



California Pacific
Medical Center

A Sutter Health Affiliate

Community Based, Not For Profit

Carrier Testing for Cystic Fibrosis Prenatal Diagnosis Center

What is Cystic Fibrosis?

Cystic fibrosis is one of the most common inherited diseases. About 1 in 3200 Caucasians (Whites) in the United States has cystic fibrosis, which means 1 in 29 Caucasians are carriers of this condition. Although it is less common in people of other ethnic or racial backgrounds, you may still wish to consider carrier testing even if you are only part Caucasian or if your partner is not Caucasian.

Cystic fibrosis causes the body to produce large amounts of abnormally thick mucus. This mucus collects in the lungs leading to congestions and pneumonia. Mucus may also collect in the intestines, resulting in diarrhea and poor growth. Treatment for these problems often requires hospitalization for one or two weeks at a time. Although cystic fibrosis is present from birth, the symptoms may not appear until childhood or in some people not until teenage or adult years. Cystic fibrosis does not affect intelligence. There is wide variability in the severity of the disease. There is no cure for cystic fibrosis at this time. Scientists are, however, making progress in improving treatment and searching for a cure. In the past, people with cystic fibrosis died in childhood, but now many are living past 30 years of age.

What Causes Cystic Fibrosis?

This is a genetic disease, not an illness you can catch from other people. It is caused by a change in our genes. Genes are packages of information that act like blueprints for making the proteins that work in our bodies. A change in genetic information can result in a gene that does not work properly either. A person who has one non-working gene and one normal gene is called a carrier. A carrier does not and never will have the disease but could pass the non-working gene to their child. The *disease* occurs as a result of having two non-working genes which code for the specific condition. Individuals who have cystic fibrosis disease inherited on non-working cystic fibrosis gene from each parent.

How is Cystic Fibrosis Inherited?

Cystic fibrosis is inherited as an autosomal recessive condition. This means a child can only have the disease if both parents are *carriers*. Carriers have no symptoms; rather they carry a genetic change (referred to as a “mutation”) that may be passed on to future generations. Although recessive diseases are inherited, there is usually no prior family history when a child is born with the disease. Therefore, because the disease is not in your family does NOT mean that you are not a carrier or could never have an affected child. If, however, there is a history of the disease in your family, the chance of having a child with the disease may be significantly higher.

What is My Chance to be a Cystic Fibrosis Carrier?

If no one in your family has cystic fibrosis, the chance that you are a carrier is based on your ethnic group:

Caucasian	1 in 29
Hispanic	1 in 46
African American	1 in 65
Asian-American	1 in 90

What Happens if We Are Both Carriers?

If you and your partner are both carriers of a cystic fibrosis gene mutation, prenatal diagnosis is available to determine whether the baby has the disease. This testing can be performed using cells from the chorionic villus sampling (CVS) or amniocentesis. If both members of a couple are carriers of a cystic fibrosis disease gene, in each pregnancy there is a 25% chance of having a child with the disease, and a 75% chance of having a child without the disease.

What is the Cystic Fibrosis Carrier Test?

Carrier testing involves taking a small blood sample. The laboratory isolates DNA, the chemicals that compose genes, and test for specific genetic mutations connected with the disease. Many but not all of the mutations connected with the disease have been discovered through research.

How Accurate is the Testing?

If you are found to be a carrier, the result is highly accurate. If you are 100% Caucasian, the DNA blood test can tell with ~90% certainty if you are a carrier for the cystic fibrosis gene. Because not all of the DNA mutations have been discovered, a negative test result means there still a chance that you could be a carrier. The detection rate is lower if you are only partially Caucasian or of non-Caucasian ancestry. The detection rate may also vary depending upon the number of mutations tested for by the laboratory. If you are interested in carrier testing for cystic fibrosis, you can discuss these issues further with your genetic counselor or health care provider.

Will My Health Insurance Pay for These Tests?

Each insurance policy is different. Your insurance company may have a contract with a particular laboratory. You may wish to check with your insurance carrier to determine if they will pay for this test. Depending on the laboratory used, carrier testing costs between \$150-\$300.

_____ YES, I would like more information about cystic fibrosis carrier testing.
Initial

_____ NO, I do NOT want more information about cystic fibrosis carrier testing. I
Initial understand the above information and decline testing.

_____ I have already been screened for cystic fibrosis. My results are _____.
Initial