

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.

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

Glycogen Storage Disease, Type Ib (SLC37A4) ACMG	Maple Syrup Urine Disease, Type II (DBT)	PCCB-Related Propionic Acidemia (PCCB)	Spinal Muscular Atrophy (SMN1) ACOG ACMG
Glycogen Storage Disease, Type III (AGL)	Medium Chain Acyl-CoA Dehydrogenase Deficiency (ACADM) ACMG	PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (PCDH15) ACMG	Spondylothoracic Dysostosis (MESP2)
GNE Myopathy (GNE)	Megalencephalic Leukoencephalopathy with Subcortical Cysts (MLC1) ACMG	Pendred Syndrome (SLC26A4) ACMG	Steroid-Resistant Nephrotic Syndrome (NPHS2)
GNPTAB-Related Disorders (GNPTAB) ACMG	Metachromatic Leukodystrophy (ARSA) ACMG	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 (PEX12)	TPP1-Related Neuronal Ceroid Lipofuscinosis (TPP1)
Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)(HBB) ACOG ACMG	Methylmalonic Acidemia, cblB Type (MMAB)	Peroxisome Biogenesis Disorder, Type 4 (PEX6)	Tyrosine Hydroxylase Deficiency (TH)
Hereditary Fructose Intolerance (ALDOB) ACMG	Methylmalonic Aciduria and Homocystinuria, cblC Type (MMACHC) ACMG	Peroxisome Biogenesis Disorder, Type 5 (PEX2)	Tyrosinemia, Type I (FAH) ACMG
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) ACOG ACMG	Mucopolysaccharidosis, Type I (including Hurler Syndrome) (IDUA) ACMG	Phenylalanine Hydroxylase Deficiency (PAH) ACMG	USH1C-Related Disorders (USH1C)
HMG-CoA Lyase Deficiency (HMGCL)	Mucopolysaccharidosis, Type I (including Hurler Syndrome) (IDUA) ACMG	POMGNT-Related Disorders (POMGNT1)	USH2A-Related Disorders (USH2A) ACMG
Holocarboxylase Synthetase Deficiency (HLCS)	Mucopolysaccharidosis, Type II (IDS) X-linked	Pompe Disease (GAA) ACMG	Usher Syndrome, Type 3 (CLRN1) ACMG
Homocystinuria, CBS-Related (CBS) ACMG	Mucopolysaccharidosis, Type IIIA (SGSH)	PPT1-Related Neuronal Ceroid Lipofuscinosis (PPT1)	Very Long Chain Acyl-CoA Dehydrogenase Deficiency (ACADVL) ACMG
Hydroletharus Syndrome (HYLS1)	Mucopolysaccharidosis, Type IIIB (NAGLU)	Primary Carnitine Deficiency (SLC22A5)	Wilson Disease (ATP7B) ACMG
Hypophosphatasia (ALPL) ACMG	Mucopolysaccharidosis, Type IIIC (HGSNAT)	Primary Hyperoxaluria, Type 1 (AGXT) ACMG	X-linked Adrenal Hypoplasia Congenita (NROB1) ACMG X-linked
Isovaleric Acidemia (IVD)	MMUT-Related Methylmalonic Acidemia (MMUT) ACMG	Primary Hyperoxaluria, Type 2 (GRHPR)	X-Linked Juvenile Retinoschisis (RS1) ACMG X-linked
Joubert Syndrome 2 (TMEM216) ACMG	MYO7A-Related Disorders (MYO7A)	Primary Hyperoxaluria, Type 3 (HOGA1)	X-Linked Myotubular Myopathy (MTM1) X-linked
Junctional Epidermolysis Bullosa, LAMC2-Related (LAMC2)	NEB-Related NemaLine Myopathy (NEB) ACMG	Pycnodysostosis (CTSK)	X-Linked Severe Combined Immunodeficiency (IL2RG) X-linked
Junctional Epidermolysis Bullosa, LAMA3-Related (LAMA3)	Nephrotic Syndrome, NPHS1-Related (NPHS1) ACMG	Pyruvate Carboxylase Deficiency (PC)	Xeroderma Pigmentosum, Group A (XPA)
Familial Hyperinsulinism, KCNJ11-Related (KCNJ11)	Niemann-Pick Disease, SMPD1-Related (SMPD1) ACMG	Rhizomelic Chondrodysplasia Punctata, Type 1 (PEX7)	Xeroderma Pigmentosum, Group C (XPC) ACMG
Krabbe Disease (GALC)	Niemann-Pick Disease, Type C1 (NPC1)	RTEL1-Related Disorders (RTEL1)	
Muscular Dystrophy, LAMA2-Related (LAMA2)	Niemann-Pick Disease, Type C2 (NPC2)	Salla Disease (SLC17A5)	
Leigh Syndrome, French-Canadian Type (LRPPRC)	Nijmegen Breakage Syndrome (NBN)	Sandhoff Disease (HEXB)	ACOG
Lipoid Congenital Adrenal Hyperplasia (STAR)	Ornithine Transcarbamylase Deficiency (OTC) ACMG X-linked	Short Chain Acyl-CoA Dehydrogenase Deficiency (ACADS)	Indicates disease listed in ACOG guidelines
Lysosomal Acid Lipase Deficiency (LIPA)	PCCA-Related Propionic Acidemia (PCCA)	Sjogren-Larsson Syndrome (ALDH3A2)	ACMG
Maple Syrup Urine Disease, Type Ia (BCKDHA)		SLC26A2-Related Disorders (SLC26A2) ACMG	Indicates disease listed in ACMG guidelines
Maple Syrup Urine Disease, Type Ib (BCKDHB) ACMG		Smith-Lemli-Opitz Syndrome (DHCR7) ACMG	X-linked
		Spastic Paraplegia, Type 15 (ZFYVE26)	Indicates X-linked disorders