FullGenomics

HERES Carrier Screening test
Information for Patients

What is carrier screening?

Carrier screening is a genetic test that can determine a couple's risk of having a child affected with a genetic condition or disease. Typically, carriers of a genetic condition are healthy and not aware of their risk. If their partner is also a carrier for the same condition, they are at an increased risk of having a child affected with that condition. Every person is a carrier for a number of genetic changes that could cause disease in their child.

Who should have carrier screening?

According to the American College of Obstetricians and Gynecologists (ACOG), expanded carrier screening should be offered to pregnant women and women considering pregnancy, regardless of ethnicity and family history. Expanded carrier screening tests for hundreds of genetic conditions is considered an appropriate approach for carrier screening during pregnancy. If a woman is found to be a carrier for a specific condition, carrier screening for her partner should be considered.

What are the benefits of carrier screening?

HERES Carrier Screening provides information to make informed decisions regarding:

- Reproductive planning
- Prenatal testing options
- Preparation for the birth of a child with a genetic disorder

What will the HERES Carrier Screening test tell me?

By analyzing the genes in your blood or saliva sample, the HERES Carrier Screening test can tell you if you are a carrier for certain genetic conditions. Carriers are usually healthy and do not show signs or symptoms of the condition, but they can potentially pass the disease on to their children. If you and your partner are carriers for the same condition, you are at increased risk of having a child affected by that condition.

Which genetic conditions are included in HERES Carrier Screening tests?

FullGenomics offers two different HERES Carrier Screening tests, genotyping and sequencing, which screens over 300 hereditary conditions. Screened conditions include:

- Cystic Fibrosis
- Fragile X
- Sickle Cell Anemia
- Spinal Muscular Atrophy
- Tay-Sachs
- Thalassemia
- Canavan Disease
- Bloom Syndrome
- Wilson Disease

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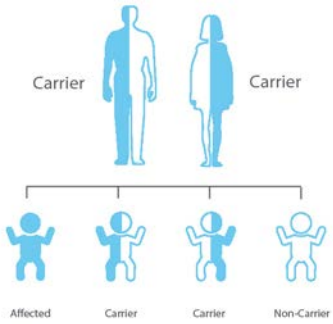
How are recessive genetic conditions inherited?

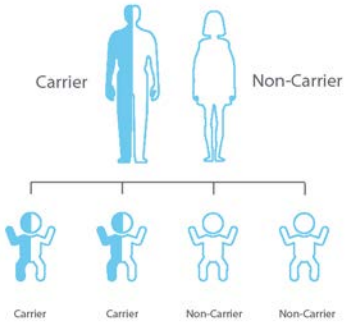
There are two broad types of conditions that are screened by this test: Autosomal recessive and X-linked.

Autosomal recessive conditions

For most genes we have two copies, one we inherit from our fathers and one we inherit from our mothers. Autosomal recessive conditions will only manifest if both copies of the gene are affected.



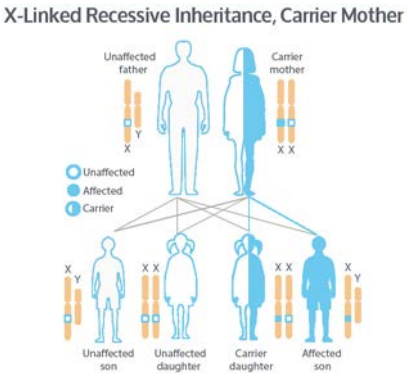
If both parents carry a disease-causing mutation in the same gene, each time they are pregnant there is a 1 in 4 (25%) chance of having an affected child.



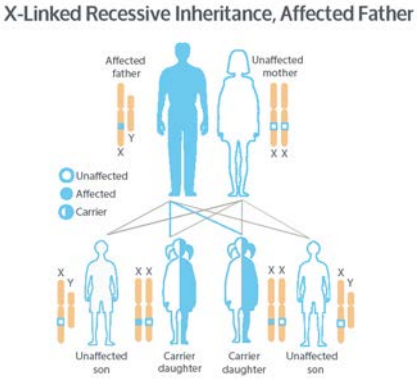
If only one parent carries a disease-causing variant but the other parent does not, their children will not be affected. The children have a chance of being healthy carriers.

X-linked conditions

Some disease-causing variants are located on the X-chromosome. Females have two X-chromosomes while males have only one. Males that inherit disease-causing variants on the X-chromosome are always affected, while females are often unaffected carriers.



If a woman carries a disease-causing variant on the X-chromosome, each of their children has a 1 in 2 (50%) chance of inheriting the variant. Sons will be affected and daughters will be carriers.



If a man is affected by an X-linked condition, all his daughters will be carriers and his sons are not at risk (there is no male-to-male transmission in X-linked conditions).

How do I request a HERES Carrier Screening test from my doctor?

Your doctor can request the proper test from FullGenomics prior to your next appointment and order the test during your next visit. Once your sample is received at the lab, your results will be available in approximately three weeks.