Mount Sinai - Counsyl Expanded Panel

ACOG = Testing for this disease recommended to be offered by the American College of Obstetricians and Gynecologists
ACMG = Testing for this disease recommended to be offered by the American College of Medical Genetics

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