

Giving parents-to-be a more complete picture

CFvantage® Cystic Fibrosis Expanded Screen



Quest
Diagnostics™

Approach your family planning with more knowledge and confidence

What every future parent should know

People are born with cystic fibrosis (CF) because they inherit altered CF genes from their parents, who usually are not affected by CF themselves but silently carry the altered gene. Unless they are tested, potential parents have no way of knowing they are CF carriers. With more than 10 million CF carriers in the United States, it is important for anyone who is planning to have children to receive a CF carrier screening test. This brochure will help you understand CF, explain how your family history and ethnic background can increase your chance of having a child with CF, and provide information about available testing, so you can make informed decisions about your family's future.

More than

10 million

Americans are CF carriers.

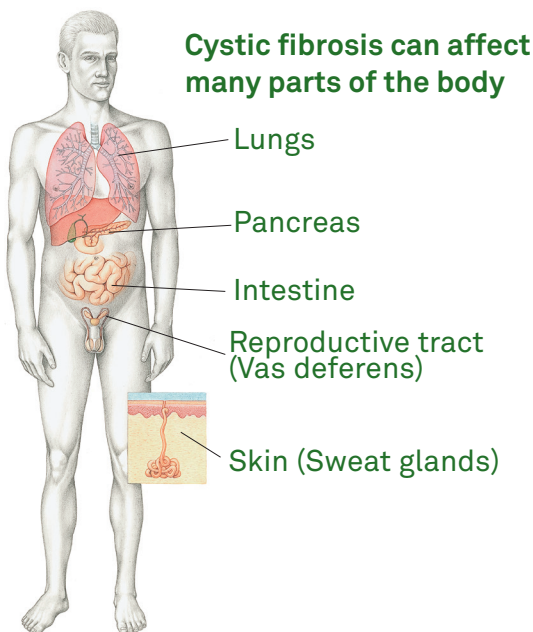


What is cystic fibrosis?

CF is a life-threatening disease that affects mainly the lungs and digestive system. People with CF can experience:

- Frequent coughing or wheezing
- Serious lung infections, such as pneumonia
- Trouble digesting food and gaining weight
- Male infertility

There currently is no cure for CF and the average life span of all affected people is 37 years. However, there are treatments that can help people with CF feel better and live longer.



What causes cystic fibrosis?

Children genetically inherit cystic fibrosis from their parents. We each receive two copies of every gene: one from each parent. Children born with CF inherit altered or mutated copies of the CF gene from both parents. Those who receive only one mutated gene and one unaffected gene typically will not have CF, but will be CF carriers who can potentially pass the mutated copy of the gene down to their children. Although this may seem rare, more than 10 million Americans are CF carriers. So the odds that both parents are CF carriers are higher than you might think.

Who has the highest risk?

Two out of three people with a brother or sister that has the disease are at risk of being CF carriers. However, cystic fibrosis is most common in people whose families come from Northern and Western Europe. For Ashkenazi Jewish and Non-Hispanic Caucasians, approximately 1 infant out of every 3,000 births will be born with CF. While CF is less common in people of other ethnic groups, CF carrier screening is recommended for all women who are pregnant or planning to become pregnant.*

Remember, your child can get CF only if both parents pass down an altered gene.

*According to the American Congress of Obstetricians and Gynecologists.

What are the risk factors for being a CF carrier?

Risk Factor	Risk
Family history of CF	As high as 2 in 3
Ethnicity**	
Ashkenazi Jewish	1 in 24
Non-Hispanic Caucasians	1 in 25
Hispanic Americans	1 in 46
African Americans	1 in 65
Asian Americans	1 in 94

**This risk is based on ethnicity alone; it does not include risk from personal or family history.

Who should have cystic fibrosis carrier screening?

This is a decision that you and your partner must make. According to medical guidelines, testing is recommended for*:

- Those who are planning a pregnancy
- All couples who are currently pregnant
- Couples in which one partner has CF
- Individuals with a family history of CF, such as those who already have a child with CF or those who have a relative with CF

Talk to your doctor if you feel you should be tested. If your doctor agrees, he or she will order the test for you.

What is the CFvantage Cystic Fibrosis Expanded Screen?

CFvantage is a blood test that can tell you if you carry an altered gene that can cause cystic fibrosis. The test will also help you define your chances of having CF. Medical guidelines recommend that all couples who are planning a pregnancy or are pregnant should be offered a cystic fibrosis carrier screening test such as CFvantage.

While the CFvantage test is highly accurate, no test is perfect. It detects the most common alterations in the CF gene that can cause CF, but not all known defects.

Like all CF carrier tests, a negative CFvantage result does not 100% guarantee the birth of a healthy baby, but it can show that there is a low level of risk. If your result is positive, your doctor will discuss further testing options for your partner and/or your pregnancy.

What makes CFvantage a good choice for your family?

CFvantage can determine your risk of having a child with cystic fibrosis.

- **Deeper insights**—an expanded test panel detects more CF gene alterations to provide more comprehensive information
- **Timely**—allows you to make decisions even before you get pregnant
- **Trusted**—offered by Quest Diagnostics, a leader in genetic testing
- **Easy**—requires just a simple blood draw

Remember, CFvantage can only tell you your risk of having a child with CF. It cannot determine whether your child definitely will have CF.

Understanding your results

CFvantage detects the most common, but not all, genetic alterations that can cause CF.

If your test results are negative, you are at low risk for being a CF carrier and you are less likely to have a child with CF.

If your test results are positive, you are a CF carrier. There is a 50% (1 in 2) chance that you'll pass the altered gene on to your child. But your child will only be affected with CF if both you and your partner each pass down an altered gene.

If your partner's result is also positive, there is a 25% (1 in 4) chance that your child will have CF, a 50% (1 in 2) chance that your child will be a carrier but will not have CF, and a 25% (1 in 4) chance that your child will not be a carrier and will not have CF.

Your CF Test Results	Your Partner's CF Test Results	Explanation
Positive	Positive	Both you and your partner are both CF carriers. There is a 50% (1 in 2) chance that your child will be a carrier but will not have CF. There is a 25% (1 in 4) chance that both of you will pass the altered gene on to your child and your child will only be affected with CF. There is a 25% (1 in 4) chance that your child will not be a carrier and will not have CF.
Positive	Negative	You are a CF carrier, and your partner is not a CF carrier. There is a 50% (1 in 2) chance that you'll pass the altered gene on to your child. There is a 50% (1 in 2) chance that you will not pass the altered gene on to your child.
Negative	Positive	You are not a CF carrier, and your partner is a CF carrier. There is a 50% (1 in 2) chance that your partner will pass the altered gene on to your child. There is a 50% (1 in 2) chance that your partner will not pass the altered gene on to your child.
Negative	Negative	Neither you nor your partner were identified as a CF carrier. There is only a very remote chance that your child will have CF.

What family planning options are available?

If your CFvantage results are negative and you do not have any close relatives with CF, you can plan your family knowing that the probability of having a child with CF is very low.

If you and your partner are both CF carriers, you can choose from several options to prevent having a child with CF, such as IVF and adoption. You may want to learn what it's like to live with and take care of a child with CF, or learn about other pregnancy options such as:

- More testing like amniocentesis to provide information on the health of the fetus
- Adoption
- Using an egg or sperm from a donor who is not a CF carrier
- Procedures in which embryos are tested to ensure that only healthy ones are transferred to a woman's uterus

Your doctor or genetic counselor can help you learn more about the options and resources available and discuss the risks and benefits.



For more information about cystic fibrosis screening, talk to your healthcare provider.

This brochure is for informational purposes only and is not a substitute for medical advice, diagnosis, or treatment. The diagnosis or treatment of any disease or condition may be based on personal history, family history, symptoms, a physical examination, laboratory test results, and other information considered important by your doctor. Always talk with your doctor about the meaning of your test results and before you stop, start, or change any medication or treatment.

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