Family Prep Screen

Our expanded carrier screen uses next-generation sequencing to look at mutations across 105 genes. Physicians offer the Family Prep Screen more than any other form of expanded carrier screening.

Disease list categories

The information presented is meant as a quick reference to diseases screened by Counsyl and is not meant to be a comprehensive guide. Individual diseases can have widely varying phenotypes not captured here. For specific disease information, please refer to counsyl.com/diseases.

<table>
<thead>
<tr>
<th>Category</th>
<th>Number</th>
<th>Description</th>
<th>Examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Have limited or no treatment options</td>
<td>79</td>
<td>These diseases do not currently have available and/or effective treatment options. While treatments may be available for some of the conditions listed here, these offerings are extremely limited in their ability to relieve or modify symptoms or may not be widely accessible.</td>
<td>Krabbe disease, Bloom syndrome, Pompe disease</td>
</tr>
<tr>
<td>Lead to shortened life expectancy</td>
<td>58</td>
<td>Shortened life expectancy is defined as a decrease in average lifespan for most (&gt;60%) individuals with these conditions. Impact on quality of life can include lifelong management of chronic symptoms or complications.</td>
<td>Cystic fibrosis: 35 years, Tay-Sachs disease: 3–5 years, Spinal muscular atrophy: less than 2 years</td>
</tr>
<tr>
<td>Carry a risk for intellectual disability</td>
<td>46</td>
<td>Conditions in this category are associated with a significant risk for intellectual disability either with or without application of standard treatment. The severity of intellectual disability is not considered in this count.</td>
<td>Fragile X syndrome, Smith-Lemli-Opitz syndrome, Metachromatic leukodystrophy</td>
</tr>
<tr>
<td>Improve with early intervention</td>
<td>61</td>
<td>There is a standard, recommended treatment that is reasonably accessible to most individuals with the disease. This does not include experimental approaches.</td>
<td>Galactosemia: normal life quality with treatment, Wilson disease: most symptoms prevented with early treatment, Phenylalanine hydroxylase deficiency: normal life quality with treatment</td>
</tr>
</tbody>
</table>

1. At-risk couples are defined as a couple who are both carriers of a mutation for the same autosomal recessive genetic disease, and have a 1/4 risk of having a child affected with the disease. This percentage also includes carriers of fragile X and other X-linked diseases.
### Disease list

Below are the 105 diseases on the Universal panel of the *Family Prep Screen*.

#### UNIVERSAL PANEL

- 21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia (12)
- ABCC8-related hyperinsulinism
- Achromatopsia
- Alkaptonuria
- Alpha-1 antitrypsin deficiency
- Alpha-mannosidosis
- Alpha-thalassemia (13)  
  - ACOG  
  - ACMG
- Andermann syndrome
- ARSACS
- Aspartylglycosaminuria
- Ataxia with vitamin E deficiency
- Ataxia-telangiectasia
- Autosomal recessive polycystic kidney disease
- Bardet-Biedl syndrome  
  - BB51-related
  - BB510-related
- Biotinidase deficiency
- Bloom syndrome  
  - ACOG
  - ACMG
- Canavan disease  
  - ACOG
  - ACMG
- Carnitine palmitoyltransferase IA deficiency
- Carnitine palmitoyltransferase II deficiency
- Cartilage-hair hypoplasia
- Choroideremia
- Cohen syndrome
- Citrullinemia type 1
- Congenital disorder of glycosylation  
  - Type 1a
  - Type 1b
- Congenital Finnish nephrosis
- Costeff optic atrophy syndrome
- Cystic fibrosis  
  - ACOG
  - ACMG
- Cystinosis
- D-bifunctional protein deficiency
- Factor XI deficiency
- Familial dysautonomia  
  - ACOG
  - ACMG
- Familial Mediterranean fever
- Fanconi anemia type C  
  - ACOG
  - ACMG
- Fragile X syndrome  
  - (female specimens only)  
  - (1)
- Galactosemia
- Gaucher disease (10)  
  - ACOG
- GJB2-related DFNB 1 Nonsyndromic hearing loss and deafness
- Glutaric acidemia type 1
- Glycogen storage disease  
  - Type 1a
  - Type 1b
  - Type III
  - Type V
- GRACILE syndrome
- Hb beta chain-related hemoglobinopathy (including beta thalassemia and sickle cell disease)  
  - ACOG
- Hereditary fructose intolerance
- Hereditary thymine-uraciluria
- Herlitz junctional epidermolysis bullosa  
  - LAMA3-related
  - LAMB3-related
  - LAMC2-related
- Hexosaminidase A deficiency  
  - (including Tay-Sachs disease)  
  - ACOG
  - ACMG
- Homocystinuria caused by cystathionine beta-synthase deficiency
- Hurler syndrome (2)
- Hypophosphatasa, autosomal recessive
- Inclusion body myopathy 2
- Isovaleric acidemia
- Joubert syndrome 2
- Krabbe disease
- Limb-Girdle muscular dystrophy  
  - Type 2D
  - 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
- Mucolipidosis IV  
  - ACOG
- Muscle-eye-brain disease
- NEB-related nemaline myopathy
- Neuronal ceroid lipofuscinosis  
  - CLN3-related
  - CLN5-related
  - PPT1-related
  - TPP1-related
- Niemann-Pick disease  
  - SMPD1-associated
  - Type C
- Nijmegen breakage syndrome
- Northern epilepsy
- Pendred syndrome
- PEX1-related Zellweger syndrome spectrum
- Phenylalanine hydroxylase deficiency
- Polyglutamyl autoimmune syndrome type 1
- Pompe disease
- Primary carnitine deficiency
- Primary hyperoxaluria  
  - Type 1
  - Type 2
- PROP1-related combined pituitary hormone deficiency
- Pseudocholinesterase deficiency
- Pycnodysostosis
- Rhizomelic chondrodysplasia punctata type 1
- Salla disease
- Segawa syndrome
- Short chain Acyl-CoA dehydrogenase deficiency
- Sjogren-Larsson syndrome
- Smith-Lemli-Opitz syndrome
- Spinal muscular atrophy (1)  
  - ACOG
- Steroid-resistant nephrotic syndrome
- Sulfate transporter-related osteochondrodysplasia
- Tyrosinemia type 1
- Usher syndrome  
  - Type 1F
  - Type 3
- Very long chain Acyl-CoA dehydrogenase deficiency
- Walker-Warburg syndrome
- Wilson disease
- X-Linked juvenile retinoschisis

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**Additional information**

In addition to the *Family Prep Screen*, further testing options may be recommended to your patients, such as a biochemical assay for Tay-Sachs disease¹ or CBC and hemoglobin electrophoresis/HPLC² for hemoglobinopathies.²

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¹Number in parenthesis represents the number of variants analyzed using targeted genotyping.