COUNSYL FAMILY PREP SCREEN DISEASE LIST

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) ACMG

Canavan Disease (4) ACOG ACMG

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) ACOG ACMG

Cystinosis (4)

D-Bifunctional Protein Deficiency (2)

Factor XI Deficiency (4)

Familial Dysautonomia (2) ACOG ACMG

Familial Mediterranean Fever (4)

Fanconi Anemia Type C (3) ACMG

Fragile X Syndrome (female specimens only) (1)

Galactosemia (8)

Gaucher Disease (10) ACMG

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) ACOG

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) 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 (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) ACMG

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) ACMG
- 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) ACMG

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)

ACOG
Indicates testing recommended by ACOG

ACMG
Indicates testing recommended by ACMG

Additional Information: Counsyl screening identifies but does not eliminate risk. Results are based on probabilities, and as such, cannot give 100% definitive conclusions, and cannot predict all disease. In addition to the Family Prep Screen, further testing options may be recommended to your patients. If only one member of a couple is of Ashkenazi Jewish background, a biochemical assay for Tay-Sachs disease can be performed.[1] Individuals of African, Asian, and Mediterranean ancestry are at increased risk for being carriers for hemoglobinopathies and should also be offered carrier testing by CBC and hemoglobin electrophoresis or HPLC.[2]

1. S Gross, BA Pletcher, KG Monaghan. Carrier screening in individuals of Ashkenazi Jewish descent. *Genetics in Medicine* (2008) 10. 54–56.
2. ACOG, Hemoglobinopathies in pregnancy. ACOG Practice Bulletin No. 78. (2007), 1–9.