

## How Can Genetic Counseling Help?

A genetic counselor is a specially trained healthcare professional who can help you make an informed decision about CF carrier testing. You can request to meet with a genetic counselor. The counselor will explain your specific risk to have a child with CF and discuss options for additional testing if you and your partner are both found to be carriers of CF gene mutations. A genetic counseling visit is usually covered by insurance.



## Who Do I Call for More Information?

Genetic counseling services are available through Greenwood Genetic Center's offices in Greenwood, Greenville, Spartanburg, and Columbia. If you have any questions, please call the office closest to where you live and ask to speak to a genetic counselor.

### **Greenwood**

1 Gregor Mendel Circle  
Greenwood, SC 29646  
1-888-442-4363 (toll free)  
(864) 941-8100

### **Greenville**

2 Doctors Drive  
Greenville, SC 29605  
1-866-478-4363 (toll free)  
(864) 250-7944  
or (864) 455-1600 for prenatal counselors

### **Spartanburg**

Regional Maternal-Fetal Medicine  
863 North Church Street, Suite 610  
Spartanburg, SC 29303  
(864) 560-1615

### **Columbia**

529 Richland Street  
Columbia, SC 29201  
1-800-679-5390 (toll free)  
(803) 799-5390

This brochure is published by Greenwood Genetic Center, a nonprofit institute organized to provide clinical genetic services and laboratory testing, to develop educational programs and materials, and to conduct research in the field of medical genetics.

## CYSTIC FIBROSIS Carrier Testing

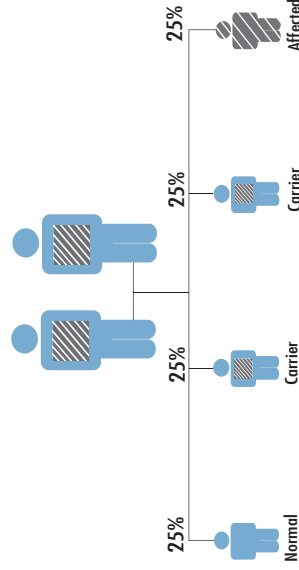


## What is Cystic Fibrosis?

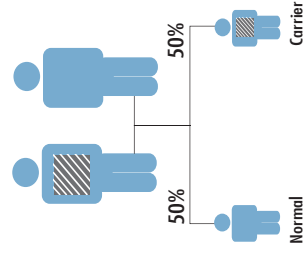
Cystic fibrosis (CF) is one of the most common inherited diseases in the United States. It is characterized by problems with breathing, digestion, and fertility. Some individuals with CF will have mild symptoms and others will have life-threatening illness. Although treatment for CF has improved, there is no cure for this disease. CF does not cause learning problems or physical birth defects.

## How is CF Inherited?

CF is an autosomal recessive genetic disease. In order for a child to be affected with a recessive genetic disease, both parents must be carriers of a disease causing gene mutation. Carriers have one normal CF gene and one CF gene mutation. CF carriers typically do not have any symptoms of the condition. When both parents carry a CF gene mutation there is a 25% chance that a child will be affected with CF, a 25% chance that a child will get no CF gene mutations, and a 50% chance that a child will be a carrier like the parents.



When only one parent carries a CF gene mutation, there is a 50% chance for the child to be a carrier, but no chance for the child to be affected with CF.



  
**Affected Individual**

  
**Normal Individual**

  
**Carrier**

## Who Could Be a Carrier for CF?

A person of any racial or ethnic background can be a carrier of CF. However CF is most common among Caucasians (people with European ancestry). The chance of being a CF carrier based on ethnic background is shown at the bottom of the page.

The chance of being a CF carrier is higher if there is a family history of the disease. A genetic counselor or physician can determine the specific chance for you to be a CF carrier based on your family history.

## Who Should Have CF Carrier Testing?

The National Institutes of Health, the American College of Obstetrics and Gynecology, and the American College of Medical Genetics recommend that all couples planning a pregnancy or seeking prenatal care be offered CF carrier testing. It is especially important for couples to consider testing when at least one member of the couple is Caucasian. Individuals with a family history of CF should also consider CF carrier testing. Some couples will be offered CF carrier testing because prenatal ultrasound results suggest an increased risk for CF. CF carrier testing is optional.

## How is the CF Carrier Test Done?

The CF carrier test requires drawing a blood sample from the arm. The blood sample is sent to the laboratory for gene mutation (DNA) analysis. Test results are generally available within 1 week. There are hundreds of different mutations in the CF gene. The Greenwood Genetic Center tests for the most common mutations. A person who has one mutation is a carrier for CF. Since there are very rare mutations in the CF gene, a negative test result does not guarantee that a person is not a carrier for CF. A negative test result means that the chance for that person to be a carrier for CF is very low. Prenatal diagnosis through amniocentesis is available if both parents are carriers of a CF mutation.

## How Do I Decide if I Want CF Carrier Testing?

Choosing to have CF carrier testing is a very important and personal decision. Take time to discuss this information with your partner and your health care provider. Some points to consider are:

- Do you think CF is a serious condition?
- Do you feel that your chance to be a CF carrier is high or low?
- Would you consider prenatal diagnosis if your baby is at risk for CF?
- Will insurance cover testing? Although CF carrier testing is recommended by the American College of Obstetricians and Gynecologists, all insurance plans may not cover this test so you should check with your insurance representative.

## ESTIMATED CARRIER RISK

Racial or Ethnic Group	Risk Before Test	Detection Rate	Risk After Negative Test
Northern European Caucasian	1/29	90%	~ 1 in 250
Ashkenazi Jewish	1/29	97%	~ 1 in 930
European Caucasian	1/29	80%	~ 1 in 140
Hispanic American	1/46	57%	~ 1 in 105
African American	1/65	69%	~ 1 in 207
Asian American	1/90	Not Available	Not Available

Adapted from ACOG and ACMG Preconception and Prenatal Carrier Screening for Cystic Fibrosis: Clinical and Laboratory Guidelines 2001