ARUP LABORATORIES

Expanded Screening Panels

The following two panels are listed with diseases and the number of mutations assayed.

ACOG = Testing for this disease recommended to be offered by ACOG
ACMG = Testing for this disease recommended to be offered by ACMG
*Disease must be specifically requested to be included in your panel.

Prenatal Panel

ABCC8-Related Hyperinsulinism (3)
Alpha-Mannosidosis (1)
Ataxia-Telangiectasia (8)
Autosomal Recessive Polycystic Kidney Disease (4)
Bardet-Biedl Syndrome, BBS10-Related (1)
Bardet-Biedl Syndrome, BBS1-Related (1)
Biotinidase Deficiency (4)
Bloom Syndrome (1) ACMG
Canavan Disease (4) ACOG ACMG
Carnitine Palmitoyltransferase IA Deficiency (1)
Carnitine Palmitoyltransferase II Deficiency (3)
Cartilage-Hair Hypoplasia (1)
Citrullinemia Type 1 (2)
CLN3-Related and CLN5-Related Neuronal Ceroid Lipofuscinosis (2)
Cohen Syndrome (1)
Congenital Disorder of Glycosylation Type Ia and Ib (5)
Congenital Finnish Nephrosis (2)
Costeff Optic Atrophy Syndrome (1)
Cystic Fibrosis (100) ACOG ACMG
D-Bifunctional Protein Deficiency (2)
Familial Dysautonomia (2) ACOG ACMG
Familial Mediterranean Fever (5)
Fanconi Anemia Type C (3) ACMG
*Fragile X Syndrome (Female blood specimens only) (1)
Galactosemia (10)
Gaucher Disease (10) ACMG
GJB2-Related DFNB 1 Nonsyndromic Hearing Loss And Deafness (7)
Glutaric Acidemia Type 1 (1)
Glycogen Storage Disease Type 1a (7)
Glycogen Storage Disease Type 1b (9)
Glycogen Storage Disease Type 3 (9)
GRACILE Syndrome (1)
Hb Beta Chain-Related Hemoglobinopathy (Including Beta Thalassemia and Sickle Cell Disease) (28) ACOG
Hereditary Fructose Intolerance (3)
Herlitz Junctional Epidermolysis Bullosa, LAMA3-, LAMB3-, LAMC2-Related (5)
Hexosaminidase A Deficiency (Including Tay-Sachs Disease) (9) ACOG ACMG
Homocystinuria Caused By Cystathionine Beta-Synthase Deficiency (1)
Hurler Syndrome (2)
Hypophosphatasia, Autosomal Recessive (4)
Inclusion Body Myopathy 2 (2)
Isovaleric Acidemia (1)
Joubert Syndrome 2 (1)
Krabbe Disease (2)
Limb-Girdle Muscular Dystrophy Type 2D and 2E (2)
Lipoamide Dehydrogenase Deficiency (2)
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (1)
Maple Syrup Urine Disease Type 1B (3)
Medium Chain Acyl-CoA Dehydrogenase Deficiency (2)
Megalencephalic Leukoencephalopathy With Subcortical Cysts (4)
Metachromatic Leukodystrophy (5)
Mucolipidosis IV (2) ACMG
Muscle-Eye-Brain Disease (1)
NEB-Related Nemaline Myopathy (1)
Niemann-Pick Disease, SMPD1-Associated (4) ACMG
Niemann-Pick Disease Type C (1)
Nijmegen Breakage Syndrome (1)
Northern Epilepsy (1)
Pendred Syndrome (5)
PEX1-Related Zellweger Syndrome Spectrum (2)
Phenylalanine Hydroxylase Deficiency (13)
Polyglandular Autoimmune Syndrome Type 1 (2)
Pompe Disease (4)
PPT1-Related Neuronal Ceroid Lipofuscinosis (3)
Primary Carnitine Deficiency (1)
PROP1-Related Combined Pituitary Hormone Deficiency (1)
Rhizomelic Chondrodysplasia Punctata Type 1 (4)
Segawa Syndrome (1)
Short Chain Acyl-CoA Dehydrogenase Deficiency (1)
Sjogren-Larsson Syndrome (1)
Smith-Lemli-Opitz Syndrome (13)
Spinal Muscular Atrophy (1) ACMG
Steroid-Resistant Nephrotic Syndrome (2)
Sulfate Transporter-Related Osteochondrodysplasia (5)
TPP1-Related Neuronal Ceroid Lipofuscinosis (3)
Tyrosinemia Type 1 (6)
Usher Syndrome Types 1F and 3 (2)
Very Long Chain Acyl-CoA Dehydrogenase Deficiency (1)
Wilson Disease (2)

Universal Panel

- ABCC8-Related Hyperinsulinism (3)
- Achromatopsia (3)
- Alkaptonuria (11)
- Alpha-1 Antitrypsin Deficiency (1)
- Alpha-Mannosidosis (1)
- Andermann Syndrome (2)
- ARSACS (2)
- Aspartylglycosaminuria (1)
- Ataxia with Vitamin E Deficiency (1)
- Ataxia-Telangiectasia (8)
- Autosomal Recessive Polycystic Kidney Disease (4)
- Bardet-Biedl Syndrome
  - BBS1-Related (1)
  - BBS10-Related (1)
- Biotinidase Deficiency (4)
- Bloom Syndrome (1) AC MG
- Canavan Disease (4) AC OG AC MG
- Carnitine Palmitoyltransferase IA Deficiency (1)
- Carnitine Palmitoyltransferase II Deficiency (3)
- Cartilage-Hair Hypoplasia (1)
- Choroideremia (1)
- Cohen Syndrome (1)
- Citrullinemia Type 1 (2)
- Congenital Disorder of Glycosylation
  - Type 1a (4)
  - Type 1b (1)
- Congenital Finnish Nephrosis (2)
- Costeff Optic Atrophy Syndrome (1)
- Cystic Fibrosis (100) AC OG AC MG
- Cystinosis (4)
- D-Bifunctional Protein Deficiency (2)
- Factor XI Deficiency (4)
- Familial Dysautonomia (2) AC OG AC MG
- Familial Mediterranean Fever (4)
- Fanconi Anemia Type C (3) AC MG
- *Fragile X Syndrome (female blood specimens only) (1)
- Galactosemia (8)
- Gaucher Disease (10) AC MG
- GJB2-Related DFNB 1 Nonsyndromic Hearing Loss and Deafness (7)
- Glutaric Acidemia Type 1 (1)
- Glycogen Storage Disease
  - Type Ia (7)
  - Type Ib (2)
  - Type III (3)
  - Type V (4)
- GRACILE Syndrome (1)
- Hb Beta Chain-Related Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (28) AC OG
- Hereditary Fructose Intolerance (3)
- Hereditary Thymine-Uraciluria (1)
- Herlitz Junctional Epidermolysis Bullosa
  - LAMA3-Related (1)
  - LAMB3-Related (3)
  - LAMC2-Related (1)
- Hexosaminidase A Deficiency (including Tay-Sachs Disease) (9) AC OG AC MG
- Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency (1)
- Hurler Syndrome (2)
- Hypophosphatasia, Autosomal Recessive (4)
- Inclusion Body Myopathy 2 (2)
- Isovaleric Acidemia (1)
- Joubert Syndrome 2 (1)
- Krabbe Disease (2)
- Limb-Girdle Muscular Dystrophy
  - Type 2D (1)
  - Type 2E (1)
- Lipoamide Dehydrogenase Deficiency (2)
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (1)
- Maple Syrup Urine Disease Type 1B (3)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (2)
- Megalencephalic Leukoencephalopathy with Subcortical Cysts (4)
- Metachromatic Leukodystrophy (5)
- Mucolipidosis IV (2) AC MG
- Muscle-Eye-Brain Disease (1)
- NEB-Related Nemaline Myopathy (1)
- Neuronal Ceroid Lipofuscinosis
  - CLN3-related (1)
  - CLN5-related (1)
  - PPT1-related (3)
  - TPP1-related (3)
- Niemann-Pick Disease
  - SMPD1-Associated (4) AC MG
  - Type C (1)
- Nijmegen Breakage Syndrome (1)
- Northern Epilepsy (1)
- Pendred Syndrome (5)
- PEX1-Related Zellweger Syndrome Spectrum (2)
- Phenylalanine Hydroxylase Deficiency (13)
- Polyglandular Autoimmune Syndrome Type 1 (2)
- Pompe Disease (4)
- Primary Carnitine Deficiency (1)
- Primary Hyperoxaluria
  - Type 1 (2)
  - Type 2 (2)
- PROP1-Related Combined Pituitary Hormone Deficiency (1)
- Pseudocholinesterase Deficiency (1)
- Pycnodysostosis (1)
- Rhizomelic Chondrodysplasia Punctata Type 1 (4)
- Salla Disease (1)
- Segawa Syndrome (1)
- Short Chain Acyl-CoA Dehydrogenase Deficiency (1)
- Sjogren-Larsson Syndrome (1)
- Smith-Lemli-Opitz Syndrome (13)
- Spinal Muscular Atrophy (1) AC MG
- Steroid-Resistant Nephrotic Syndrome (2)
- Sulfate Transporter-Related Osteochondrodysplasia (4)
- Tyrosinemia Type I (6)
- Usher Syndrome
  - Type 1F (1)
  - Type 3 (1)
- Very Long Chain Acyl-CoA Dehydrogenase Deficiency (1)
- Wilson Disease (2)
- X-Linked Juvenile Retinoschisis (3)

Counsyl is a medical genomics company founded by social entrepreneurs and philanthropists with the audacious belief that every child deserves a chance in life. With the Counsyl Test, many previously surprising and incurable childhood diseases can now be prevented.

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180 Kimball Way | South San Francisco, CA 94080

www.counsyl.com | contact@counsyl.com | (888) COUNSYL