

Universal panel disease list

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

6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (PTS)	Bardet-Biedl Syndrome, BBS1-Related (BBS1) ACMG	COL4A3-Related Alport Syndrome (COL4A3)	Fabry Disease (GLA) ACMG X-linked
Adenosine Deaminase Deficiency (ADA)	Bardet-Biedl Syndrome, BBS10-Related (BBS10)	COL4A4-Related Alport Syndrome (COL4A4)	Familial Dysautonomia (ELP1) ACOG ACMG
Adrenoleukodystrophy, X-Linked (ABCD1) ACMG X-linked	Bardet-Biedl Syndrome, BBS12-Related (BBS12)	Combined Pituitary Hormone Deficiency, PROP1-Related (PROP1)	Familial Hyperinsulinism, ABCC8-Related (ABCC8) ACMG
Alpha Thalassemia (HBA1/HBA2) ACOG ACMG	Bardet-Biedl Syndrome, BBS2-Related (BBS2) ACMG	Congenital Adrenal Hyperplasia, CYP11B1-Related (CYP11B1)	Familial Mediterranean Fever (MEFV)
Alpha-Mannosidosis (MAN2B1)	BCS1L-Related Disorders (BCS1L)	Congenital Adrenal Hyperplasia, CYP21A2-Related (CYP21A2) ACMG	Fanconi Anemia Complementation, Group A (FANCA)
Alpha-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2D) (SGCA)	Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) (SGCB)	Congenital Disorder of Glycosylation, MPI-Related (MPI)	Fanconi Anemia, FANCC-Related (FANCC) ACMG
Alport Syndrome, X-Linked (COL4A5) X-linked	Biotinidase Deficiency (BTD) ACMG	Congenital Disorder of Glycosylation, Type Ia (PMM2) ACMG	FKRP-Related Disorders (FKRP) ACMG
Alstrom Syndrome (ALMS1)	Bloom Syndrome (BLM) ACMG	Congenital Disorder of Glycosylation, Type Ic (ALG6) ACMG	FKTN-Related Disorders (including Walker-Warburg Syndrome) (FKTN) ACMG
Glycine Encephalopathy, AMT-Related (AMT)	Calpainopathy (CAPN3)	Congenital Disorder of Glycosylation, Type Ie (ALG3) ACMG	Fragile X Syndrome (FMR1) ACMG X-linked
Andermann Syndrome (SLC12A6)	Canavan Disease (ASPA) ACOG ACMG	Costeff Optic Atrophy Syndrome (OPA3)	Galactokinase Deficiency (GALK1)
Argininemia (ARG1)	Carbamoylphosphate Synthetase I Deficiency (CPS1)	Cystic Fibrosis (CFTR) ACOG ACMG	Galactosemia (GALT) ACMG
Argininosuccinic Aciduria (ASL) ACMG	Carnitine Palmitoyltransferase IA Deficiency (CPT1A)	Cystinosis (CTNS)	Gamma-Sarcoglycanopathy (SGCG)
Aspartylglycosaminuria (AGA) ACMG	Carnitine Palmitoyltransferase II Deficiency (CPT2) ACMG	D-Bifunctional Protein Deficiency (HSD17B4)	Gaucher Disease (GBA) ACMG
Ataxia with Vitamin E Deficiency (TTPA)	Cartilage-Hair Hypoplasia (RMRP)	Delta-Sarcoglycanopathy (SGCD)	GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) (GJB2) ACMG
Ataxia-Telangiectasia (ATM)	Cerebrotendinous Xanthomatosis (CYP27A1) ACMG	Dihydroliipoamide Dehydrogenase Deficiency (DLD) ACMG	GLB1-Related Disorders (GLB1)
ATP7A-Related Disorders (ATP7A) X-linked	Citrullinemia, Type 1 (ASS1)	Dysferlinopathy (DYSF)	GLDC-Related Glycine Encephalopathy (GLDC)
Autoimmune Polyglandular Syndrome Type 1 (AIRE) ACMG	CLN3-Related Neuronal Ceroid Lipofuscinosis (CLN3)	Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy)(DMD) ACMG X-linked	Glutaric Acidemia, GCDH-Related (GCDH)
Autosomal Recessive Osteopetrosis, Type 1 (TCIRG1)	CLN5-Related Neuronal Ceroid Lipofuscinosis (CLN5)	ERCC6-Related Disorders (ERCC6)	Glycogen Storage Disease, Type Ia (G6PC1) ACMG
Autosomal Recessive Polycystic Kidney Disease, PKHD1-Related (PKHD1) ACMG	Neuronal Ceroid Lipofuscinosis, CLN6-Related (CLN6)	ERCC8-Related Disorders (ERCC8)	Glycogen Storage Disease, Type Ib (SLC37A4) ACMG
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (SACS)	CLN8-Related Neuronal Ceroid Lipofuscinosis (CLN8)	EVC-Related Ellis-Van Creveld Syndrome (EVC)	
	Cohen Syndrome (VPS13B)	EVC2-Related Ellis-Van Creveld Syndrome (EVC2) ACMG	

Glycogen Storage Disease, Type III (<i>AGL</i>)	Maple Syrup Urine Disease, Type II (<i>DBT</i>)	PCCB-Related Propionic Acidemia (<i>PCCB</i>)	Spastic Paraplegia, Type 15 (<i>ZFYVE26</i>)
GNE Myopathy (<i>GNE</i>)	Medium Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADM</i>) ACMG	PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (<i>PCDH15</i>) ACMG	Spinal Muscular Atrophy (<i>SMN1</i>) ACOG ACMG
GNPTAB-Related Disorders (<i>GNPTAB</i>) ACMG	Megalencephalic Leukoencephalopathy with Subcortical Cysts (<i>MLC1</i>) ACMG	Pendred Syndrome (<i>SLC26A4</i>) ACMG	Spondylothoracic Dysostosis (<i>MESP2</i>)
HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) (<i>HADHA</i>)	Metachromatic Leukodystrophy (<i>ARSA</i>) ACMG	Peroxisome Biogenesis Disorder, Type 1 (<i>PEX1</i>)	Steroid-Resistant Nephrotic Syndrome (<i>NPHS2</i>)
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (<i>HBB</i>) ACOG ACMG	Methylmalonic Acidemia, cblA Type (<i>MMAA</i>)	Peroxisome Biogenesis Disorder, Type 3 (<i>PEX12</i>)	TGM1-Related Autosomal Recessive Congenital Ichthyosis (<i>TGM1</i>)
Hereditary Fructose Intolerance (<i>ALDOB</i>) ACMG	Methylmalonic Acidemia, cblB Type (<i>MMAB</i>)	Peroxisome Biogenesis Disorder, Type 4 (<i>PEX6</i>)	TPP1-Related Neuronal Ceroid Lipofuscinosis (<i>TPP1</i>)
Junctional Epidermolysis Bullosa, LAMB3-Related (<i>LAMB3</i>)	Methylmalonic Aciduria and Homocystinuria, cblC Type (<i>MMACHC</i>) ACMG	Peroxisome Biogenesis Disorder, Type 5 (<i>PEX2</i>)	Tyrosine Hydroxylase Deficiency (<i>TH</i>)
Hexosaminidase A Deficiency (including Tay-Sachs Disease) (<i>HEXA</i>) ACOG ACMG	MKS1-Related Disorders (<i>MKS1</i>)	Peroxisome Biogenesis Disorder, Type 6 (<i>PEX10</i>)	Tyrosinemia, Type I (<i>FAH</i>) ACMG
HMG-CoA Lyase Deficiency (<i>HMGL</i>)	Mucopolipidosis III Gamma (<i>GNPTG</i>)	Phenylalanine Hydroxylase Deficiency (<i>PAH</i>) ACMG	Tyrosinemia, Type II (<i>TAT</i>)
Holocarboxylase Synthetase Deficiency (<i>HLCS</i>)	Mucopolipidosis IV (<i>MCOLN1</i>) ACMG	POMGNT-Related Disorders (<i>POMGNT1</i>)	USH1C-Related Disorders (<i>USH1C</i>)
Homocystinuria, CBS-Related (<i>CBS</i>) ACMG	Mucopolysaccharidosis, Type I (including Hurler Syndrome) (<i>IDUA</i>) ACMG	Pompe Disease (<i>GAA</i>) ACMG	USH2A-Related Disorders (<i>USH2A</i>) ACMG
Hydroletharus Syndrome (<i>HYLS1</i>)	Mucopolysaccharidosis, Type II (<i>IDS</i>) X-linked	PPT1-Related Neuronal Ceroid Lipofuscinosis (<i>PPT1</i>)	Usher Syndrome, Type 3 (<i>CLRN1</i>) ACMG
Hypophosphatasia (<i>ALPL</i>) ACMG	Mucopolysaccharidosis, Type IIIA (<i>SGSH</i>)	Primary Carnitine Deficiency (<i>SLC22A5</i>)	Very Long Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADVL</i>) ACMG
Isovaleric Acidemia (<i>IVD</i>)	Mucopolysaccharidosis, Type IIIB (<i>NAGLU</i>)	Primary Hyperoxaluria, Type 1 (<i>AGXT</i>) ACMG	Wilson Disease (<i>ATP7B</i>) ACMG
Joubert Syndrome 2 (<i>TMEM216</i>) ACMG	Mucopolysaccharidosis, Type IIIC (<i>HGSNAT</i>)	Primary Hyperoxaluria, Type 2 (<i>GRHPR</i>)	X-linked Adrenal Hypoplasia Congenita (<i>NROB1</i>) ACMG X-linked
Junctional Epidermolysis Bullosa, LAMC2-Related (<i>LAMC2</i>)	MMUT-Related Methylmalonic Acidemia (<i>MMUT</i>) ACMG	Primary Hyperoxaluria, Type 3 (<i>HOGA1</i>)	X-Linked Juvenile Retinoschisis (<i>RS1</i>) ACMG X-linked
Junctional Epidermolysis Bullosa, LAMA3-Related (<i>LAMA3</i>)	MYO7A-Related Disorders (<i>MYO7A</i>)	Pycnodysostosis (<i>CTSK</i>)	X-Linked Myotubular Myopathy (<i>MTM1</i>) X-linked
Familial Hyperinsulinism, KCNJ11-Related (<i>KCNJ11</i>)	NEB-Related Nemaline Myopathy (<i>NEB</i>) ACMG	Pyruvate Carboxylase Deficiency (<i>PC</i>)	X-Linked Severe Combined Immunodeficiency (<i>IL2RG</i>) X-linked
Krabbe Disease (<i>GALC</i>)	Nephrotic Syndrome, NPHS1-Related (<i>NPHS1</i>) ACMG	Rhizomelic Chondrodysplasia Punctata, Type 1 (<i>PEX7</i>)	Xeroderma Pigmentosum, Group A (<i>XPA</i>)
Muscular Dystrophy, LAMA2-Related (<i>LAMA2</i>)	Niemann-Pick Disease, SMPD1-Related (<i>SMPD1</i>) ACMG	RTEL1-Related Disorders (<i>RTEL1</i>)	Xeroderma Pigmentosum, Group C (<i>XPC</i>) ACMG
Leigh Syndrome, French-Canadian Type (<i>LRPPRC</i>)	Niemann-Pick Disease, Type C1 (<i>NPC1</i>)	Salla Disease (<i>SLC17A5</i>)	
Lipoid Congenital Adrenal Hyperplasia (<i>STAR</i>)	Niemann-Pick Disease, Type C2 (<i>NPC2</i>)	Sandhoff Disease (<i>HEXB</i>)	
Lysosomal Acid Lipase Deficiency (<i>LIPA</i>)	Nijmegen Breakage Syndrome (<i>NBN</i>)	Short Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADS</i>)	
Maple Syrup Urine Disease, Type Ia (<i>BCKDHA</i>)	Ornithine Transcarbamylase Deficiency (<i>OTC</i>) ACMG X-linked	Sjogren-Larsson Syndrome (<i>ALDH3A2</i>)	
Maple Syrup Urine Disease, Type Ib (<i>BCKDHB</i>) ACMG	PCCA-Related Propionic Acidemia (<i>PCCA</i>)	SLC26A2-Related Disorders (<i>SLC26A2</i>) ACMG	
		Smith-Lemli-Opitz Syndrome (<i>DHCR7</i>) ACMG	

[ACOG](#) Indicates disease listed in ACOG guidelines

[ACMG](#) Indicates disease listed in ACMG guidelines

[X-linked](#) Indicates X-linked disorders