrier Screen focuses on serious, clinically-actionable, and prevalent conditions to ensure you are providing meaningful information to your patients.

- 11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CYP11B1)
- 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (CYP21A2)*
- 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (PTS)
- ABCC8-Related Hyperinsulinism (ABCC8
- Adenosine Deaminase Deficiency (ADA)
- Adrenoleukodystrophy: X-Linked (ABCD1) X-linked
- Alpha Thalassemia (HBA1/HBA2)* ACOG ACMG
- Alpha-Mannosidosis (MAN2B1)
- Alpha-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2D) (SGCA)
- Alport Syndrome, X-Linked (COL4A5) X-linked
- Alstrom Syndrome (ALMS1)
- AMT-Related Glycine Encephalopathy (AMT)
- Andermann Syndrome (SLC12A6)
- Argininemia (ARG1)
- Argininosuccinic Aciduria (ASL)
- ARSACS (SACS)
- Aspartylglycosaminuria (AGA)
- Ataxia with Vitamin E Deficiency (TTPA)
- Ataxia-Telangiectasia (ATM)
- ATP7A-Related Disorders (ATP7A) X-linked
- Autosomal Recessive Osteopetrosis, Type 1 (TCIRG1)
- Bardet-Biedl Syndrome, BBS1-Related (BBS1)
- Bardet-Biedl Syndrome, BBS10-Related (BBS10)
- Bardet-Biedl Syndrome, BBS12-Related (BBS12)
- Bardet-Biedl Syndrome, BBS2-Related (BBS2)
- Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) (SGCB)
- Biotinidase Deficiency (BTD)
- Bloom Syndrome (BLM) ACMG
- Calpainopathy (CAPN3)
- Canavan Disease (ASPA) ACOG ACMG
- Carbamoylphosphate Synthetase I Deficiency (CPS1)
- Carnitine Palmitoyltransferase IA Deficiency (CPT1A)
- Carnitine Palmitoyltransferase II Deficiency (CPT2)
- Cartilage-Hair Hypoplasia (RMRP)
- Cerebrotendinous Xanthomatosis (CYP27A1)
- Citrullinemia, Type 1 (ASS1)
- CLN3-Related Neuronal Ceroid Lipofuscinosis (CLN3)
- CLN5-Related Neuronal Ceroid Lipofuscinosis (CLN5)
- CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 (CLN6)
- Cohen Syndrome (VPS13B)
- COL4A3-Related Alport Syndrome (COL4A3)
- COL4A4-Related Alport Syndrome (COL4A4)
- Congenital Disorder of Glycosylation, Type Ia (PMM2)
- Congenital Disorder of Glycosylation, Type Ib (MPI)
- Congenital Disorder of Glycosylation, Type Ic (ALG6)
- Congenital Finnish Nephrosis (NPHS1)
- Costeff Optic Atrophy Syndrome (OPA3)
- Cystic Fibrosis (CFTR) ACOG ACMG
- Cystinosis (CTNS)
- D-Bifunctional Protein Deficiency (HSD17B4)
- Delta-Sarcoglycanopathy (SGCD)
- Dysferlinopathy (DYSF)
- Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy)(DMD) X-linked
- ERCC6-Related Disorders (ERCC6)
- ERCC8-Related Disorders (ERCC8)
- EVC-Related Ellis-Van Creveld Syndrome (EVC)
- EVC2-Related Ellis-Van Creveld Syndrome (EVC2)
- Fabry Disease (GLA) X-linked
- Familial Dysautonomia (IKBKAP) ACOG ACMG
- Familial Mediterranean Fever (MEFV)
- Fanconi Anemia Complementation, Group A (FANCA)
- Fanconi Anemia, Type C (FANCC) ACMG
- FKRP-Related Disorders (FKRP)
- FKTN-Related Disorders (including Walker-Warburg Syndrome) (FKTN)
- Fragile X Syndrome (FMR1)* X-linked
- Galactokinase Deficiency (GALK1)
- Galactosemia (GALT)
- Gamma-Sarcoglycanopathy (SGCG)
- Gaucher Disease (GBA)* ACMG
- GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) (GJB2)
- GLB1-Related Disorders (GLB1)
- GLDC-Related Glycine Encephalopathy (GLDC)
- Glutaric Acidemia, Type 1 (GCDH)
- Glycogen Storage Disease, Type Ia (G6PC)
- Glycogen Storage Disease, Type Ib (SLC37A4)
- Glycogen Storage Disease, Type III (AGL)
- GNPTAB-Related Disorders (GNPTAB)
- GRACILE Syndrome (BCS1L)
- HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (HADHA)
- Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (HBB) ACOG
- Hereditary Fructose Intolerance (ALDOB)
- Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related (LAMA3)
- Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related (LAMB3)
- Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (LAMC2)

Abbott Pathology · Dorevitch Pathology · Laverty Pathology · ML Pathology (Specialists in Private Pathology since the 1920s) · TML Pathology (Tasmanian Medical Laboratories) · Western Diagnostic Pathology

- Hexosaminidase A Deficiency (including Tay-Sachs Disease) (HEXA) ACOG ACMG
- HMG-CoA Lyase Deficiency (HMGCL)
- Holocarboxylase Synthetase Deficiency (HLCS)
- Homocystinuria caused by Cystathionine Beta-Synthase Deficiency (CBS)
- Hydrolethalus Syndrome (HYLS1)
- Hypophosphatasia, Autosomal Recessive (ALPL)
- Inclusion Body Myopathy 2 (GNE)
- Isovaleric Acidemia (IVD)
- Joubert Syndrome 2 (TMEM216)
- KCNJ11-Related Familial Hyperinsulinism (KCNJ11)
- Krabbe Disease (GALC)
- LAMA2-Related Muscular Dystrophy (LAMA2)
- Leigh Syndrome, French-Canadian Type (LRPPRC)
- Lipoamide Dehydrogenase Deficiency (DLD)
- Lipoid Congenital Adrenal Hyperplasia (STAR)
- Lysosomal Acid Lipase Deficiency (LIPA)
- Maple Syrup Urine Disease, Type Ia (BCKDHA)
- Maple Syrup Urine Disease, Type IB (BCKDHB)
- Maple Syrup Urine Disease, Type II (DBT)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM)
- Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1)
- Metachromatic Leukodystrophy (ARSA)
- Methylmalonic Acidemia, cblA Type (MMAA)
- Methylmalonic Acidemia, cblB Type (MMAB)
- Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC)
- MKS1-Related Disorders (MKS1)
- Mucolipidosis III Gamma (GNPTG)
- Mucolipidosis IV (MCOLN1) ACMG
- Mucopolysaccharidosis, Type I (including Hurler Syndrome) (IDUA)
- Mucopolysaccharidosis, Type II (IDS) X-linked
- Mucopolysaccharidosis, Type IIIA (SGSH)
- Mucopolysaccharidosis, Type IIIB (NAGLU)
- Mucopolysaccharidosis, Type IIIC (HGSNAT)
- Muscle-Eye-Brain Disease (POMGNT1)
- MUT-Related Methylmalonic Acidemia (MUT)
- MYO7A-Related Disorders (MYO7A)
- NEB-Related Nemaline Myopathy (NEB)
- Niemann-Pick Disease, Type C (NPC1)
- Niemann-Pick Disease, Type C2 (NPC2)
- Niemann-Pick Disease, SMPD1-Associated (SMPD1) ACMG
- Nijmegen Breakage Syndrome (NBN)
- Northern Epilepsy (CLN8)
- Ornithine Transcarbamylase Deficiency (OTC) X-linked
- PCCA-Related Propionic Acidemia (PCCA)
- PCCB-Related Propionic Acidemia (PCCB)
- PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (PCDH15)
- Pendred Syndrome (SLC26A4)
- Peroxisome Biogenesis Disorder, Type 3 (PEX12)
- Peroxisome Biogenesis Disorder, Type 4 (PEX6)
- Peroxisome Biogenesis Disorder, Type 5 (PEX2)
- Peroxisome Biogenesis Disorder, Type 6 (PEX10)
- PEX1-Related Zellweger Syndrome Spectrum (PEX1)
- Phenylalanine Hydroxylase Deficiency (PAH)
- PKHD1-Related Autosomal Recessive Polycystic Kidney Disease (PKHD1)
- Polyglandular Autoimmune Syndrome, Type 1 (AIRE)
- Pompe Disease (GAA)
- PPT1-Related Neuronal Ceroid Lipofuscinosis (PPT1)
- Primary Carnitine Deficiency (SLC22A5)
- Primary Hyperoxaluria, Type 1 (AGXT)
- Primary Hyperoxaluria, Type 2 (GRHPR)
- Primary Hyperoxaluria, Type 3 (HOGA1)
- PROP1-Related Combined Pituitary Hormone Deficiency (PROP1)
- Pycnodysostosis (CTSK)
- Pyruvate Carboxylase Deficiency (PC)
- Rhizomelic Chondrodysplasia Punctata, Type 1 (PEX7)
- RTEL1-Related Disorders (RTEL1)
- Salla Disease (SLC17A5)
- Sandhoff Disease (HEXB)
- Segawa Syndrome (TH)
- Short Chain Acyl-CoA Dehydrogenase Deficiency (ACADS)
- Sjogren-Larsson Syndrome (ALDH3A2)
- Smith-Lemli-Opitz Syndrome (DHCR7)
- Spastic Paraplegia, Type 15 (ZFYVE26)
- Spinal Muscular Atrophy (SMN1)* ACOG ACMG
- Spondylothoracic Dysostosis (MESP2)
- Steroid-Resistant Nephrotic Syndrome (NPHS2)
- Sulfate Transporter-Related Osteochondrodysplasia (SLC26A2)
- TGM1-Related Autosomal Recessive Congenital Ichthyosis (TGM1)
- TPP1-Related Neuronal Ceroid Lipofuscinosis (TPP1)
- Tyrosinemia, Type I (FAH)
- Tyrosinemia, Type II (TAT)
- USH1C-Related Disorders (USH1C)
- USH2A-Related Disorders (USH2A)
- Usher Syndrome, Type 3 (CLRN1)
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL)
- Wilson Disease (ATP7B)
- X-Linked Congenital Adrenal Hypoplasia (NR0B1) X-linked
- X-Linked Juvenile Retinoschisis (RS1) X-linked
- X-Linked Myotubular Myopathy (MTM1) X-linked
- X-Linked Severe Combined Immunodeficiency (IL2RG) X-linked
- Xeroderma Pigmentosum, Group A (XPA)
- Xeroderma Pigmentosum, Group C (XPC)

ACOG
Indicates testing recommended by American College of Obstetricians & Gynaecologists (ACOG)

ACMG
Indicates testing recommended by American College of Medical Genetics (ACMG)

X-linked
Indicates X-linked disorders

*Analysed using custom assay

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