

Universal panel 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.

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*)

Dihydroliipoamide 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 (<i>G6PC1</i>) ACMG	Maple Syrup Urine Disease, Type Ib (<i>BCKDHB</i>) ACMG	PCCA-Related Propionic Acidemia (<i>PCCA</i>)	Spastic Paraplegia, Type 15 (<i>ZFYVE26</i>)
Glycogen Storage Disease, Type Ib (<i>SLC37A4</i>) ACMG	Maple Syrup Urine Disease, Type II (<i>DBT</i>)	PCCB-Related Propionic Acidemia (<i>PCCB</i>)	Spinal Muscular Atrophy (<i>SMN1</i>)* ACOG ACMG
Glycogen Storage Disease, Type III (<i>AGL</i>)	Medium Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADM</i>) ACMG	PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (<i>PCDH15</i>) ACMG	Spondylothoracic Dysostosis (<i>MESP2</i>)
GNE Myopathy (<i>GNE</i>)	Megalencephalic Leukoencephalopathy with Subcortical Cysts (<i>MLC1</i>) ACMG	Pendred Syndrome (<i>SLC26A4</i>) ACMG	Steroid-Resistant Nephrotic Syndrome (<i>NPHS2</i>)
GNPTAB-Related Disorders (<i>GNPTAB</i>) ACMG	Metachromatic Leukodystrophy (<i>ARSA</i>) ACMG	Peroxisome Biogenesis Disorder, Type 1 (<i>PEX1</i>)	TGM1-Related Autosomal Recessive Congenital Ichthyosis (<i>TGM1</i>)
HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) (<i>HADHA</i>)	Methylmalonic Acidemia, cblA Type (<i>MMAA</i>)	Peroxisome Biogenesis Disorder, Type 3 (<i>PEX2</i>)	TPP1-Related Neuronal Ceroid Lipofuscinosis (<i>TPP1</i>)
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)(<i>HBB</i>) ACOG ACMG	Methylmalonic Acidemia, cblB Type (<i>MMAB</i>)	Peroxisome Biogenesis Disorder, Type 4 (<i>PEX6</i>)	Tyrosine Hydroxylase Deficiency (<i>TH</i>)
Hereditary Fructose Intolerance (<i>ALDOB</i>) ACMG	Methylmalonic Aciduria and Homocystinuria, cblC Type (<i>MMACHC</i>) ACMG	Peroxisome Biogenesis Disorder, Type 5 (<i>PEX2</i>)	Tyrosinemia, Type I (<i>FAH</i>) ACMG
Junctional Epidermolysis Bullosa, LAMB3-Related (<i>LAMB3</i>)	MKS1-Related Disorders (<i>MKS1</i>)	Peroxisome Biogenesis Disorder, Type 6 (<i>PEX10</i>)	Tyrosinemia, Type II (<i>TAT</i>)
Hexosaminidase A Deficiency (including Tay-Sachs Disease) (<i>HEXA</i>) ACOG ACMG	Mucopolysaccharidosis, Type I (including Hurler Syndrome) (<i>IDUA</i>) ACMG	Phenylalanine Hydroxylase Deficiency (<i>PAH</i>) ACMG	USH1C-Related Disorders (<i>USH1C</i>)
HMG-CoA Lyase Deficiency (<i>HMGL1</i>)	Mucopolysaccharidosis, Type II (<i>IDS</i>) X-linked	POMGNT-Related Disorders (<i>POMGNT1</i>)	USH2A-Related Disorders (<i>USH2A</i>) ACMG
Holocarboxylase Synthetase Deficiency (<i>HLCS</i>)	Mucopolysaccharidosis, Type IIIA (<i>SGSH</i>)	Pompe Disease (<i>GAA</i>) ACMG	Usher Syndrome, Type 3 (<i>CLRN1</i>) ACMG
Homocystinuria, CBS-Related (<i>CBS</i>) ACMG	Mucopolysaccharidosis, Type IIIB (<i>NAGLU</i>)	PPT1-Related Neuronal Ceroid Lipofuscinosis (<i>PPT1</i>)	Very Long Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADVL</i>) ACMG
Hydroletharus Syndrome (<i>HYLS1</i>)	Mucopolysaccharidosis, Type IIIC (<i>HGSNAT</i>)	Primary Carnitine Deficiency (<i>SLC22A5</i>)	Wilson Disease (<i>ATP7B</i>) ACMG
Hypophosphatasia (<i>ALPL</i>) ACMG	MMUT-Related Methylmalonic Acidemia (<i>MMUT</i>) ACMG	Primary Hyperoxaluria, Type 1 (<i>AGXT</i>) ACMG	X-linked Adrenal Hypoplasia Congenita (<i>NROB1</i>) ACMG X-linked
Isovaleric Acidemia (<i>IVD</i>)	MYO7A-Related Disorders (<i>MYO7A</i>)	Primary Hyperoxaluria, Type 2 (<i>GRHPR</i>)	X-Linked Juvenile Retinoschisis (<i>RS1</i>) ACMG X-linked
Joubert Syndrome 2 (<i>TMEM216</i>) ACMG	NEB-Related NemaLine Myopathy (<i>NEB</i>) ACMG	Primary Hyperoxaluria, Type 3 (<i>HOGA1</i>)	X-Linked Myotubular Myopathy (<i>MTM1</i>) X-linked
Junctional Epidermolysis Bullosa, LAMC2-Related (<i>LAMC2</i>)	Nephrotic Syndrome, NPHS1-Related (<i>NPHS1</i>) ACMG	Pycnodysostosis (<i>CTSK</i>)	X-Linked Severe Combined Immunodeficiency (<i>IL2RG</i>) X-linked
Junctional Epidermolysis Bullosa, LAMA3-Related (<i>LAMA3</i>)	Niemann-Pick Disease, SMPD1-Related (<i>SMPD1</i>) ACMG	Pyruvate Carboxylase Deficiency (<i>PC</i>)	Xeroderma Pigmentosum, Group A (<i>XPA</i>)
Familial Hyperinsulinism, KCNJ11-Related (<i>KCNJ11</i>)	Niemann-Pick Disease, Type C1 (<i>NPC1</i>)	Rhizomelic Chondrodysplasia Punctata, Type 1 (<i>PEX7</i>)	Xeroderma Pigmentosum, Group C (<i>XPC</i>) ACMG
Krabbe Disease (<i>GALC</i>)	Niemann-Pick Disease, Type C2 (<i>NPC2</i>)	RTEL1-Related Disorders (<i>RTEL1</i>)	
Muscular Dystrophy, LAMA2-Related (<i>LAMA2</i>)	Nijmegen Breakage Syndrome (<i>NBN</i>)	Salla Disease (<i>SLC17A5</i>)	ACOG
Leigh Syndrome, French-Canadian Type (<i>LRPPRC</i>)	Ornithine Transcarbamylase Deficiency (<i>OTC</i>) ACMG X-linked	Sandhoff Disease (<i>HEXB</i>)	Indicates disease listed in ACOG guidelines
Lipoid Congenital Adrenal Hyperplasia (<i>STAR</i>)		Short Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADS</i>)	ACMG
Lysosomal Acid Lipase Deficiency (<i>LIPA</i>)		Sjogren-Larsson Syndrome (<i>ALDH3A2</i>)	Indicates disease listed in ACMG guidelines
Maple Syrup Urine Disease, Type Ia (<i>BCKDHA</i>)		SLC26A2-Related Disorders (<i>SLC26A2</i>) ACMG	X-linked
		Smith-Lemli-Opitz Syndrome (<i>DHCR7</i>) ACMG	Indicates X-linked disorders

*Analyzed using custom assay