

# Cystic Fibrosis Carrier Screening

## **What Is Cystic Fibrosis?**

Cystic Fibrosis (CF) is a disorder that causes the body to make a thick, sticky mucus that clogs the pancreas and lungs, most often leading to problems with breathing, infections, digestion and fertility. This disease can affect males or females. Thirty years ago, most babies born with cystic fibrosis died in early childhood, but advances in diagnosing and treating the disease have significantly improved the outlook for people born with CF. Even though there is no cure for CF, more than 60% of babies born with cystic fibrosis reach adulthood.

## **What causes CF?**

CF is a genetic disorder. People with CF have inherited two nonworking, or mutated, copies of the CF gene; one from each parent. When both copies of the CF gene do not work, that person produces a thicker mucus in the lungs and pancreas, which leads to the symptoms of the disease.

## **What is a carrier of CF?**

A carrier of CF is someone who “carries” one copy of the faulty CF gene. CF carriers are not sick and do not have CF. Genes do not change in one’s lifetime. Therefore a carrier will always be a carrier, but will NEVER get sick with CF. Carriers are at risk to have a child with CF.

## **How could my baby have CF?**

To have CF, a baby must inherit two nonworking copies of the gene, one from each parent. The disease can only occur in babies with two carrier parents. When both parents are carriers, they have a 25% chance with each pregnancy of having a baby with CF. When both parents are carriers, prenatal testing, by either amniocentesis or chorionic villus sampling, can be done if the couple wants to know, during the pregnancy, whether the fetus is affected or not. Details about these options would be discussed with a genetic counselor. If one parent is a carrier and the other parent is not a carrier, the couple is at risk to have a child that is a carrier, but they are not at risk to have a child that is affected with CF.

## **What is the chance that I am a CF carrier?**

Your chance of being a carrier depends on your family history and/or your ethnicity. Your chance is highest if you have a close relative with CF. Even without a family history of this disease, you could be a carrier. If you are of Caucasian and/or Jewish, there is about a 4% (1/25) chance that you could be a carrier. If you are Asian, Latin American, Native American and/or African American, your chances are lower.

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

CF carrier testing is a blood test that looks at your DNA to see if you “carry” a nonworking copy of the gene involved with CF.

## **Is CF carrier testing recommend?**

The American College of Obstetrics and Gynecology (ACOG) recommends that information about CF carrier testing be offered to all women prior to or early in pregnancy, regardless of race or ethnicity and family history. It is a personal choice to pursue CF carrier testing and only you and your partner can decide if this test is right for you.

**Why would I want to have CF carrier testing?**

Some people want to know if they have a higher chance of having a baby with CF. If that chance is higher, they may decide to pursue further testing to find out for certain if the baby has CF during the pregnancy so they can plan and prepare. Others may want to know that the risk is higher, but will decided to pursue further testing after their baby is born.

**Why would I NOT want to have CF carrier testing?**

Some people do not want to know during a pregnancy if they have a higher chance of having a baby with CF because this information would not be helpful for them at that time. All babies are screened for CF at birth as part of the Newborn Screen.

**What if I find out during my pregnancy that my baby has CF?**

When a baby is found to have CF during the pregnancy, the family is provided with more information about CF, including the option to meet with specialists and/or other families that have a child with CF. Some families choose to continue the pregnancy and other parents may choose to end the pregnancy if they find out their child has CF. These are personal decisions and both options are supported.

**How accurate is CF carrier testing?**

It depends on your ethnicity or race. It is most accurate for people who are Caucasian and/or Jewish. It is least accurate for Hispanic Americans and Native Americans. A genetic counselor can explain in more detail how accurate the test will be for you. A negative test results significantly reduces your chance to be a carrier, but it never eliminates the chance.

**When should I have CF carrier testing?**

CF carrier testing can be completed any time, although the best times are before pregnancy or within the first trimester of pregnancy.

**Will my insurance pay for CF carrier testing?**

Most insurance companies cover the cost of the test, however it is recommended that you check with your own insurance company.