UW Medicine
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Cystic Fibrosis

About carrier testing

This handout gives information to help you decide if you want to have a test to see if you are a carrier of cystic fibrosis. Having this test is up to you. Some people do not find this type of screening test to be helpful. You may refuse testing at any time. Talk with your healthcare provider to learn more.

What is cystic fibrosis?

Cystic fibrosis (CF) is a lifelong condition that causes problems with breathing and often with digestion. CF does not affect how smart someone is or how they look.

There is no cure for CF, but there are many therapies and treatments for the symptoms. Many people with CF live into their 40s and older. But, the disease often gets worse over time and becomes harder to treat.

What causes CF?

Cystic fibrosis is an *inherited* disease. Every baby inherits 2 sets of *genes*, 1 from their mother and 1 from their father. Genes are the hereditary instructions that tell our bodies how to grow and develop. Each gene is part of a pair.

Every baby inherits 2 sets of genes, 1 from each of their parents.

When a person inherits an abnormal CF *gene* from **both** parents, their body makes thick, sticky mucus. This mucus clogs the lungs, leading to life-threatening lung infections. In most people with CF, the mucus also blocks the pancreas, which keeps the body from digesting food properly.

A person who has 1 normal CF gene and 1 abnormal CF gene is known as a *carrier* of CF. Carriers are healthy and have no symptoms of CF. But, if 2 carriers of CF have a child, there is a 25% (1 out of 4) chance that the child will have CF.

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What is the chance of being a CF carrier?

Cystic fibrosis occurs most often in people whose ancestors came from northern European countries, such as Ireland, England, or Holland. But, anyone can be a carrier of CF, as shown in this chart:

Ethnic Group	Chance of Being a Carrier	
Eastern European (Ashkenazi) Jewish	4%	(1 out of 24)
Non-Hispanic Caucasian	4%	(1 out of 25)
Hispanic American	2 %	(1 out of 46)
African American	1.5%	(1 out of 65)
Asian American	1%	(1 out of 94)

Data from ACOG Committee Opinion 325, December 2005

Most CF carriers do **not** have family members with cystic fibrosis. If someone in your family does have CF, your chance of being a carrier is probably higher than other people in your ethnic group. Be sure to tell your healthcare provider about your family's health history.

How can I find out if I am a CF carrier?

To have your CF genes tested, you will need to give a small blood sample. The lab checks for the most common changes, known as *mutations*, in the CF gene. Over time, scientists have found more than 1,600 mutations in the CF gene. The standard test looks for the 23 mutations that are most common.

CF carrier testing is most reliable for Caucasians, because that is the group of people who usually have CF. Carrier testing is less reliable in people of other ethnic groups, as shown in this chart:

Ethnic Group	Accuracy of Carrier Test
Eastern European (Ashkenazi) Jewish	94%
Non-Hispanic Caucasian	88%
Hispanic American	72%
African American	65%
Asian American	49%

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What does a "negative" test result mean?

A *negative* test result means you do not have any of the CF gene mutations you were tested for. It does **not** mean that you're not a carrier, although chances are lower that you are. You could have one of the rare gene mutations that we do not search for.

What does a "positive" test result mean?

A *positive* test result means that you **are** a carrier of cystic fibrosis. There are no "false positives" with this test. If your CF test is positive, your partner can then be tested to see if you are both CF carriers.

What if my partner's test is negative?

If your partner's test is negative, it means their chance of being a carrier of CF is lower, but not zero. There is still a very small risk that your child will have CF.

What if my partner's test is positive?

If your partner's test is positive, there is:

- A 25% (1 out of 4) chance your baby **will** have CF
- A 75% (3 out of 4) chance your baby will **not** have CF

Can we test my baby for CF?

- *Amniocentesis* and *chorionic villus sampling* (CVS) are 2 tests that can be done while you are pregnant to test your baby for CF. These can be done as early as 10 weeks of pregnancy.
- After birth, your baby will be screened for CF at birth as a part of newborn screening. But, newborn screening does not pick up all cases of CF. If you and your partner are both carriers of CF, do not rely on newborn screening results to diagnose CF.
- For more complete genetic testing, we can take a small blood sample from your newborn baby to diagnose CF.

Questions?

Your questions are important. If you have any questions about amniocentesis or the risks, benefits, or alternatives to it, talk with your provider before signing any consent forms.

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Published PFES: 2003, 2009, 2020
Clinician Review: 02/2009
Reprints on Health Online: https://healthonline.washington.edu

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