horizon™
ADVANCED CARRIER SCREEN
POWERED BY NATERA™

Knowing matters

The Horizon™ carrier screen is a DNA screening test that provides information on your chance of having a child with a genetic condition

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What is carrier screening?

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

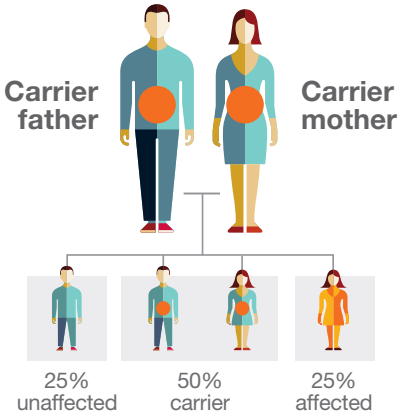
What does it mean to be a carrier?

A carrier of a genetic condition has a change (or "mutation") in one gene copy of a pair of genes.

- 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
- Carrier couples are at increased chance to have a child with a genetic condition

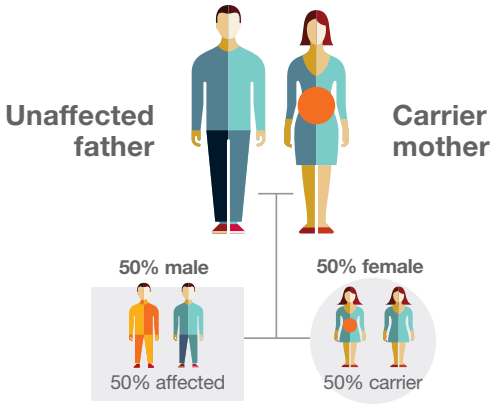
How are genetic conditions passed down from carrier parents to children?

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 274 genetic conditions. Your healthcare provider discuss the choice for carrier screening with you. This may include screening for a few or all of the conditions available through Horizon.

Our standard general population panel, Horizon 27, screens for the following conditions:

- Adrenoleukodystrophy, X-Linked
- Alpha-thalassemia
- 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
- 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

How is Horizon different?

Horizon screens for up to 274 genetic conditions. Your healthcare provider will discuss the choice for carrier screening with you. This may include screening for a few or all of the conditions available through Horizon.

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 do I get started with Horizon?

Horizon is available through your healthcare provider. Not sure if your healthcare provider offers Horizon? Contact your doctor or email support@natera.com

You can also learn more about Horizon by visiting our website: **http://www.natera.com/horizon-carrier-screen**

What do Horizon results tell me, and when?

Results are returned to your provider in about 2-3 weeks.

A positive result means that a disease-causing or likely disease-causing mutation was detected. It is important to determine your partner's carrier status to understand the chances of passing a genetic condition to your child.

A negative result means that no mutations for the conditions screened were found. While a negative result indicates a significantly lower chance to be a carrier, carrier screening cannot detect all disease-causing mutations.

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 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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