

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 (*CYP11B1*)

21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (*CYP21A2*)*

6-Pyruvoyl-Tetrahydropterin Synthase Deficiency (*PTS*)

ABCC8-Related Hyperinsulinism (*ABCC8*)

Adenosine Deaminase Deficiency (*ADA*)

Adrenoleukodystrophy: X-Linked (*ABCD1*) [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*)

AMT-Related Glycine Encephalopathy (*AMT*)

Andermann Syndrome (*SLC12A6*)

Argininemia (*ARG1*)

Argininosuccinic Aciduria (*ASL*)

ARSACS (*SACS*)

Aspartylglycosaminuria (*AGA*)

Ataxia with Vitamin E Deficiency (*TTPA*)

Ataxia-Telangiectasia (*ATM*)

ATP7A-Related Disorders (*ATP7A*) [X-linked](#)

Autosomal Recessive Osteopetrosis, Type 1 (*TCIRG1*)

Bardet-Biedl Syndrome, BBS1-Related (*BBS1*)

Bardet-Biedl Syndrome, BBS10-Related (*BBS10*)

Bardet-Biedl Syndrome, BBS12-Related (*BBS12*)

Bardet-Biedl Syndrome, BBS2-Related (*BBS2*)

Beta-Sarcoglycanopathy (including Limb-Girdle Muscular Dystrophy, Type 2E) (*SGCB*)

Biotinidase Deficiency (*BTD*)

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*)

Cartilage-Hair Hypoplasia (*RMRP*)

Cerebrotendinous Xanthomatosis (*CYP27A1*)

Citrullinemia, Type 1 (*ASS1*)

CLN3-Related Neuronal Ceroid Lipofuscinosis (*CLN3*)

CLN5-Related Neuronal Ceroid Lipofuscinosis (*CLN5*)

CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 (*CLN6*)

Cohen Syndrome (*VPS13B*)

COL4A3-Related Alport Syndrome (*COL4A3*)

COL4A4-Related Alport Syndrome (*COL4A4*)

Congenital Disorder of Glycosylation, Type Ia (*PMM2*)

Congenital Disorder of Glycosylation, Type Ib (*MPI*)

Congenital Disorder of Glycosylation, Type Ic (*ALG6*)

Congenital Finnish Nephrosis (*NPHS1*)

Costeff Optic Atrophy Syndrome (*OPA3*)

Cystic Fibrosis (*CFTR*) [ACOG](#) [ACMG](#)

Cystinosis (*CTNS*)

D-Bifunctional Protein Deficiency (*HSD17B4*)

Delta-Sarcoglycanopathy (*SGCD*)

Dysferlinopathy (*DYSF*)

Dystrophinopathies (including Duchenne/Becker Muscular Dystrophy)(*DMD*) [X-linked](#)

ERCC6-Related Disorders (*ERCC6*)

ERCC8-Related Disorders (*ERCC8*)

EVC-Related Ellis-Van Creveld Syndrome (*EVC*)

EVC2-Related Ellis-Van Creveld Syndrome (*EVC2*)

Fabry Disease (*GLA*) [X-linked](#)

Familial Dysautonomia (*IKBKAP*) [ACOG](#) [ACMG](#)

Familial Mediterranean Fever (*MEFV*)

Fanconi Anemia Complementation, Group A (*FANCA*)

Fanconi Anemia, Type C (*FANCC*) [ACMG](#)

FKRP-Related Disorders (*FKRP*)

FKTN-Related Disorders (including Walker-Warburg Syndrome) (*FKTN*)

Fragile X Syndrome (*FMR1*)* [X-linked](#)

Galactokinase Deficiency (*GALK1*)

Galactosemia (*GALT*)

Gamma-Sarcoglycanopathy (*SGCG*)

Gaucher Disease (*GBA*)* [ACMG](#)

GJB2-Related DFNB1 Nonsyndromic Hearing Loss and Deafness (including two GJB6 deletions) (*GJB2*)

GLB1-Related Disorders (*GLB1*)

GLDC-Related Glycine Encephalopathy (*GLDC*)

Glutaric Acidemia, Type 1 (*GCDH*)

Glycogen Storage Disease, Type Ia (*G6PC*)

Glycogen Storage Disease, Type Ib (*SLC37A4*)

Glycogen Storage Disease, Type III (*AGL*)

GNPTAB-Related Disorders (*GNPTAB*)

GRACILE Syndrome (*BCS1L*)

HADHA-Related Disorders (including Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency) (*HADHA*)

Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease)(*HBB*) [ACOG](#)

Hereditary Fructose Intolerance (*ALDOB*)

Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related (*LAMA3*)

Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related (*LAMB3*)

Herlitz Junctional Epidermolysis Bullosa, LAMC2-related (*LAMC2*)

Hexosaminidase A Deficiency (including Tay-Sachs Disease) (<i>HEXA</i>) ACOG ACMG	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>)	Mucopolisaccharidosis 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>)	Mucopolisaccharidosis IV (<i>MCOLN1</i>) ACMG	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>) X-linked	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 Nematine Myopathy (<i>NEB</i>)	Primary Hyperoxaluria, Type 3 (<i>HOGA1</i>)	X-Linked Congenital Adrenal Hypoplasia (<i>NROB1</i>) X-linked
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>) X-linked
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>) X-linked
Lysosomal Acid Lipase Deficiency (<i>LIPA</i>)	Niemann-Pick Disease, SMPD1-Associated (<i>SMPD1</i>) ACMG	Pyruvate Carboxylase Deficiency (<i>PC</i>)	X-Linked Severe Combined Immunodeficiency (<i>IL2RG</i>) X-linked
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>) X-linked	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>)	ACOG Indicates testing recommended by ACOG
Methylmalonic Acidemia, cblA Type (<i>MMAA</i>)	Pendred Syndrome (<i>SLC26A4</i>)	Sjogren-Larsson Syndrome (<i>ALDH3A2</i>)	ACMG 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>)	X-linked Indicates X-linked disorders
		Spastic Paraplegia, Type 15 (<i>ZFYVE26</i>)	
		Spinal Muscular Atrophy (<i>SMN1</i>)* ACOG ACMG	*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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