



Cystic Fibrosis: Prenatal Screening and Diagnosis

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What is cystic fibrosis (CF)?

Cystic fibrosis (CF) is a disease that affects a person's long-term health and lifespan. It often causes problems with digestion and breathing. In some cases, CF can be a mild disease. But in most people with CF, it poses a serious risk to a person's health. The average lifespan of a person with CF is 37 years. Those with a milder form can live into their 50s.

What causes CF?

Cystic fibrosis is a **genetic disorder** caused by a **gene** that is passed from parent to child. It takes two genes—one from the mother and one from the father—for a person to have CF. If a person has only one copy of a gene for CF, he or she is known as a **carrier**. Carriers often do not know that they have a gene for CF. They usually do not have symptoms or may have only mild symptoms. If both parents are carriers, each of their children has a 25% chance of having the disorder. Put another way, this couple has a 1-in-4 chance of having a child with CF.

What are the symptoms of CF?

The symptoms of CF can vary in type and severity. Many people with CF produce a thick, sticky mucus in their bodies. This mucus builds up and clogs the lungs, which makes it hard to breathe and can lead to infection. Mucus buildup also can affect the digestive organs, making it hard for the body to break down food and absorb nutrients. Most males with CF are sterile and cannot have children.

Is treatment available for CF?

New drugs and treatments have improved the outlook for people with CF, but there is no cure. The disease gets worse the longer a person has it. To treat lung problems, children with CF need to have physical therapy for about a half hour every day. This therapy helps clear mucus from the lungs. The therapy can be done by parents or other family members.

What are risk factors for CF?

The risk of being a CF carrier is increased in families with a history of CF. The risk also is higher in certain races and ethnic groups. It occurs more often in non-Hispanic white people than in other racial groups. But as our population has become more diverse, it is harder to assign a person to just one ethnicity. For this reason, the American College of Obstetricians and Gynecologists recommends offering **carrier screening** to all women who are thinking about becoming pregnant or are currently pregnant.

What is carrier screening?

Carrier screening uses a sample of blood, saliva, or tissue from the inside of the cheek. Carrier testing is voluntary. You can choose to have carrier screening or not to have it. Currently, there are several approaches to carrier screening:

- Testing based on your ethnicity if your ethnic group is known to be at higher risk (**ethnic-based screening**)
- Testing for many disorders at once (**expanded carrier screening**)
- Testing for just a few specific disorders

When should I have carrier screening?

Carrier screening can be done before pregnancy or during pregnancy. If you have carrier screening before you become pregnant and both you and your partner are carriers, you have more options. If you have carrier screening while you are pregnant, you have fewer options. Prenatal **diagnostic tests** are available to test whether the **fetus** has CF or is a CF carrier. This type of testing can be done as early as 10 weeks of pregnancy.

How is carrier screening for CF done?

You are usually tested first. If results show that you are a carrier, your partner is tested. If you already are pregnant, you and your partner can be tested together. If your partner has a family history of CF, he may be tested first.

What does it mean if the test result for one partner is negative?

If your test result is negative, the chance that you are a CF carrier is small, but no screening test checks for every known CF **mutation**. For this reason, if your test result is negative, there still is a very small chance that you could be a carrier of a mutated gene that was not detected by the test.

What does it mean if the test result for one partner is positive?

If your test result is positive, it means that you are a CF carrier. The next step is to test your partner. Both parents must be CF carriers for the baby to have CF. If one parent has a negative test result, the chance that the baby will have CF is small. If only one partner is a carrier and the other has a negative result, no further testing is recommended.

What does it mean if the test results for both partners are positive?

If two people who are both CF carriers have a baby, there is a 25% (1-in-4) chance that the baby will have CF. However, it is more likely that the baby will be a carrier and not have the disease. There also is a 1-in-4 chance that the baby will not have CF and will not be a CF carrier.

If both partners are positive, what are the follow-up tests and what can they show?

If you are pregnant and you and your partner are CF carriers, prenatal diagnostic testing can be done to detect whether the fetus has CF. The results of these prenatal tests can tell you with a high degree of certainty whether the fetus has CF or is a CF carrier. They cannot tell you exactly how severe or mild the disease will be if the fetus has the disorder.

Prenatal diagnostic tests include **chorionic villus sampling (CVS)** and **amniocentesis** (see [FAQ164 Prenatal Genetic Diagnostic Tests](#)). CVS can be performed between 10 weeks and 13 weeks of pregnancy. Amniocentesis usually is done between 15 weeks and 20 weeks of pregnancy, but it also can be done up until you have the baby.

Diagnostic testing is voluntary. Some parents want to know this information before the birth of the baby. Other parents do not want to know. There is no right or wrong answer.

What are my options if diagnostic test results show that the fetus has CF?

Two options are available:

1. Continue the pregnancy and prepare for a child with CF. Couples can use this time to learn as much as possible about the disease, current treatment options, and the experiences of other families who have a child with CF.
2. End the pregnancy. Each state has its own laws on pregnancy termination. Your health care professional can answer any questions you may have. You also may want to talk with your partner, counselors, and close friends.

What about future pregnancies?

If a test result shows that you are a CF carrier, the result is definite and will not change. If both partners are carriers, it means that in each pregnancy the fetus will have a 25% (1-in-4) chance of having CF. In this case, you have several options for future pregnancies:

- You can accept the level of risk and become pregnant. You may choose to have prenatal diagnostic testing in each pregnancy, or you may not. If you want to know whether your baby will have CF, you will need to have amniocentesis or CVS in each pregnancy.

- You can adopt.
- You can use **in vitro fertilization (IVF)** with donor sperm or donor eggs (but the donor should be tested for CF carrier status).
- You can use IVF with your own sperm and eggs, and then use **preimplantation genetic diagnosis** to see if the fertilized egg has CF or is a CF carrier.

Glossary

Amniocentesis: A procedure in which a needle is used to withdraw and test a small amount of amniotic fluid and cells from the sac surrounding the fetus.

Carrier: A person who shows no signs of a particular disorder but could pass the gene on to his or her children.

Carrier Screening: A test done on a person without signs or symptoms to find out whether he or she carries a gene for a genetic disorder.

Chorionic Villus Sampling (CVS): A procedure in which a small sample of cells is taken from the placenta and tested.

Cystic Fibrosis (CF): An inherited disorder that causes problems in digestion and breathing.

Diagnostic Tests: Tests that look for a disease or cause of a disease.

Ethnic-Based Screening: Carrier screening recommended for people who belong to an ethnic group or race that has a high rate of carriers of a specific genetic disorder.

Expanded Carrier Screening: A carrier screening technology that allows a large number of disorders to be screened for simultaneously using a sample of a person's blood and without regard to the person's race or ethnicity.

Fetus: The stage of prenatal development that starts 8 weeks after fertilization and lasts until the end of pregnancy.

Gene: A segment of DNA that contains instructions for the development of a person's physical traits and control of the processes in the body. It is the basic unit of heredity and can be passed down from parent to child.

Genetic Disorder: A disorder caused by a change in genes or chromosomes.

In Vitro Fertilization (IVF): A procedure in which an egg is removed from a woman's ovary, fertilized in a laboratory with the man's sperm, and then transferred to the woman's uterus to achieve a pregnancy.

Mutation: A permanent change in a gene that can be passed from parent to child.

Preimplantation Genetic Diagnosis: A type of genetic testing that can be done during in vitro fertilization. Tests are performed on the fertilized egg before it is transferred to the uterus.

If you have further questions, contact your obstetrician–gynecologist.

FAQ171: Designed as an aid to patients, this document sets forth current information and opinions related to women's health. The information does not dictate an exclusive course of treatment or procedure to be followed and should not be construed as excluding other acceptable methods of practice. Variations, taking into account the needs of the individual patient, resources, and limitations unique to the institution or type of practice, may be appropriate.

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