

## Fact Sheet: Cystic Fibrosis (CF) Carrier Testing

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**Note:** The American College of Obstetrics and Gynecology and the American College of Medical Genetics recommend CF carrier testing for:

- *Individuals who have a partner with CF, or a family history of CF*
- *Couples in whom one or both partners are Caucasian and are planning a pregnancy or seeking pregnancy care*

Carrier screening is made available to couples in other racial and ethnic groups who may want testing.

### What is CF?

CF is an inherited disorder in which the body produces abnormally thick mucus, which accumulates in the lungs. In severe cases, this leads to chronic lung infections and pulmonary failure. Other problems include chronic diarrhea, poor growth and infertility. In severe cases, CF is present from birth. In mild cases, the symptoms sometimes do not appear until late childhood or even in the teenage years. The average live span of most individuals with CF is 20 – 30 years. With research and new medical treatment, it is hoped that the life span of those with CF will be increased.

### What causes CF?

CF occurs in a child who inherits two improperly functioning CF genes, one from each parent. Each of us has about 30,000 genes. The job of each gene is to manufacture a protein. Each protein has a specific purpose in the body. In CF there is an error in the gene causing no protein, or an abnormal protein to be made. Genes come in pairs. Individuals with one improperly functioning CF gene are called “carriers” of CF. Carriers of CF have no symptoms of CF. People with symptoms of CF have inherited two improperly working, or non-working CF genes; one from each parent.

### **How common is it to be a carrier of CF?**

The chance of being a carrier of CF depends on your ethnic and racial background. When one of your relatives has CF, then no matter what your ethnic or racial background your chance of carrying the CF gene is increased.

### **How can I find out if I am a carrier of CF?**

A special DNA test detects most carriers of the CF gene. Carrier testing is done as a blood test, or from cells taken with a brush from the inside of the cheek. However, the ability of the test to detect carriers is different for people of different ethnic and racial backgrounds.

The table below shows the chance of being a carrier in various racial and ethnic groups and the CF carrier detection rate for the different groups.

<b>Ethnicity</b>	<b>Chance of Being a Carrier</b>	<b>Carrier Detection Rate</b>
Northern European	1 in 29	90%
Southern European	1 in 29	70%
Ashkenazi Jewish	1 in 29	97%
Hispanic	1 in 46	57%
African American	1 in 65	72%
Asian	1 in 90	30%
Other backgrounds	Variable	70% (average)

### **What if my partner and I are carriers of CF?**

When both members of a couple are known to be carriers of CF, their chance of having a child with CF is 25% (1 in 4). Or, more optimistically, there is a 75% (3 out of 4) chance, with each pregnancy, that their child will not have CF. The chance remains the same if the child is a boy or a girl.

When both members of a couple have been tested and both are known to be carriers of CF, they may choose to use prenatal testing, (amniocentesis or chronic villi sampling), to find out whether their developing baby has CF. Carrier testing should be done early in pregnancy or if possible, before pregnancy. Genetic counselors, physicians and other health care providers can explain the details of carrier testing and prenatal diagnosis.

### **What if prenatal diagnosis shows that my baby will be born with CF?**

You will have the choice of using this information to prepare for a child with special health needs, or you can terminate pregnancy. Ask your physician, genetic counselor, or other health provider if carrier testing or prenatal diagnosis is appropriate for you.