



#### What is carrier screening?

Carrier screening is a type of genetic testing that determines your risks for passing on an inherited genetic condition to your child.

Carrier screening is a simple blood test that determines if you are a carrier of one or more autosomal recessive and / or X-linked genetic conditions.

#### Did you know that...

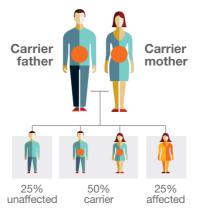
Most people are carriers of at least four to six genetic conditions

- Most carriers are healthy because the other copy of the gene works normally
- Most have no family history of the genetic condition they carry
- Carrier couples for the same condition are at increased chance to have a child with a genetic condition



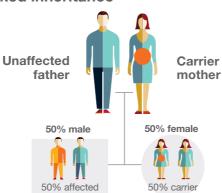
## How are genetic conditions passed down from carrier parents to children?

#### **Autosomal recessive inheritance**



If a woman and her partner are both carriers of the same condition, they have a 1-in-4, or 25%, chance with each pregnancy of having a child affected with the condition.

#### X-linked inheritance



If a woman is a carrier of an X-linked condition, she has a 1-in-2, or 50%, chance with each pregnancy of passing the gene mutation to her child. If the child is a boy, and he inherits the mutation, he will be affected with the condition.

### What does Horizon screen for?

Horizon screens for up to 603 genetic conditions. Here is a brief overview of the genetic conditions included in the screening\*:

- Adrenoleukodystrophy, X-Linked
- Alpha-thalassemia
- AR Spastic Ataxia of Charlevoix-Saguenay (SACS)
- Andermann syndrome (SLC12A6)
- Batten disease, CLN3-related
- Beta-hemoglobinopathies (including sickle-cell anemia)
- Bloom syndrome
- Canavan disease
- Citrullinemia, type 1
- Cystic fibrosis
- Duchenne/Becker muscular dystrophy
- · Familial dysautonomia
- Fanconi anemia group C
- Fragile X syndrome
- Galactosemia
- Gaucher disease
- · Glycogen storage disease, type 1a
- Isovaleric acidemia
- Leigh Syndrome (LRPPRC)
- Methylmalonic aciduria and homocystinuria, type cblC
- Mucolipidosis, type IV
- Mucopolysaccharidosis, type I (Hurler syndrome)
- Niemann-Pick disease, types A/B
- Polycystic kidney disease, autosomal recessive
- Rhizomelic chondrodysplasia punctata, type 1
- Smith-Lemli-Opitz syndrome
- Spinal muscular atrophy
- Tay-Sachs disease
- Tyrosinemia, type 1
- Zellweger spectrum disorders, PEX1-related

Horizon includes the most frequent French Canadian and Jewish-Ashekenaze conditions.

<sup>\*</sup> Men are only screened for autosomal recessive conditions.

# When should I have Horizon carrier screening?

Horizon can be performed any time before or during pregnancy. Some people may want to know their carrier status before pregnancy to inform reproductive decisions.

### How to get the test and receive the results?

Horizon is precribed by your healthcare provider and the blood draw is performed at ovo labo.



You can also book your appointment directly on our website: rdv-ovolabo.cliniqueovo.com

Results are returned to your provider in about 10-21 calendar days.

### What type of results can I get?

A negative result indicates a significantly lower chance to be a carrier, carrier screening cannot detect all disease causing mutations.

A positive result means that a mutation / change in one of your genes was detected. A positive result is not a diagnosis of the disease for yourself, it means that you are a carrier of a genetic condition. To assess the risk of passing the condition onto your child, your reproductive partner may need to be tested.

## What are my reproductive options if I am a carrier?

If you and your partner are both carriers of the same autosomal recessive condition, or if you are a carrier of an X-linked condition, you can choose the best course of action for your family. You may consider:



**Natural conception**, with an option of prenatal testing, such as amniocentesis or chorionic villi sampling, for the specific condition



**In vitro fertilization (IVF)** with preimplantation genetic diagnosis (PGD)



**Use of a sperm or egg donor** who is not a carrier for the condition



Adoption



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