### SOGC-RECOMMENDED CONDITIONS

<table>
<thead>
<tr>
<th>Condition</th>
<th>Notes</th>
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<tbody>
<tr>
<td>Alpha Thalassemia (HBA1/HBA2)</td>
<td>* Indicates testing also recommended by ACOG</td>
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<tr>
<td>Andersen-Armstrong Syndrome (SLC12A6)</td>
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<td>ARSACS (SACS)</td>
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<tr>
<td>Bartel-Bied Syndrome, BB1-Related (BB1)</td>
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<tr>
<td>Bartel-Bied Syndrome, BB11-Related (BB11)</td>
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<tr>
<td>Bartel-Bied Syndrome, BB12-Related (BB12)</td>
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<tr>
<td>Bartel-Bied Syndrome, BB12-Related (BB12)</td>
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<tr>
<td>Bloom Syndrome (BLU)</td>
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<tr>
<td>Canavan Disease (ASPA)</td>
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<tr>
<td>CLN3-Related Neuronal Ceroid Lipofuscinoses (CLN3)</td>
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<td>CLN5-Related Neuronal Ceroid Lipofuscinoses (CLN5)</td>
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<tr>
<td>CLN9-Related Neuronal Ceroid Lipofuscinoses, Type 9 (CLN9)</td>
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<tr>
<td>Congenital Disorder of Glycosylation, Type Ib (MNGI)</td>
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<tr>
<td>Cystic Fibrosis (CFTR)</td>
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<td>Eccrine-Gland-Related Perforating Lesions (EIRCE)</td>
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<td>Ferritin-Related Disorders (FIRDR)</td>
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<td>Fanconi Anemia, Type C (FANCIC)</td>
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<tr>
<td>FXTN-Related Disorders (including Walker-Warburg Syndrome) (FXN7)</td>
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<tr>
<td>Fragile X Syndrome (FMR1)</td>
<td>* Indicates X-linked disorders</td>
</tr>
<tr>
<td>Gaucher Disease (GBA)</td>
<td>* Indicates X-linked disorders</td>
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* Indicates analyzed using targeted genotyping

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### DISEASE CATEGORIES

#### EXPANDED CARRIER SCREENING

Many of the diseases included in our screen are vital to know about. The categories below provide an overview of the types of conditions included in the Foresight Carrier Screen.

#### Early intervention
Some of the conditions on the Foresight Carrier Screen can be treated early in life, like Wilson disease and PKU.

#### Intellectual disability
Some result in intellectual disabilities, as with fragile X syndrome and Niemann-Pick disease.

#### Shortened life expectancy
Others are chronic and require lifelong management, like cystic fibrosis and Bloom syndrome.

#### Limited or no treatment
Finally, some of the conditions have no treatments available, like spinal muscular atrophy and Canavan disease.

- You can take the Expanded Carrier Screen before or during pregnancy
- It’s normal to be a carrier – what you really want to know is if both partners are carriers of the same disease
- Most carriers have no history of the condition within their family

Knowing whether you are a carrier of genetic conditions provides valuable health information when planning a family.

Please visit LifeLabsGenetics.com for the full list of conditions covered on the Counsyl Foresight Carrier Screen. The following five conditions are also available, however, they are not routinely included in the Foresight Screen due to low clinical utility: MTHFR, hemochromatosis, G6PD deficiency, factor V Leiden, and prothrombin. Please contact LifeLabs Genetics for more information.

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1-844-363-4357
The Counsyl Foresight Carrier Screen covers many types of conditions:

- Spinal muscular atrophy
- Tay-Sachs disease
- X-linked spinal muscular atrophy
- Beta thalassemia
- Wilson Disease
- Costeff optic atrophy
- Smith-Lemli-Opitz Syndrome
- Diphtheria
- Hairy cell leukemia
- Fabry disease
- Gaucher type 1 disease
- Gaucher type 3 disease
- Glycogen storage disease type III
- Hereditary motor and sensory neuropathy type V
- Hereditary motor and sensory neuropathy type VII
- Hurler syndrome
- Kearn-Sayre syndrome
- Krabbe disease
- McLeod syndrome
- Niemann-Pick C
- Phenylketonuria
- Refsum disease
- Retinitis pigmentosa
- Spastic paraplegia
- Tay-Sachs disease
- Wiskott-Aldrich Syndrome
- Wilson disease

The option of (Expanded Carrier Screening) should be part of the informed consent process.

I am the type of person that likes to have as much information as possible to prepare.

"All individuals, regardless of race or ethnicity, are offered screening for the same set of conditions." – ACOG, ACMG, NSGC, PGFM, SMFM Joint Statement

The percentage of affected pregnancies missed by the current, ethnicity-based guidelines include:

- East Asian: 23%
- South Asian: 10%
- African American: 12%
- Hispanic: 12%
- Middle Eastern: 19%
- North European: 5%
- South European: 10%
- North American: 15%

WHAT IF AN INDIVIDUAL IS NOT A CARRIER?

When two parents are carriers of the same genetic mutation, each child has a 1 in 4 (or 25%) chance of being affected by the associated condition. For certain conditions, such as Fragile X syndrome, only the mother needs to be carrier for the child to be at an increased risk of being affected by the condition.

WHAT IS CARRIER SCREENING?

A carrier screen analyzes a person's genes to determine if they carry a recessive genetic condition. A screen is able to detect if you are a carrier of many, but not all, conditions.

WHAT IS A RECESSIVE DISEASE AND WHAT IS A CARRIER?

Recessive conditions are caused by changes, called mutations, in persons' genes. Each person has 2 copies of any given gene, one copy inherited from each parent. A recessive condition occurs when both copies of the same gene have a mutation. A carrier is someone who has only one gene with a mutation and one gene that is normal. Carriers are typically symptom-free and do not know they carry a mutation.

WHAT IF AN INDIVIDUAL IS NOT A CARRIER?

Generally, no further testing is suggested for the candidate screening. It is important to understand that a screening is able to identify every carrier of every condition. You should also know that while the Expanded Carrier Screen covers a comprehensive analysis for detection of genetic mutations. Next-generation sequencing of the entire gene of interest provides the most comprehensive analysis for detection of genetic mutations. The percentage of affected pregnancies missed by the current, ethnicity-based guidelines include: 23%, 10%, 12%, 12%, 19%, 5%, 10%, 15%.

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