CYSTIC FIBROSIS

Carrier Testing

What You Need to Know

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What is cystic fibrosis?

Cystic fibrosis (CF) is a genetic condition that affects more than 30,000 people in the United States. Affected individuals are usually diagnosed in infancy due to a positive newborn screen, recurrent lung infections, and poor growth resulting from difficulty absorbing nutrients from food.

Is there a cure for CF?

Although there is no cure, treatment improves the length and quality of life of affected individuals by reducing lung damage and optimizing nutrition. CF symptoms and disease severity vary from patient to patient. The average life expectancy for an affected individual in developed countries is approximately 50 years.

Medications that target the specific disease-causing *CFTR* gene variants are available for some patients. Digestive problems may be treated with a high calorie, high fat diet, and vitamin, enzyme, and mineral supplements. Inhaled bronchodilators are used to maintain adequate airflow, and airway clearance techniques such as daily chest physical therapy are needed to help clear mucus from the lungs and prevent respiratory infections. Even with careful treatment, lung infections can develop, requiring antibiotics, aerosol inhalants, and hospitalization. Lung infections usually worsen over the course of life due to antibiotic-resistant bacteria. Treatments and hospital visits are costly.

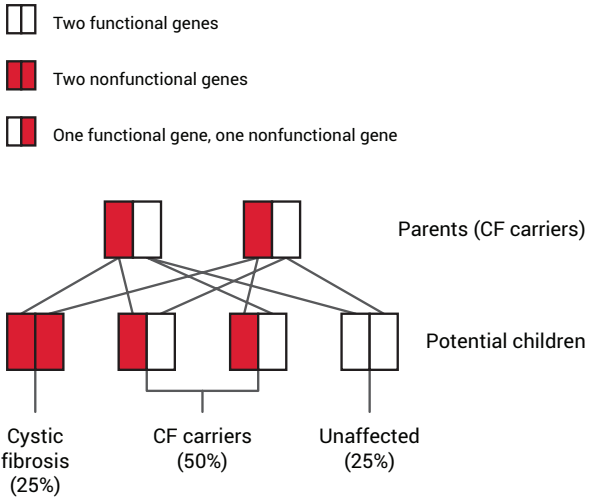
The leading cause of death in individuals with CF is respiratory failure from progressive lung damage.

Can my children have CF even if it is not in my family?

Yes. In fact, most couples who have a child with CF have no family history of CF and are surprised to learn that they carry a variant in the *CFTR* gene, which causes the condition.

Genes are the basic hereditary units determining an individual's traits, such as hair and eye color. CF carriers inherit a single nonfunctional gene from one parent, along with a functional gene from their other parent. Carriers of CF usually have no symptoms, as they have one normal copy of the gene. Both parents must be CF carriers to have an affected child.

CF Inheritance:



Who should consider carrier testing?

An estimated 10 million people in the United States are carriers of CF. Individuals with a relative who is affected with or a carrier of CF should consider testing. The American College of Medical Genetics and the American College of Obstetrics and Gynecology recommend that carrier screening be offered to women who are pregnant or planning a pregnancy.

Your chance of being a CF carrier depends on your ethnic background, unless someone in your family has CF or is a carrier. Assuming you and your partner are from the same ethnic group and have no family history of CF, the following chart shows the estimated chance of having a child with CF before testing, the test's detection rate, and the chance of having a child with CF if one partner has a normal test result.

Your chance of being a CF carrier depends on your ethnic background.

Ethnicity	Chance of Child with CF Before Test	CF Carriers Detected	Chance of CF Child after Normal Test Result in One Parent
Ashkenazi Jewish	1 in 2,300	96%	1 in 53,000
White	1 in 2,500	92%	1 in 30,000
African-American	1 in 15,100	78%	1 in 67,000
Hispanic	1 in 13,500	80%	1 in 66,000
Asian	1 in 35,100	55%	1 in 79,000

Some couples may decide against carrier testing if their ethnic group has a low risk of having a child with CF or because the test's detection rate is low for their ethnicity. Other individuals may forego testing due to a lack of insurance coverage or the potential anxiety it may cause.

Whether to have CF-carrier testing is a complex personal decision. Some couples may decide to undergo carrier testing to learn as much as possible about the health of their future child. The majority of couples that undergo testing will be reassured that their chance of having a child with CF is low. Screening identifies couples who are at high risk for having a baby with CF, allowing them to make informed decisions about prenatal diagnosis or testing after delivery.

What if my test results do not show a CF gene variant?

Like many medical tests, CF-carrier testing has its limitations. Even with a normal test result, a small chance exists that you could still be a carrier. There are certain rare changes in the *CFTR* gene that this test does not detect. However, if your test results are normal, you can be reassured your baby has a low chance of being affected with CF.

What if my test results show a *CFTR* gene variant?

If you are found to be a CF carrier, then your partner should also be tested. If one test result is normal, then you and your partner can be reassured your baby has a low chance of being affected with CF.

What if we both have a *CFTR* gene variant?

If both you and your partner are found to be CF carriers, then with each pregnancy there is a one in four (25%) chance of having a child with CF or three in four (75%) chance of having a child without CF. It is highly recommended that you and your partner meet with a genetic counselor who can thoroughly review your reproductive options.

Preimplantation genetic diagnosis may be performed prior to pregnancy to select only unaffected embryos for transfer. Adoption or donor sperm could also be considered. In already established pregnancies, prenatal diagnostic testing, such as amniocentesis or chorionic villus sampling, may be performed to determine if the pregnancy is affected. Prenatal testing can help a couple prepare for the birth of a child who may have special healthcare needs (approximately 10% of infants with CF have meconium ileus, a bowel obstruction requiring surgical repair in the newborn period) or determine if they wish to discontinue an affected pregnancy. Other couples choose not to proceed with prenatal diagnostic testing and instead test the baby after birth.

If you would like to learn more about CF carrier testing, please talk with your healthcare provider or contact a genetic counselor in your area (www.nsgc.org/page/find-a-genetic-counselor).

References

1. Watson MS, Cutting GR, Desnick RJ, et al. Cystic fibrosis population carrier screening: 2004 revision of American College of Medical Genetics mutation panel. *Genet Med.* 2004;6(5):387-391.

2. Cystic Fibrosis Foundation. www.cff.org (accessed on August 23, 2021).

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