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) (*HEXA*) ACOG ACMG

HMG-CoA Lyase Deficiency (*HMGCL*)

Holocarboxylase Synthetase Deficiency (*HLCS*)

Homocystinuria caused by Cystathionine Beta-Synthase Deficiency (*CBS*)

Hydrolethalus Syndrome (*HYLS1*)

Hypophosphatasia, Autosomal Recessive (*ALPL*)

Inclusion Body Myopathy 2 (*GNE*)

Isovaleric Acidemia (*IVD*)

Joubert Syndrome 2 (*TMEM216*)

KCNJ11-Related Familial Hyperinsulinism (*KCNJ11*)

Krabbe Disease (*GALC*)

LAMA2-Related Muscular Dystrophy (*LAMA2*)

Leigh Syndrome, French-Canadian Type (*LRPPRC*)

Lipoamide Dehydrogenase Deficiency (*DLD*)

Lipoid Congenital Adrenal Hyperplasia (*STAR*)

Lysosomal Acid Lipase Deficiency (*LIPA*)

Maple Syrup Urine Disease, Type Ia (*BCKDHA*)

Maple Syrup Urine Disease, Type IB (*BCKDHB*)

Maple Syrup Urine Disease, Type II (*DBT*)

Medium Chain Acyl-CoA Dehydrogenase Deficiency (*ACADM*)

Megalencephalic Leukoencephalopathy with Subcortical Cysts (*MLC1*)

Metachromatic Leukodystrophy (*ARSA*)

Methylmalonic Acidemia, cblA Type (*MMAA*)

Methylmalonic Acidemia, cblB Type (*MMAB*)

Methylmalonic Aciduria and Homocystinuria, cblC Type (*MMACHC*)

MKS1-Related Disorders (*MKS1*)

Mucolipidosis III Gamma (*GNPTG*)

Mucolipidosis IV (*MCOLN1*) ACMG

Mucopolysaccharidosis, Type I (including Hurler Syndrome) (*IDUA*)

Mucopolysaccharidosis, Type II (*IDS*) X-linked

Mucopolysaccharidosis, Type IIIA (*SGSH*)

Mucopolysaccharidosis, Type IIIB (*NAGLU*)

Mucopolysaccharidosis, Type IIIC (*HGSNAT*)

Muscle-Eye-Brain Disease (*POMGNT1*)

MUT-Related Methylmalonic Acidemia (*MUT*)

MYO7A-Related Disorders (*MYO7A*)

NEB-Related Nemaline Myopathy (*NEB*)

Niemann-Pick Disease, Type C (*NPC1*)

Niemann-Pick Disease, Type C2 (*NPC2*)

Niemann-Pick Disease, SMPD1-Associated (*SMPD1*) ACMG

Nijmegen Breakage Syndrome (*NBN*)

Northern Epilepsy (*CLN8*)

Ornithine Transcarbamylase Deficiency (*OTC*) X-linked

PCCA-Related Propionic Acidemia (*PCCA*)

PCCB-Related Propionic Acidemia (*PCCB*)

PCDH15-Related Disorders (including Usher Syndrome, Type 1F) (*PCDH15*)

Pendred Syndrome (*SLC26A4*)

Peroxisome Biogenesis Disorder, Type 3 (*PEX12*)

Peroxisome Biogenesis Disorder, Type 4 (*PEX6*)

Peroxisome Biogenesis Disorder, Type 5 (*PEX2*)

Peroxisome Biogenesis Disorder, Type 6 (*PEX10*)

PEX1-Related Zellweger Syndrome Spectrum (*PEX1*)

Phenylalanine Hydroxylase Deficiency (*PAH*)

PKHD1-Related Autosomal Recessive Polycystic Kidney Disease (*PKHD1*)

Polyglandular Autoimmune Syndrome, Type 1 (*AIRE*)

Pompe Disease (*GAA*)

PPT1-Related Neuronal Ceroid Lipofuscinosis (*PPT1*)

Primary Carnitine Deficiency (*SLC22A5*)

Primary Hyperoxaluria, Type 1 (*AGXT*)

Primary Hyperoxaluria, Type 2 (*GRHPR*)

Primary Hyperoxaluria, Type 3 (*HOGA1*)

PROP1-Related Combined Pituitary Hormone Deficiency (*PROP1*)

Pycnodysostosis (*CTSK*)

Pyruvate Carboxylase Deficiency (*PC*)

Rhizomelic Chondrodysplasia Punctata, Type 1 (*PEX7*)

RTEL1-Related Disorders (*RTEL1*)

Salla Disease (*SLC17A5*)

Sandhoff Disease (*HEXB*)

Segawa Syndrome (*TH*)

Short Chain Acyl-CoA Dehydrogenase Deficiency (*ACADS*)

Sjogren-Larsson Syndrome (*ALDH3A2*)

Smith-Lemli-Opitz Syndrome (*DHCR7*)

Spastic Paraplegia, Type 15 (*ZFYVE26*)

Spinal Muscular Atrophy (*SMN1*)* ACOG ACMG

Spondylothoracic Dysostosis (*MESP2*)

Steroid-Resistant Nephrotic Syndrome (*NPHS2*)

Sulfate Transporter-Related Osteochondrodysplasia (*SLC26A2*)

TGM1-Related Autosomal Recessive Congenital Ichthyosis (*TGM1*)

TPP1-Related Neuronal Ceroid Lipofuscinosis (*TPP1*)

Tyrosinemia, Type I (*FAH*)

Tyrosinemia, Type II (*TAT*)

USH1C-Related Disorders (*USH1C*)

USH2A-Related Disorders (*USH2A*)

Usher Syndrome, Type 3 (*CLRN1*)

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (*ACADVL*)

Wilson Disease (*ATP7B*)

X-Linked Congenital Adrenal Hypoplasia (*NR0B1*) X-linked

X-Linked Juvenile Retinoschisis (*RS1*) X-linked

X-Linked Myotubular Myopathy (*MTM1*) X-linked

X-Linked Severe Combined Immunodeficiency (*IL2RG*) X-linked

Xeroderma Pigmentosum, Group A (*XPA*)

Xeroderma Pigmentosum, Group C (*XPC*)

ACOG
Indicates testing recommended by ACOG

ACMG
Indicates testing recommended by ACMG

X-linked
Indicates X-linked disorders

*Analysed using custom assay

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

1800 822 999

or email us at info@genomicdiagnostics.com.au

www.genomicdiagnostics.com.au

Phone: 1800 822 999 | **Email:** info@genomicdiagnostics.com.au | **URL:** genomicdiagnostics.com.au

Specialist Diagnostic Services, ABN 84 007 190 043 APA No. 000042, trading as Laverty Pathology. IS-MKT-119. V1 July 2019