

# COUNSYL FORESIGHT™ CARRIER SCREEN DISEASE LIST

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

11-Beta-Hydroxylase-Deficient Congenital Adrenal Hyperplasia ( <i>CYP11B1</i> )	Bardet-Biedl Syndrome, BBS1-Related ( <i>BBS1</i> )	Congenital Disorder of Glycosylation, Type Ic ( <i>ALG6</i> )	Galactokinase Deficiency ( <i>GALK1</i> )
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia ( <i>CYP21A2</i> )*	Bardet-Biedl Syndrome, BBS10-Related ( <i>BBS10</i> )	Congenital Finnish Nephrosis ( <i>NPFS1</i> )	Galactosemia ( <i>GALT</i> )
6-Pyruvoyl-Tetrahydropterin Synthase Deficiency ( <i>PTS</i> )	Bardet-Biedl Syndrome, BBS12-Related ( <i>BBS12</i> )	Costeff Optic Atrophy Syndrome ( <i>OPA3</i> )	Gamma-Sarcoglycanopathy ( <i>SGCG</i> )
ABCC8-Related Hyperinsulinism ( <i>ABCC8</i> )	Bardet-Biedl Syndrome, BBS2-Related ( <i>BBS2</i> )	Cystic Fibrosis ( <i>CFTR</i> ) <a href="#">ACOG</a> <a href="#">ACMG</a>	Gaucher Disease ( <i>GBA</i> )* <a href="#">ACMG</a>
Adenosine Deaminase Deficiency ( <i>ADA</i> )	Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) ( <i>SGCB</i> )	Cystinosis ( <i>CTNS</i> )	GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) ( <i>GJB2</i> )
Adrenoleukodystrophy: X-Linked ( <i>ABCD1</i> ) <a href="#">X-linked</a>	Biotinidase Deficiency ( <i>BTD</i> )	D-Bifunctional Protein Deficiency ( <i>HSD17B4</i> )	GLB1-Related Disorders ( <i>GLB1</i> )
Alpha Thalassemia ( <i>HBA1/HBA2</i> )* <a href="#">ACOG</a> <a href="#">ACMG</a>	Bloom Syndrome ( <i>BLM</i> ) <a href="#">ACMG</a>	Delta-Sarcoglycanopathy ( <i>SGCD</i> )	GLDC-Related Glycine Encephalopathy ( <i>GLDC</i> )
Alpha-Mannosidosis ( <i>MAN2B1</i> )	Calpainopathy ( <i>CAPN3</i> )	Dysferlinopathy ( <i>DYSF</i> )	Glutaric Acidemia, Type 1 ( <i>GCDH</i> )
Alpha-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2D) ( <i>SGCA</i> )	Canavan Disease ( <i>ASPA</i> ) <a href="#">ACOG</a> <a href="#">ACMG</a>	Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy)( <i>DMD</i> ) <a href="#">X-linked</a>	Glycogen Storage Disease, Type Ia ( <i>G6PC</i> )
Alport Syndrome, X-Linked ( <i>COL4A5</i> ) <a href="#">X-linked</a>	Carbamoylphosphate Synthetase I Deficiency ( <i>CPS1</i> )	ERCC6-Related Disorders ( <i>ERCC6</i> )	Glycogen Storage Disease, Type Ib ( <i>SLC37A4</i> )
Alstrom Syndrome ( <i>ALMS1</i> )	Carnitine Palmitoyltransferase IA Deficiency ( <i>CPT1A</i> )	ERCC8-Related Disorders ( <i>ERCC8</i> )	Glycogen Storage Disease, Type III ( <i>AGL</i> )
AMT-Related Glycine Encephalopathy ( <i>AMT</i> )	Carnitine Palmitoyltransferase II Deficiency ( <i>CPT2</i> )	EVC-Related Ellis-Van Creveld Syndrome ( <i>EVC</i> )	GNPTAB-Related Disorders ( <i>GNPTAB</i> )
Andermann Syndrome ( <i>SLC12A6</i> )	Cartilage-Hair Hypoplasia ( <i>RMRP</i> )	EVC2-Related Ellis-Van Creveld Syndrome ( <i>EVC2</i> )	GRACILE Syndrome ( <i>BCS1L</i> )
Argininemia ( <i>ARG1</i> )	Cerebrotendinous Xanthomatosis ( <i>CYP27A1</i> )	Fabry Disease ( <i>GLA</i> ) <a href="#">X-linked</a>	HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) ( <i>HADHA</i> )
Argininosuccinic Aciduria ( <i>ASL</i> )	Citrullinemia, Type 1 ( <i>ASS1</i> )	Familial Dysautonomia ( <i>IKBKAP</i> ) <a href="#">ACOG</a> <a href="#">ACMG</a>	Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)( <i>HBB</i> ) <a href="#">ACOG</a>
ARSACS ( <i>SACS</i> )	CLN3-Related Neuronal Ceroid Lipofuscinosis ( <i>CLN3</i> )	Familial Mediterranean Fever ( <i>MEFV</i> )	Hereditary Fructose Intolerance ( <i>ALDOB</i> )
Aspartylglycosaminuria ( <i>AGA</i> )	CLN5-Related Neuronal Ceroid Lipofuscinosis ( <i>CLN5</i> )	Fanconi Anemia Complementation, Group A ( <i>FANCA</i> )	Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related ( <i>LAMA3</i> )
Ataxia with Vitamin E Deficiency ( <i>TTPA</i> )	CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 ( <i>CLN6</i> )	Fanconi Anemia, Type C ( <i>FANCC</i> ) <a href="#">ACMG</a>	Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related ( <i>LAMB3</i> )
Ataxia-Telangiectasia ( <i>ATM</i> )	Cohen Syndrome ( <i>VPS13B</i> )	FKRP-Related Disorders ( <i>FKRP</i> )	Herlitz Junctional Epidermolysis Bullosa, LAMC2-related ( <i>LAMC2</i> )
ATP7A-Related Disorders ( <i>ATP7A</i> ) <a href="#">X-linked</a>	COL4A3-Related Alport Syndrome ( <i>COL4A3</i> )	FKTN-Related Disorders (including Walker-Warburg Syndrome) ( <i>FKTN</i> )	
Autosomal Recessive Osteopetrosis, Type 1 ( <i>TCIRG1</i> )	COL4A4-Related Alport Syndrome ( <i>COL4A4</i> )	Fragile X Syndrome ( <i>FMR1</i> )* <a href="#">X-linked</a>	
	Congenital Disorder of Glycosylation, Type Ia ( <i>PMM2</i> )		
	Congenital Disorder of Glycosylation, Type Ib ( <i>MPL</i> )		

Hexosaminidase A Deficiency (including Tay-Sachs Disease) ( <i>HEXA</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACOG</span> <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACMG</span>	Methylmalonic Aciduria and Homocystinuria, cblC Type ( <i>MMACHC</i> )	Peroxisome Biogenesis Disorder, Type 4 ( <i>PEX6</i> )	Spondylothoracic Dysostosis ( <i>MESP2</i> )
HMG-CoA Lyase Deficiency ( <i>HMGCL</i> )	MKS1-Related Disorders ( <i>MKS1</i> )	Peroxisome Biogenesis Disorder, Type 5 ( <i>PEX2</i> )	Steroid-Resistant Nephrotic Syndrome ( <i>NPHS2</i> )
Holocarboxylase Synthetase Deficiency ( <i>HLCS</i> )	Mucopolipidosis III Gamma ( <i>GNPTG</i> )	Peroxisome Biogenesis Disorder, Type 6 ( <i>PEX10</i> )	Sulfate Transporter-Related Osteochondrodysplasia ( <i>SLC26A2</i> )
Homocystinuria caused by Cystathionine Beta-Synthase Deficiency ( <i>CBS</i> )	Mucopolipidosis IV ( <i>MCOLN1</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACMG</span>	PEX1-Related Zellweger Syndrome Spectrum ( <i>PEX1</i> )	TGM1-Related Autosomal Recessive Congenital Ichthyosis ( <i>TGM1</i> )
Hydrolethalus Syndrome ( <i>HYLS1</i> )	Mucopolysaccharidosis, Type I (including Hurler Syndrome) ( <i>IDUA</i> )	Phenylalanine Hydroxylase Deficiency ( <i>PAH</i> )	TPP1-Related Neuronal Ceroid Lipofuscinosis ( <i>TPP1</i> )
Hypophosphatasia, Autosomal Recessive ( <i>ALPL</i> )	Mucopolysaccharidosis, Type II ( <i>IDS</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">X-linked</span>	PKHD1-Related Autosomal Recessive Polycystic Kidney Disease ( <i>PKHD1</i> )	Tyrosinemia, Type I ( <i>FAH</i> )
Inclusion Body Myopathy 2 ( <i>GNE</i> )	Mucopolysaccharidosis, Type IIIA ( <i>SGSH</i> )	Polyglandular Autoimmune Syndrome, Type 1 ( <i>AIRE</i> )	Tyrosinemia, Type II ( <i>TAT</i> )
Isovaleric Acidemia ( <i>IVD</i> )	Mucopolysaccharidosis, Type IIIB ( <i>NAGLU</i> )	Pompe Disease ( <i>GAA</i> )	USH1C-Related Disorders ( <i>USH1C</i> )
Joubert Syndrome 2 ( <i>TMEM216</i> )	Mucopolysaccharidosis, Type IIIC ( <i>HGSNAT</i> )	PPT1-Related Neuronal Ceroid Lipofuscinosis ( <i>PPT1</i> )	USH2A-Related Disorders ( <i>USH2A</i> )
KCNJ11-Related Familial Hyperinsulinism ( <i>KCNJ11</i> )	Muscle-Eye-Brain Disease ( <i>POMGNT1</i> )	Primary Carnitine Deficiency ( <i>SLC22A5</i> )	Usher Syndrome, Type 3 ( <i>CLRN1</i> )
Krabbe Disease ( <i>GALC</i> )	MUT-Related Methylmalonic Acidemia ( <i>MUT</i> )	Primary Hyperoxaluria, Type 1 ( <i>AGXT</i> )	Very Long Chain Acyl-CoA Dehydrogenase Deficiency ( <i>ACADVL</i> )
LAMA2-Related Muscular Dystrophy ( <i>LAMA2</i> )	MYO7A-Related Disorders ( <i>MYO7A</i> )	Primary Hyperoxaluria, Type 2 ( <i>GRHPR</i> )	Wilson Disease ( <i>ATP7B</i> )
Leigh Syndrome, French-Canadian Type ( <i>LRPPRC</i> )	NEB-Related Nemaline Myopathy ( <i>NEB</i> )	Primary Hyperoxaluria, Type 3 ( <i>HOGA1</i> )	X-Linked Congenital Adrenal Hypoplasia ( <i>NROB1</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">X-linked</span>
Lipoamide Dehydrogenase Deficiency ( <i>DLD</i> )	Niemann-Pick Disease, Type C ( <i>NPC1</i> )	PROP1-Related Combined Pituitary Hormone Deficiency ( <i>PROP1</i> )	X-Linked Juvenile Retinoschisis ( <i>RS1</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">X-linked</span>
Lipoid Congenital Adrenal Hyperplasia ( <i>STAR</i> )	Niemann-Pick Disease, Type C2 ( <i>NPC2</i> )	Pycnodysostosis ( <i>CTSK</i> )	X-Linked Myotubular Myopathy ( <i>MTM1</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">X-linked</span>
Lysosomal Acid Lipase Deficiency ( <i>LIPA</i> )	Niemann-Pick Disease, SMPD1-Associated ( <i>SMPD1</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACMG</span>	Pyruvate Carboxylase Deficiency ( <i>PC</i> )	X-Linked Severe Combined Immunodeficiency ( <i>IL2RG</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">X-linked</span>
Maple Syrup Urine Disease, Type Ia ( <i>BCKDHA</i> )	Nijmegen Breakage Syndrome ( <i>NBN</i> )	Rhizomelic Chondrodysplasia Punctata, Type 1 ( <i>PEX7</i> )	Xeroderma Pigmentosum, Group A ( <i>XPA</i> )
Maple Syrup Urine Disease, Type IB ( <i>BCKDHB</i> )	Northern Epilepsy ( <i>CLN8</i> )	RTEL1-Related Disorders ( <i>RTEL1</i> )	Xeroderma Pigmentosum, Group C ( <i>XPC</i> )
Maple Syrup Urine Disease, Type II ( <i>DBT</i> )	Ornithine Transcarbamylase Deficiency ( <i>OTC</i> ) <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">X-linked</span>	Salla Disease ( <i>SLC17A5</i> )	
Medium Chain Acyl-CoA Dehydrogenase Deficiency ( <i>ACADM</i> )	PCCA-Related Propionic Acidemia ( <i>PCCA</i> )	Sandhoff Disease ( <i>HEXB</i> )	
Megalencephalic Leukoencephalopathy with Subcortical Cysts ( <i>MLC1</i> )	PCCB-Related Propionic Acidemia ( <i>PCCB</i> )	Segawa Syndrome ( <i>TH</i> )	
Metachromatic Leukodystrophy ( <i>ARSA</i> )	PCDH15-Related Disorders (including Usher Syndrome, Type 1F) ( <i>PCDH15</i> )	Short Chain Acyl-CoA Dehydrogenase Deficiency ( <i>ACADS</i> )	<span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACOG</span> Indicates testing recommended by ACOG
Methylmalonic Acidemia, cblA Type ( <i>MMAA</i> )	Pendred Syndrome ( <i>SLC26A4</i> )	Sjogren-Larsson Syndrome ( <i>ALDH3A2</i> )	<span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACMG</span> Indicates testing recommended by ACMG
Methylmalonic Acidemia, cblB Type ( <i>MMAB</i> )	Peroxisome Biogenesis Disorder, Type 3 ( <i>PEX12</i> )	Smith-Lemli-Opitz Syndrome ( <i>DHCR7</i> )	<span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">X-linked</span> Indicates X-linked disorders
		Spastic Paraplegia, Type 15 ( <i>ZFYVE26</i> )	
		Spinal Muscular Atrophy ( <i>SMN1</i> )* <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACOG</span> <span style="background-color: #c6e0b4; border: 1px solid black; padding: 0 2px;">ACMG</span>	*Analysed using custom assay

For information on this test or to request a referral form, please call customer care (9:00am – 5:00pm)

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