



What is Cystic Fibrosis

Cystic fibrosis is a genetic disease that affects about 1 in 3300 people in the United States. Symptoms of the disease range from mild to severe. Most often, people with CF have chronic lung problems that will continue to worsen over time. CF may also affect digestion resulting in chronic diarrhea and poor weight gain and growth. There are some treatments available for the lung and digestive problems, but most people with CF have a shortened lifespan. Most people do not reach adulthood with CF. Men are almost always infertile. CF does NOT affect intelligence.

What is a CF carrier?

Each person has two copies of a gene, one inherited from each parent. If a person has one normal CF gene and one abnormal CF gene, that person is a carrier of the disease and, therefore, does not have symptoms of the disease, but may be able to transmit this risk to offspring, including the disease. This would happen if both parents were carriers of the same abnormal gene. In this scenario, the risk of disease would be 25% (1 out of 4). The risk of carrier state would be 50% (2 out of 4) and the risk of complete normal genes, would be 25%.

Can anyone be a CF carrier?

Yes. Your ethnic background can determine your carrier state risk. CF is more common in people of Caucasian and Ashkenazi Jewish descent. CF is less common in Hispanic, African American, Native American, or Asian backgrounds. If someone in your family has CF, then your risk of being a carrier is increased above what your risk is based on ethnic background.

What is the purpose of carrier testing?

The purpose of testing is to see if a couple is at increased risk for giving birth to a child who will have CF. Carrier-testing is a laboratory test on blood or saliva, used to show if a couple is at high risk. Additional testing on the developing baby may then be necessary.

If my test is negative, could I still be a carrier?

The answer to this is "yes." There are some mutations of the CF gene that the current test cannot find. Currently the test includes a minimum of 25 specific CF gene mutations, most of which are common in the higher risk ethnic groups. Like most medical tests, there are limitations because not all the CF mutations are known. However, these other mutations are rare.

INFORMED CONSENT:

I understand that the decision to be tested for CF carrier status is completely mine.

I understand that the test does not detect all CF carriers.

I understand that if I am a carrier, testing the baby's father will help me learn more about the chance that my baby could have CF.

I understand that if one parent is a carrier, and the other is not, it is still possible that the baby will have CF, but the chance of this is very small.

I understand that if both parents are carriers, additional testing can be done in order to know whether or not the baby will have CF.

I understand that if the baby has inherited a changed CF gene from each parent, that the baby will have the disease Cystic fibrosis, and further genetic counseling and recommendations from a perinatal center will be offered.

I have read and understand the information presented to me regarding CF testing.

I do not want CF carrier testing.

I want CF carrier testing.

Signed: _____

Date: _____

Witness: _____

Date: _____

(Please note that insurance coverage for this test is dependent upon your own individual insurance policy.)