Genomic Diagnostics
LEADING THE WAY TO IMPROVE HEALTH

Myriad genetics

Myriad Genetics Foresight® Carrier Screen Disease List

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

Congenital Adrenal Hyperplasia, CYP11B1-Related *(CYP11B1)*

6-Pyruvoyl-Tetrahydropterin Synthase Deficiency *(PTS)*

Familial Hyperinsulinism, ABCC8-Related *(ABCC8)* ACMG

Adenosine Deaminase Deficiency *(ADA)*

Adrenoleukodystrophy, X-Linked *(ABCD1)* ACMG 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)*

Glycine Encephalopathy, AMT-Related *(AMT)*

Andermann Syndrome *(SLC12A6)*

Argininemia *(ARG1)*

Argininosuccinic Aciduria *(ASL)* ACMG

Aspartylglycosaminuria *(AGA)* ACMG

Ataxia with Vitamin E Deficiency *(TTPA)*

Ataxia-Telangiectasia *(ATM)*

ATP7A-Related Disorders *(ATP7A)* X-linked

Autoimmune Polyglandular Syndrome Type 1 *(AIRE)* ACMG

Autosomal Recessive Osteopetrosis, Type 1 *(TCIRG1)*

Autosomal Recessive Polycystic Kidney Disease, PKHD1-Related *(PKHD1)* ACMG

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay *(SACS)*

Bardet-Biedl Syndrome, BBS1-Related *(BBS1)* ACMG

Bardet-Biedl Syndrome, BBS10-Related *(BBS10)*

Bardet-Biedl Syndrome, BBS12-Related *(BBS12)*

Bardet-Biedl Syndrome, BBS2-Related *(BBS2)* ACMG

BCS1L-Related Disorders *(BCS1L)*

Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) *(SGCB)*

Biotinidase Deficiency *(BTD)* ACMG

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)* ACMG

Cartilage-Hair Hypoplasia *(RMRP)*

Cerebrotendinous Xanthomatosis *(CYP27A1)* ACMG

Citrullinemia, Type 1 *(ASS1)*

CLN3-Related Neuronal Ceroid Lipofuscinosis *(CLN3)*

CLN5-Related Neuronal Ceroid Lipofuscinosis *(CLN5)*

Neuronal Ceroid Lipofuscinosis, CLN6-Related *(CLN6)*

CLN8-Related Neuronal Ceroid Lipofuscinosis *(CLN8)*

Cohen Syndrome *(VPS13B)*

COL4A3-Related Alport Syndrome *(COL4A3)*

COL4A4-Related Alport Syndrome *(COL4A4)*

Combined Pituitary Hormone Deficiency, PROP1-Related *(PROP1)*

Congenital Adrenal Hyperplasia, CYP21A2-Related *(CYP21A2)** ACMG

Congenital Disorder of Glycosylation, MPI-Related *(MPI)*

Congenital Disorder of Glycosylation, Type Ia *(PMM2)* ACMG

Congenital Disorder of Glycosylation, Type Ic *(ALG6)*

Costeff Optic Atrophy Syndrome *(OPA3)*

Cystic Fibrosis *(CFTR)* ACOG ACMG

Cystinosis *(CTNS)*

D-Bifunctional Protein Deficiency *(HSD17B4)*

Delta-Sarcoglycanopathy *(SGCD)*

Dihydrolipoamide Dehydrogenase Deficiency *(DLD)* ACMG

Dysferlinopathy *(DYSF)*

Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy)*(DMD)* ACMG X-linked

ERCC6-Related Disorders *(ERCC6)*

ERCC8-Related Disorders *(ERCC8)*

EVC-Related Ellis-Van Creveld Syndrome *(EVC)*

EVC2-Related Ellis-Van Creveld Syndrome *(EVC2)* ACMG

Fabry Disease *(GLA)* ACMG X-linked

Familial Dysautonomia *(ELP1)* ACOG ACMG

Familial Mediterranean Fever *(MEFV)*

Fanconi Anemia Complementation, Group A *(FANCA)*

Fanconi Anemia, FANCC-Related *(FANCC)* ACMG

FKRP-Related Disorders *(FKRP)* ACMG

FKTN-Related Disorders (including Walker-Warburg Syndrome) *(FKTN)* ACMG

Fragile X Syndrome *(FMR1)** ACMG X-linked

Galactokinase Deficiency *(GALK1)*

Galactosemia *(GALT)* ACMG

Gamma-Sarcoglycanopathy *(SGCG)*

Gaucher Disease *(GBA)** ACMG

GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) *(GJB2)* ACMG

GLB1-Related Disorders *(GLB1)*

GLDC-Related Glycine Encephalopathy *(GLDC)*

Glutaric Acidemia, GCDH-Related *(GCDH)*

Glycogen Storage Disease, Type Ia *(G6PC1)* ACMG

Glycogen Storage Disease, Type Ib *(SLC37A4)* ACMG

Glycogen Storage Disease, Type III *(AGL)*

GNE Myopathy *(GNE)*

GNPTAB-Related Disorders *(GNPTAB)* ACMG

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 ACMG

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Hereditary Fructose Intolerance *(ALDOB)* ACMG

Junctional Epidermolysis Bullosa, LAMB3-Related *(LAMB3)*

Hexosaminidase A Deficiency (including Tay-Sachs Disease) *(HEXA)* ACOG ACMG

HMG-CoA Lyase Deficiency *(HMGCL)*

Holocarboxylase Synthetase Deficiency *(HLCS)*

Homocystinuria, CBS-Related *(CBS)* ACMG

Hydrolethalus Syndrome *(HYLS1)*

Hypophosphatasia *(ALPL)* ACMG

Isovaleric Acidemia *(IVD)*

Joubert Syndrome 2 *(TMEM216)* ACMG

Junctional Epidermolysis Bullosa, LAMC2-Related *(LAMC2)*

Junctional Epidermolysis Bullosa, LAMA3-Related *(LAMA3)*

Familial Hyperinsulinism, KCNJ11-Related *(KCNJ11)*

Krabbe Disease *(GALC)*

Muscular Dystrophy, LAMA2-Related *(LAMA2)*

Leigh Syndrome, French-Canadian Type *(LRPPRC)*

Lipoid Congenital Adrenal Hyperplasia *(STAR)*

Lysosomal Acid Lipase Deficiency *(LIPA)*

Maple Syrup Urine Disease, Type Ia *(BCKDHA)*

Maple Syrup Urine Disease, Type Ib *(BCKDHB)* ACMG

Maple Syrup Urine Disease, Type II *(DBT)*

Medium Chain Acyl-CoA Dehydrogenase Deficiency *(ACADM)* ACMG

Megalencephalic Leukoencephalopathy with Subcortical Cysts *(MLC1)* ACMG

Metachromatic Leukodystrophy *(ARSA)* ACMG

Methylmalonic Acidemia, cblA Type *(MMAA)*

Methylmalonic Acidemia, cblB Type *(MMAB)*

Methylmalonic Aciduria and Homocystinuria, cblC Type *(MMACHC)* ACMG

MKS1-Related Disorders *(MKS1)*

Mucolipidosis III Gamma *(GNPTG)*

Mucolipidosis IV *(MCOLN1)* ACMG

Mucopolysaccharidosis, Type I (including Hurler Syndrome) *(IDUA)* ACMG

Mucopolysaccharidosis, Type II *(IDS)* X-linked

Mucopolysaccharidosis, Type IIIA *(SGSH)*

Mucopolysaccharidosis, Type IIIB *(NAGLU)*

Mucopolysaccharidosis, Type IIIC *(HGSNAT)*

MMUT-Related Methylmalonic Acidemia *(MMUT)* ACMG

MYO7A-Related Disorders *(MYO7A)*

NEB-Related Nemaline Myopathy *(NEB)* ACMG

Nephrotic Syndrome, NPHS1-Related *(NPHS1)* ACMG

Niemann-Pick Disease, SMPD1-Related *(SMPD1)* ACMG

Niemann-Pick Disease, Type C1 *(NPC1)*

Niemann-Pick Disease, Type C2 *(NPC2)*

Nijmegen Breakage Syndrome *(NBN)*

Ornithine Transcarbamylase Deficiency *(OTC)* ACMG X-linked

PCCA-Related Propionic Acidemia *(PCCA)*

PCCB-Related Propionic Acidemia *(PCCB)*

PCDH15-Related Disorders (including Usher Syndrome, Type 1F) *(PCDH15)* ACMG

Pendred Syndrome *(SLC26A4)* ACMG

Peroxisome Biogenesis Disorder, Type 1 *(PEX1)*

Peroxisome Biogenesis Disorder, Type 3 *(PEX12)*

Peroxisome Biogenesis Disorder, Type 4 *(PEX6)*

Peroxisome Biogenesis Disorder, Type 5 *(PEX2)*

Peroxisome Biogenesis Disorder, Type 6 *(PEX10)*

Phenylalanine Hydroxylase Deficiency *(PAH)* ACMG

POMGNT-Related Disorders *(POMGNT1)*

Pompe Disease *(GAA)* ACMG

PPT1-Related Neuronal Ceroid Lipofuscinosis *(PPT1)*

Primary Carnitine Deficiency *(SLC22A5)*

Primary Hyperoxaluria, Type 1 *(AGXT)* ACMG

Primary Hyperoxaluria, Type 2 *(GRHPR)*

Primary Hyperoxaluria, Type 3 *(HOGA1)*

Pycnodysostosis *(CTSK)*

Pyruvate Carboxylase Deficiency *(PC)*

Rhizomelic Chondrodysplasia Punctata, Type 1 *(PEX7)*

RTEL1-Related Disorders *(RTEL1)*

Salla Disease *(SLC17A5)*

Sandhoff Disease *(HEXB)*

Short Chain Acyl-CoA Dehydrogenase Deficiency *(ACADS)*

Sjogren-Larsson Syndrome *(ALDH3A2)*

SLC26A2-Related Disorders *(SLC26A2)* ACMG

Smith-Lemli-Opitz Syndrome *(DHCR7)* ACMG

Spastic Paraplegia, Type 15 *(ZFYVE26)*

Spinal Muscular Atrophy *(SMN1)** ACOG ACMG

Spondylothoracic Dysostosis *(MESP2)*

Steroid-Resistant Nephrotic Syndrome *(NPHS2)*

TGM1-Related Autosomal Recessive Congenital Ichthyosis *(TGM1)*

TPP1-Related Neuronal Ceroid Lipofuscinosis *(TPP1)*

Tyrosine Hydroxylase Deficiency *(TH)*

Tyrosinemia, Type I *(FAH)* ACMG

Tyrosinemia, Type II *(TAT)*

USH1C-Related Disorders *(USH1C)*

USH2A-Related Disorders *(USH2A)* ACMG

Usher Syndrome, Type 3 *(CLRN1)* ACMG

Very Long Chain Acyl-CoA Dehydrogenase Deficiency *(ACADVL)* ACMG

Wilson Disease *(ATP7B)* ACMG

X-linked Adrenal Hypoplasia Congenita *(NR0B1)* ACMG X-linked

X-Linked Juvenile Retinoschisis *(RS1)* ACMG 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)* ACMG

ACOG Indicates disease listed in American Congress of Obstetricians and Gynaecologists (ACOG) guidelines

ACMG
Indicates disease listed in American College of Medical Genetics (ACMG) guidelines

X-linked
Indicates X-linked disorders

*Analyzed using custom assay

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