DISEASE CATEGORIES

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.

The SOGC-recommended conditions listed above are included in the 175 that are tested on the Expanded Carrier Screen.

Glycogen Storage Disease, Type I (GPPC)
GM1E-Related Disorders (GM1E)
Hemoglobinopathies (including Beta-Thalassemia and Sickle Cell Disease) (HBB)
Hemochromatosis A-Deficiency (including Tar-Sachs Disease) (HEXA)
Joubert Syndrome 2 (MKS2B16)
KCNJ11-Related Familial Hypertension (KCNJ11)
Leigh Syndrome, French-Canadian Type (UPPPC)
Lipoxygenase Oxidoreduction Deficiency (LOX)
Maple Syrup Urine Disease, Type B (RCK2HB)
Mucolipidosis IV (MCLIV)
Niemann-Pick Disease, Type C2 (NPC2)
Spinal Muscular Atrophy (SMN1)*
Tyrosinemia, Type I (FAH)
Usher Syndrome, Type 3 (CLRN1)

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- **GPPC** indicates testing also recommended by ACOG
- **GM1E** indicates testing also recommended by ACMG
- **X-linked** indicates X-linked disorders

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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.
The Counsyl Foresight Carrier Screen covers many types of conditions:

- **Down Syndrome**: Affected births
- **Neural Tube Defects**: Affected births
- **Open Neutral Tube Defects**: Affected births

### ABOUT THE CATEGORIES

The information presented in this document is meant as a quick reference to diseases screened by Counsyl Family Prep Screen and Counsyl Expanded Carrier Screen. A comprehensive analysis of detection for genetic mutations is beyond the scope of this document.

### PATIENT FLOW

1. **Ordering**
   - Physicians complete requisition if interested. Parents have the option to obtain a local carrier counselor or request information elsewhere.

2. **Results**
   - Physicians share report with patients.
   - Patients have access to view their results through their personal portal.

3. **Counselling**
   - Counsellors provide education to patients.
   - Physicians have access to view the results.

4. **Follow Up**
   - Physicians share results with patients.
   - Patients have access to view their results through their personal portal.

### PATIENTS TO CONSIDER:

- "All individuals, regardless of race or ethnicity, are offered screening for the same set of conditions." – ACMG, ACNSG, PGF, SMFM Joint Statement

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

  - **94% East Asian**
  - **79% Hispanic**
  - **65% Northeast European**
  - **55% Ashkenazi Jewish**

  "The option of (Expanded Carrier Screening) should be part of the informed consent process." – CCMS and SOGC Joint Opinion

### WHAT IS CARRIER SCREENING?

A carrier screen is a process 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 a person’s genes. Each person has two 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 unaffected. Carriers are typically symptom-free and do not know they carry a mutation.

When two parents are carriers of the same genetic mutation, each child has a 1 in 4 (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 IF AN INDIVIDUAL IS NOT A CARRIER?

If you are not a carrier, it means you do not have the associated condition. You should also know that if you are a carrier, you and your partner have an equal chance for each child to be a carrier of the condition.

### WHEN TO CONSIDER?

- When two parents are carriers of the same genetic mutation, each child has a 1 in 4 (25%) chance of being affected by the associated condition.

### WHEN TO INSTALL?

- When two parents are carriers of the same genetic mutation, each child has a 1 in 4 (25%) chance of being affected by the associated condition.

### WHAT IF AN INDIVIDUAL IS NOT A CARRIER?

If you are not a carrier, it means you do not have the associated condition. You should also know that if you are a carrier, you and your partner have an equal chance for each child to be a carrier of the condition.