

## 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 (G6PC) <a href="#">ACMG</a>	Maple Syrup Urine Disease, Type Ib (BCKDHB) <a href="#">ACMG</a>	PCCA-Related Propionic Acidemia (PCCA)	Spastic Paraplegia, Type 15 (ZFYE26)
Glycogen Storage Disease, Type Ib (SLC37A4) <a href="#">ACMG</a>	Maple Syrup Urine Disease, Type II (DBT)	PCCB-Related Propionic Acidemia (PCCB)	Spinal Muscular Atrophy (SMN1)* <a href="#">ACOG</a> <a href="#">ACMG</a>
Glycogen Storage Disease, Type III (AGL)	Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM) <a href="#">ACMG</a>	PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (PCDH15) <a href="#">ACMG</a>	Spondylothoracic Dysostosis (MESP2)
GNE Myopathy (GNE)	Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1) <a href="#">ACMG</a>	Pendred Syndrome (SLC26A4) <a href="#">ACMG</a>	Steroid-Resistant Nephrotic Syndrome (NPHS2)
GNPTAB-Related Disorders (GNPTAB) <a href="#">ACMG</a>	Metachromatic Leukodystrophy (ARSA) <a href="#">ACMG</a>	Peroxisome Biogenesis Disorder, Type 1 (PEX1)	TGM1-Related Autosomal Recessive Congenital Ichthyosis (TGM1)
HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) (HADHA)	Methylmalonic Acidemia, cblA Type (MMAA)	Peroxisome Biogenesis Disorder, Type 3 (PEX2)	TPP1-Related Neuronal Ceroid Lipofuscinosis (TPP1)
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)(HBB) <a href="#">ACOG</a> <a href="#">ACMG</a>	Methylmalonic Acidemia, cblB Type (MMAB)	Peroxisome Biogenesis Disorder, Type 4 (PEX6)	Tyrosine Hydroxylase Deficiency (TH)
Hereditary Fructose Intolerance (ALDOB) <a href="#">ACMG</a>	Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC) <a href="#">ACMG</a>	Peroxisome Biogenesis Disorder, Type 5 (PEX2)	Tyrosinemia, Type I (FAH) <a href="#">ACMG</a>
Junctional Epidermolysis Bullosa, LAMB3-Related (LAMB3)	MKS1-Related Disorders (MKS1)	Peroxisome Biogenesis Disorder, Type 6 (PEX10)	Tyrosinemia, Type II (TAT)
Hexosaminidase A Deficiency (including Tay-Sachs Disease) (HEXA) <a href="#">ACOG</a> <a href="#">ACMG</a>	Mucopolipidosis III Gamma (GNPTG)	Phenylalanine Hydroxylase Deficiency (PAH) <a href="#">ACMG</a>	USH1C-Related Disorders (USH1C)
HMG-CoA Lyase Deficiency (HMGL)	Mucopolipidosis IV (MCOLN1) <a href="#">ACMG</a>	POMGNT-Related Disorders (POMGNT1)	USH2A-Related Disorders (USH2A) <a href="#">ACMG</a>
Holocarboxylase Synthetase Deficiency (HLCS)	Mucopolysaccharidosis, Type I (including Hurler Syndrome) (IDUA) <a href="#">ACMG</a>	Pompe Disease (GAA) <a href="#">ACMG</a>	Usher Syndrome, Type 3 (CLRN1) <a href="#">ACMG</a>
Homocystinuria, CBS-Related (CBS) <a href="#">ACMG</a>	Mucopolysaccharidosis, Type II (IDS) <a href="#">X-linked</a>	PPT1-Related Neuronal Ceroid Lipofuscinosis (PPT1)	Very Long Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) <a href="#">ACMG</a>
Hydroletharus Syndrome (HYLS1)	Mucopolysaccharidosis, Type IIIA (SGSH)	Primary Carnitine Deficiency (SLC22A5)	Wilson Disease (ATP7B) <a href="#">ACMG</a>
Hypophosphatasia (ALPL) <a href="#">ACMG</a>	Mucopolysaccharidosis, Type IIIB (NAGLU)	Primary Hyperoxaluria, Type 1 (AGXT) <a href="#">ACMG</a>	X-linked Adrenal Hypoplasia Congenita (NROB1) <a href="#">ACMG</a> <a href="#">X-linked</a>
Isovaleric Acidemia (IVD)	Mucopolysaccharidosis, Type IIIC (HGSNAT)	Primary Hyperoxaluria, Type 2 (GRHPR)	X-Linked Juvenile Retinoschisis (RS1) <a href="#">ACMG</a> <a href="#">X-linked</a>
Joubert Syndrome 2 (TMEM216) <a href="#">ACMG</a>	MUT-Related Methylmalonic Acidemia (MUT)	Primary Hyperoxaluria, Type 3 (HOGA1)	X-Linked Myotubular Myopathy (MTM1) <a href="#">X-linked</a>
Junctional Epidermolysis Bullosa, LAMC2-Related (LAMC2)	MYO7A-Related Disorders (MYO7A)	Pycnodysostosis (CTSK)	X-Linked Severe Combined Immunodeficiency (IL2RG) <a href="#">X-linked</a>
Junctional Epidermolysis Bullosa, LAMA3-Related (LAMA3)	NEB-Related NemaLine Myopathy (NEB) <a href="#">ACMG</a>	Pyruvate Carboxylase Deficiency (PC)	Xeroderma Pigmentosum, Group A (XPA)
Familial Hyperinsulinism, KCNJ11-Related (KCNJ11)	Nephrotic Syndrome, NPHS1-Related (NPHS1) <a href="#">ACMG</a>	Rhizomelic Chondrodysplasia Punctata, Type 1 (PEX7)	Xeroderma Pigmentosum, Group C (XPC) <a href="#">ACMG</a>
Krabbe Disease (GALC)	Niemann-Pick Disease, SMPD1-Related (SMPD1) <a href="#">ACMG</a>	RTEL1-Related Disorders (RTEL1)	<a href="#">ACOG</a>
Muscular Dystrophy, LAMA2-Related (LAMA2)	Niemann-Pick Disease, Type C1 (NPC1)	Salla Disease (SLC17A5)	Indicates disease listed in ACOG guidelines
Leigh Syndrome, French-Canadian Type (LRPPRC)	Niemann-Pick Disease, Type C2 (NPC2)	Sandhoff Disease (HEXB)	<a href="#">ACMG</a>
Lipoid Congenital Adrenal Hyperplasia (STAR)	Nijmegen Breakage Syndrome (NBN)	Short Chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	Indicates disease listed in ACMG guidelines
Lysosomal Acid Lipase Deficiency (LIPA)	Ornithine Transcarbamylase Deficiency (OTC) <a href="#">ACMG</a> <a href="#">X-linked</a>	Sjogren-Larsson Syndrome (ALDH3A2)	<a href="#">X-linked</a>
Maple Syrup Urine Disease, Type Ia (BCKDHA)		SLC26A2-Related Disorders (SLC26A2) <a href="#">ACMG</a>	Indicates X-linked disorders
		Smith-Lemli-Opitz Syndrome (DHCR7) <a href="#">ACMG</a>	*Analyzed using custom assay