

Foresight™ Carrier Screen disease list

Using a systematic approach to improve our panel for both patients and providers, Counsyl has updated its Universal Panel — introducing the latest breakthrough in expanded carrier screening.

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (<i>CYP11B1</i>)	Bardet-Biedl Syndrome, BBS10-Related (<i>BBS10</i>)	Congenital Finnish Nephrosis (<i>NPHS1</i>)	Gaucher Disease (<i>GBA</i>) * ACMG
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (<i>CYP21A2</i>)*	Bardet-Biedl Syndrome, BBS12-Related (<i>BBS12</i>)	Costeff Optic Atrophy Syndrome (<i>OPA3</i>)	GJB2-Related DFNB1
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (<i>PTS</i>)	Bardet-Biedl Syndrome, BBS2-Related (<i>BBS2</i>)	Cystic Fibrosis (<i>CFTR</i>) ACOG ACMG	Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) (<i>GJB2</i>)
ABCC8-Related Hyperinsulinism (<i>ABCC8</i>)	Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) (<i>SGCB</i>)	Cystinosis (<i>CTNS</i>)	GLB1-Related Disorders (<i>GLB1</i>)
Adenosine Deaminase Deficiency (<i>ADA</i>)	Biotinidase Deficiency (<i>BTBD</i>)	D-Bifunctional Protein Deficiency (<i>HSD17B4</i>)	GLDC-Related Glycine Encephalopathy (<i>GLDC</i>)
Adrenoleukodystrophy: X-Linked (<i>ABCD1</i>) X-linked	Bloom Syndrome (<i>BLM</i>) ACMG	Delta-Sarcoglycanopathy (<i>SGCD</i>)	Glutaric Acidemia, Type 1 (<i>GCDH</i>)
Alpha Thalassemia (<i>HBA1/HBA2</i>) * ACOG ACMG	Calpainopathy (<i>CAPN3</i>)	Dysferlinopathy (<i>DYSF</i>)	Glycogen Storage Disease, Type Ia (<i>G6PC</i>)
Alpha-Mannosidosis (<i>MAN2B1</i>)	Canavan Disease (<i>ASPA</i>) ACOG ACMG	Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy) (<i>DMD</i>) X-linked	Glycogen Storage Disease, Type Ib (<i>SLC37A4</i>)
Alpha-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2D) (<i>SGCA</i>)	Carbamoylphosphate Synthetase I Deficiency (<i>CPS1</i>)	ERCC6-Related Disorders (<i>ERCC6</i>)	Glycogen Storage Disease, Type III (<i>AGL</i>)
Alport Syndrome, X-Linked (<i>COL4A5</i>) X-linked	Carnitine Palmitoyltransferase IA Deficiency (<i>CPT1A</i>)	ERCC8-Related Disorders (<i>ERCC8</i>)	GNPTAB-Related Disorders (<i>GNPTAB</i>)
Alstrom Syndrome (<i>ALMS1</i>)	Carnitine Palmitoyltransferase II Deficiency (<i>CPT2</i>)	EVC-Related Ellis-Van Creveld Syndrome (<i>EVC</i>)	GRACILE Syndrome (<i>BCS1L</i>)
AMT-Related Glycine Encephalopathy (<i>AMT</i>)	Cartilage-Hair Hypoplasia (<i>RMRP</i>)	EVC2-Related Ellis-Van Creveld Syndrome (<i>EVC2</i>)	HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) (<i>HADHA</i>)
Andermann Syndrome (<i>SLC12A6</i>)	Cerebrotendinous Xanthomatosis (<i>CYP27A1</i>)	Fabry Disease (<i>GLA</i>) X-linked	Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (<i>HBB</i>) ACOG
Argininemia (<i>ARG1</i>)	Citrullinemia, Type 1 (<i>ASS1</i>)	Familial Dysautonomia (<i>IKBKAP</i>) ACOG ACMG	Hereditary Fructose Intolerance (<i>ALDOB</i>)
Argininosuccinic Aciduria (<i>ASL</i>)	CLN3-Related Neuronal Ceroid Lipofuscinosis (<i>CLN3</i>)	Familial Mediterranean Fever (<i>MEFV</i>)	Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related (<i>LAMA3</i>)
ARSACS (<i>SACS</i>)	CLN5-Related Neuronal Ceroid Lipofuscinosis (<i>CLN5</i>)	Fanconi Anemia Complementation, Group A (<i>FANCA</i>)	Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related (<i>LAMB3</i>)
Aspartylglycosaminuria (<i>AGA</i>)	CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 (<i>CLN6</i>)	Fanconi Anemia, Type C (<i>FANCC</i>) ACMG	Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (<i>LAMC2</i>)
Ataxia with Vitamin E Deficiency (<i>TTPA</i>)	Cohen Syndrome (<i>VPS13B</i>)	FKRP-Related Disorders (<i>FKRP</i>)	Hexosaminidase A Deficiency (including Tay-Sachs Disease) (<i>HEXA</i>) ACOG ACMG
Ataxia-Telangiectasia (<i>ATM</i>)	COL4A3-Related Alport Syndrome (<i>COL4A3</i>)	FKTN-Related Disorders (including Walker-Warburg Syndrome) (<i>FKTN</i>)	HMG-CoA Lyase Deficiency (<i>HMGCL</i>)
ATP7A-Related Disorders (<i>ATP7A</i>) X-linked	COL4A4-Related Alport Syndrome (<i>COL4A4</i>)	Fragile X Syndrome (<i>FMR1</i>) * X-linked	
Autosomal Recessive Osteopetrosis, Type 1 (<i>TCIRG1</i>)	Congenital Disorder of Glycosylation, Type Ia (<i>PMM2</i>)	Galactokinase Deficiency (<i>GALK1</i>)	
Bardet-Biedl Syndrome, BBS1-Related (<i>BBS1</i>)	Congenital Disorder of Glycosylation, Type Ib (<i>MPI</i>)	Galactosemia (<i>GALT</i>)	
	Congenital Disorder of Glycosylation, Type Ic (<i>ALG6</i>)	Gamma-Sarcoglycanopathy (<i>SGCG</i>)	

Holocarboxylase Synthetase Deficiency (<i>HLCS</i>)	Mucopolipidosis IV (<i>MCOLN1</i>) ACMG	PEX1-Related Zellweger Syndrome Spectrum (<i>PEX1</i>)	Sulfate Transporter-Related Osteochondrodysplasia (<i>SLC26A2</i>)
Homocystinuria caused by Cystathionine Beta-Synthase Deficiency (<i>CBS</i>)	Mucopolysaccharidosis, Type I (including Hurler Syndrome) (<i>IDUA</i>)	Phenylalanine Hydroxylase Deficiency (<i>PAH</i>)	TGM1-Related Autosomal Recessive Congenital Ichthyosis (<i>TGM1</i>)
Hydroletharus Syndrome (<i>HYLS1</i>)	Mucopolysaccharidosis, Type II (<i>IDS</i>) X-linked	PKHD1-Related Autosomal Recessive Polycystic Kidney Disease (<i>PKHD1</i>)	TPP1-Related Neuronal Ceroid Lipofuscinosis (<i>TPP1</i>)
Hypophosphatasia, Autosomal Recessive (<i>ALPL</i>)	Mucopolysaccharidosis, Type IIIA (<i>SGSH</i>)	Polyglandular Autoimmune Syndrome, Type 1 (<i>AIRE</i>)	Tyrosinemia, Type I (<i>FAH</i>)
Inclusion Body Myopathy 2 (<i>GNE</i>)	Mucopolysaccharidosis, Type IIIB (<i>NAGLU</i>)	Pompe Disease (<i>GAA</i>)	Tyrosinemia, Type II (<i>TAT</i>)
Isovaleric Acidemia (<i>IVD</i>)	Mucopolysaccharidosis, Type IIIC (<i>HGSNAT</i>)	PPT1-Related Neuronal Ceroid Lipofuscinosis (<i>PPT1</i>)	USH1C-Related Disorders (<i>USH1C</i>)
Joubert Syndrome 2 (<i>TMEM216</i>)	Muscle-Eye-Brain Disease (<i>POMGNT1</i>)	Primary Carnitine Deficiency (<i>SLC22A5</i>)	USH2A-Related Disorders (<i>USH2A</i>)
KCNJ11-Related Familial Hyperinsulinism (<i>KCNJ11</i>)	MUT-Related Methylmalonic Acidemia (<i>MUT</i>)	Primary Hyperoxaluria, Type 1 (<i>AGXT</i>)	Usher Syndrome, Type 3 (<i>CLRN1</i>)
Krabbe Disease (<i>GALC</i>)	MYO7A-Related Disorders (<i>MYO7A</i>)	Primary Hyperoxaluria, Type 2 (<i>GRHPR</i>)	Very Long Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADVL</i>)
LAMA2-Related Muscular Dystrophy (<i>LAMA2</i>)	NEB-Related Nemaline Myopathy (<i>NEB</i>)	Primary Hyperoxaluria, Type 3 (<i>HOGA1</i>)	Wilson Disease (<i>ATP7B</i>)
Leigh Syndrome, French-Canadian Type (<i>LRPPRC</i>)	Niemann-Pick Disease, Type C (<i>NPC1</i>)	PROP1-Related Combined Pituitary Hormone Deficiency (<i>PROPT</i>)	X-Linked Congenital Adrenal Hypoplasia (<i>NROB1</i>) X-linked
Lipoamide Dehydrogenase Deficiency (<i>DLD</i>)	Niemann-Pick Disease, Type C2 (<i>NPC2</i>)	Pycnodysostosis (<i>CTSK</i>)	X-Linked Juvenile Retinoschisis (<i>RS1</i>) X-linked
Lipoid Congenital Adrenal Hyperplasia (<i>STAR</i>)	Niemann-Pick Disease, SMPD1-Associated (<i>SMPD1</i>) ACMG	Pyruvate Carboxylase Deficiency (<i>PC</i>)	X-Linked Myotubular Myopathy (<i>MTM1</i>) X-linked
Lysosomal Acid Lipase Deficiency (<i>LIPA</i>)	Nijmegen Breakage Syndrome (<i>NBN</i>)	Rhizomelic Chondrodysplasia Punctata, Type 1 (<i>PEX7</i>)	X-Linked Severe Combined Immunodeficiency (<i>IL2RG</i>) X-linked
Maple Syrup Urine Disease, Type Ia (<i>BCKDHA</i>)	Northern Epilepsy (<i>CLN8</i>)	RTEL1-Related Disorders (<i>RTEL1</i>)	Xeroderma Pigmentosum, Group A (<i>XPA</i>)
Maple Syrup Urine Disease, Type IB (<i>BCKDHB</i>)	Ornithine Transcarbamylase Deficiency (<i>OTC</i>) X-linked	Salla Disease (<i>SLC17A5</i>)	Xeroderma Pigmentosum, Group C (<i>XPC</i>)
Maple Syrup Urine Disease, Type II (<i>DBT</i>)	PCCA-Related Propionic Acidemia (<i>PCCA</i>)	Sandhoff Disease (<i>HEXB</i>)	
Medium Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADM</i>)	PCCB-Related Propionic Acidemia (<i>PCCB</i>)	Segawa Syndrome (<i>TH</i>)	
Megalencephalic Leukoencephalopathy with Subcortical Cysts (<i>MLC1</i>)	PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (<i>PCDH15</i>)	Short Chain Acyl-CoA Dehydrogenase Deficiency (<i>ACADS</i>)	
Metachromatic Leukodystrophy (<i>ARSA</i>)	Pendred Syndrome (<i>SLC26A4</i>)	Sjogren-Larsson Syndrome (<i>ALDH3A2</i>)	
Methylmalonic Acidemia, cblA Type (<i>MMAA</i>)	Peroxisome Biogenesis Disorder, Type 3 (<i>PEX12</i>)	Smith-Lemli-Opitz Syndrome (<i>DHCR7</i>)	ACOG Indicates testing recommended by ACOG
Methylmalonic Acidemia, cblB Type (<i>MMAB</i>)	Peroxisome Biogenesis Disorder, Type 4 (<i>PEX6</i>)	Spastic Paraplegia, Type 15 (<i>ZFYVE26</i>)	ACMG Indicates testing recommended by ACMG
Methylmalonic Aciduria and Homocystinuria, cblC Type (<i>MMACHC</i>)	Peroxisome Biogenesis Disorder, Type 5 (<i>PEX2</i>)	Spinal Muscular Atrophy (<i>SMN1</i>)* ACOG ACMG	X-linked Indicates X-linked disorders
MKS1-Related Disorders (<i>MKS1</i>)	Peroxisome Biogenesis Disorder, Type 6 (<i>PEX10</i>)	Steroid-Resistant Nephrotic Syndrome (<i>NPHS2</i>)	
Mucopolipidosis III Gamma (<i>GNPTG</i>)			

*Analyzed using targeted genotyping