UW Medicine

Expanded Carrier Testing

A genetic test for hereditary diseases

This handout gives information to help you decide if you want to have a test to see if you are a carrier of a genetic disease.

Having this test is up to you. Some people do not find this type of screening to be helpful. You may choose not to be tested at any time.

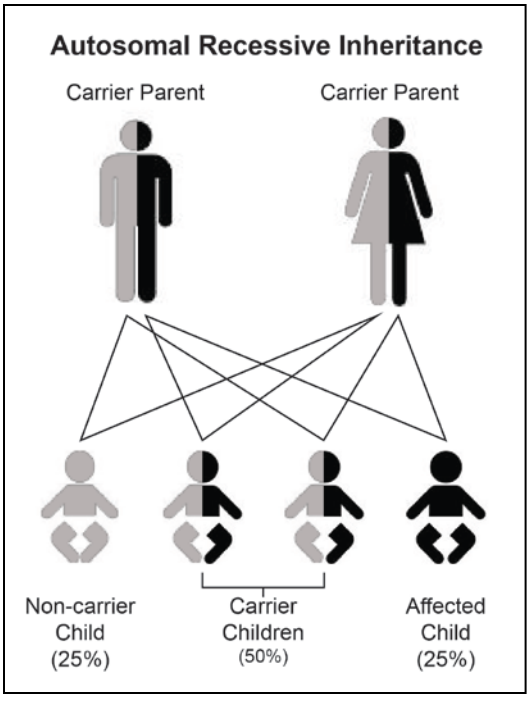
Talk with your healthcare provider to learn more.

What is expanded carrier testing?

Expanded carrier testing is a test to see if you could pass certain rare *hereditary* diseases to your children. A hereditary disease is a disease that is *inherited* (passed from parents to their children). It is also called a *genetic* disease.

What is a "carrier"?

People with 1 normal copy and 1 abnormal copy of a *gene* (see "Definitions" on the left side of page 2) for a hereditary disease are called *carriers*. Carriers are healthy and do not have symptoms of the disease. But, couples who are both carriers for the same disease could have a child who has that disease.



This drawing shows the chance of inheriting a genetic disease when both parents are carriers of the disease.

What is the chance of being a carrier for any of these rare diseases?

We know that each person may be a carrier for 3 or 4 rare diseases. The chance that you are a carrier for a specific disease may simply be the chance in the general population. But it may be higher, depending on your ancestry.

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1959 N.E. Pacific St., Seattle, WA 98195 | 206.598.4072

Definitions:

Gene: *The basic unit of heredity. Each gene has a specific code. The code is used to make a specific protein for a function or "job" in the body.*

Mutation: *Any change in a gene from its natural state. A mutation may or may not cause a disease.*

For example, the table below shows the chance of being a carrier of cystic fibrosis based on your ancestry.

Example: Cystic Fibrosis

Ancestry Group	Chance of Being a Carrier
Eastern European (Ashkenazi) Jewish	about 4% (1 out of 24)
Non-Hispanic Caucasian	about 4% (1 out of 25)
Hispanic American	about 2% (1 out of 46)
African American	about 2% (1 out of 46)
Asian American	about 1% (1 out of 94)

Who should consider having expanded carrier testing?

- Couples who are related to each other (for example, cousins)
- People who do not have health information about their birth family (for example, people who are adopted)
- People with a family history of any of the diseases included in the test
- Couples who are at higher risk for many diseases because they have mixed ancestry

How is expanded carrier testing done?

You will need to give a small blood sample. It will be taken from a vein in your arm.

What are the benefits of this test?

Most people who are carriers for a hereditary disease have no family history of the disease. Without expanded carrier testing, couples may find out that they are carriers only after they have a child who is diagnosed with the disease.

Expanded carrier testing allows you to find out if you are a carrier before pregnancy or early in pregnancy. If the test shows you are a carrier, more tests would be needed to find out whether your baby is actually at risk or affected.

What does a "negative" test result mean?

A negative result means you do not have any of the common gene *mutations* you were tested for (see "Definitions" on the left side of this page). A negative result **lowers the chance** you are a carrier. But, it does not mean you are not a carrier. You might carry a rare mutation that was not included in the test.

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What does a "positive" test result mean?

A positive result means you are a carrier of a hereditary disease. If your test is positive, your partner can then be tested to see if you are both carriers for the same hereditary condition.

What if my partner's test result is "negative"?

A negative result **lowers the chance** that your partner is a carrier. It also lowers the chance that you would have a child with the disorder. But, your partner might carry an uncommon mutation that was not included in the test.

What if both of our test results are "positive?"

If you and your partner both have a positive test result that shows you are carriers of the **same** disease:

- There is a 25% (1 in 4) chance your baby will have the disease. There is a greater chance (75%, or 3 in 4) that your baby will **not** have the disease.
- Amniocentesis and *chorionic villus sampling* (CVS) are 2 tests that can be done while you are pregnant to test to see if your baby is affected by the disease. Or, a small blood sample can be taken from your baby after birth for testing.

What are the limits of expanded carrier testing?

- It does not test for all genetic diseases.
- It does not test for all mutations that could lead to a certain disease.
- The accuracy of the test is affected by your ancestry. For some diseases, ancestry will make the test more accurate or less accurate.

Questions?

Your questions are important. If you have questions or concerns about expanded carrier testing, talk with your healthcare provider before signing any consent forms.

- ❑ **UW Medical Center Prenatal Diagnosis Clinic:** 206.598.4072

  Clinic hours: Weekdays 8 a.m. to 5 p.m.

  Address: 1959 N.E. Pacific St., Seattle, WA 98195

- ❑ **UW Medicine Maternal Fetal Medicine Clinic at Yakima Valley Memorial Hospital:** 206.598.3900 or toll-free 855.464.6298

  Clinic hours: Weekdays 8 a.m. to 5 pm.

  Address: 3003 Tieton Dr., Suite 240, Yakima, WA 98902

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