Counsyl

# 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.

**3.4%**

of couples are identified as at-risk couples by the *Family Prep Screen*[1]

## 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**.

### 79 Have limited or no treatment options

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.

**Diseases include:**

- Krabbe disease
- Bloom syndrome
- Pompe disease

### 58 Lead to shortened life expectancy

Shortened life expectancy is defined as a decrease in average lifespan for most (>60%) individuals with these conditions. Impact on quality of life can include lifelong management of chronic symptoms or complications.

**Diseases include:**

- Cystic fibrosis: *35 years*
- Tay-Sachs disease: *3–5 years*
- Spinal muscular atrophy: *less than 2 years*

### 46 Carry a risk for intellectual disability

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.

**Diseases include:**

- Fragile X syndrome
- Smith-Lemli-Opitz syndrome
- Metachromatic leukodystrophy

### 61 Improve with early intervention

There is a standard, recommended treatment that is reasonably accessible to most individuals with the disease. This does not include experimental approaches.

**Diseases include:**

- Galactosemia: *normal life quality with treatment*
- Wilson disease: *most symptoms prevented with early treatment*
- Phenylalanine hydroxylase deficiency: *normal life quality with treatment*

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.

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## 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
- BBS1-related
- BBS10-related

Biotinidase deficiency

Bloom syndrome 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 ACMG

Fragile X syndrome (female specimens only) (1)

Galactosemia

Gaucher disease (10) ACMG

GJB2-related DFNB 1 Nonsyndromic hearing loss and deafness

Glutaric acidemia type 1

Glycogen storage disease
- Type Ia
- Type Ib
- 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)

Hypophosphatasia, 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 ACMG

Muscle-eye-brain disease

NEB-related nemaline myopathy

Neuronal ceroid lipofuscinosis
- CLN3-related
- CLN5-related
- PPT1-related
- TPP1-related

Niemann-Pick disease
- SMPD1-associated ACMG
- Type C

Nijmegen breakage syndrome

Northern epilepsy

Pendred syndrome

PEX1-related Zellweger syndrome spectrum

Phenylalanine hydroxylase deficiency

Polyglandular 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) ACMG

Steroid-resistant nephrotic syndrome

Sulfate transporter-related osteochondrodysplasia

Tyrosinemia type I

Usher syndrome
- Type 1F
- Type 3

Very long chain Acyl-CoA dehydrogenase deficiency

Walker-Warburg syndrome

Wilson disease

X-Linked juvenile retinoschisis

ACOG
Indicates testing recommended by ACOG

ACMG
Indicates testing recommended by ACMG

### 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 disease[1] or CBC and hemoglobin electrophoresis/HPLC[2] for hemoglobinopathies.[2]

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

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.