



Cystic Fibrosis Carrier Screening

◆What is Cystic Fibrosis?

Cystic Fibrosis (CF) is a genetic disease that occurs most often in people whose ancestors came from Northern and Western Europe and people of Ashkenazi Jewish descent. In Northern European Caucasians and people of Ashkenazi Jewish descent, approximately one infant out of every 3,300 live births will be born with CF. People of other ethnic groups also have CF, but not as frequently. For example, one in 8,400 Hispanic Americans, one in 14,400 African Americans and only one in 32,400 Asian Americans will be born with CF.

Individuals who have Cystic Fibrosis have high levels of sodium and chloride (salt) in their sweat. Thick sticky mucus in the lungs causes persistent coughing, wheezing, and frequent lung infections, including pneumonia. Some affected children have difficulty gaining weight because they have low levels of pancreatic enzymes, which are needed to break down food for growth. In males, infertility may result from congenital absence of the vas deferens, the tube that carries sperm from the testes before being released from the body, also called CBAVD.

Although not curable, some treatments may increase the life span of patients with CF. High protein, high calorie, vitamin-rich diets, and intake of pancreatic enzymes increase physical growth. Physical therapy aids in clearing mucus from the lungs. Antibiotics and other medications are also used. However, despite treatment, half of people born with CF die before age 30, primarily from lung disease.

◆What is a Cystic Fibrosis Carrier?

People who receive one altered CF gene and one normal gene are called CF carriers and do NOT have the disease. Carriers have a 50% chance of passing the altered CF gene on to their child. In order for a child to have the disease, he or she must receive one altered gene from both parents. A child cannot have CF when only one parent is a carrier.

◆What is Cystic Fibrosis Carrier screening?

Inherited or genetic diseases like Cystic Fibrosis (CF) are passed from parents to their children. Cystic Fibrosis can occur even when neither parent has the disease. For an individual to have the disease, both parents must carry an altered CF gene. Cystic Fibrosis carrier screening lets you know what your risk is for carrying an altered gene, as well as your chance of having a child with CF. Additional testing is needed to determine if your child will have CF.

◆Who should have Cystic Fibrosis Carrier screening?

This is a decision that you, your partner, and physician must make. Carrier screening is generally offered to couples if one partner has CF or to individuals that have a family history of CF. Additionally, carrier screening is offered to non-Jewish Caucasians and Ashkenazi Jewish people. Individuals in both these groups have a 1 in 29 chance of being a carrier. Other ethnic groups have lower risks. For example, Hispanic Americans have a 1 in 46, African Americans a 1 in 60 and Asian Americans a 1 in 90 chance of being carriers based on ethnicity alone; it does not include risk from personal or family history.

◆How is Cystic Fibrosis Carrier screening done?

Over 900 changes have been discovered in the CF gene; however, the screening test is better at finding CF carriers in some ethnic groups than in others. For example, in Northern European and Ashkenazi Jewish people, 90-97% of the changes will be detected, while in Asian Americans only 30% will be detected. CF carrier screening is performed on a small sample of blood. The laboratory will find out if you carry one of the more common changes in the CF gene. Information about your race, ethnicity, and personal family history of CF is essential for correct interpretation of results.

◆What does a negative screen mean?

Cystic Fibrosis carrier screening provides information that modifies your “pre-test” risk. A negative screen test means that no change in your CF genes was detected. Your “post-test” risk of being a CF carrier is lower than your pre-test risk; you are less likely to have a child with CF. Since the screening test detects only the most common changes in the CF gene, a negative screen does not guarantee that you are not a carrier.

◆What does a positive screen mean?

A positive screen means that the laboratory found a change in one of your two CF genes and that you are a carrier. There is a 50% chance that you will pass this

gene on to your child. The chance that your child will have the disease depends upon whether your partner is also a carrier or has Cystic Fibrosis.

If your partner is also a carrier, then your child has a 25% chance of having CF. There is a 50% chance that your child will not have CF, but will be a carrier. Finally, there is a 25% chance that your child will not even be a carrier.

If your partner does have Cystic Fibrosis, then there is a 50% chance that your child will have CF. There is also a 50% chance that your child will be a carrier and not have CF.

If your partner has a negative CF carrier screen and yours is positive, the only way your child could have CF is if your partner has a rare change in one of his or her CF genes that was not detected in the screen. However, there is a 50% chance that your child will be a CF carrier.

Remember that the risks described above will be the same for each child you and your partner conceive. Also remember that CF carrier screening can only tell you the risk of your child having CF, not whether he or she will actually have CF.

◆What options are available?

If you and your parents are both CF carriers, you may want to know about medical procedures that can tell whether an unborn child will have the disease. Chorionic villi sampling (CVS) and amniocentesis are medical procedures to sample cells from the placenta or fluid surrounding the baby. These cells can be tested for the parents' altered genes. Other options such as egg or sperm donor and adoption should be discussed with your doctor or genetic counselor.

◆Possible reasons to be tested

- If CF seems like a very serious disorder to you
- If the chance of being a CF carrier seems high to you; this may be especially likely if a member of your family or your partners family has CF or is a known carrier
- If you and the baby's father would consider amniocentesis or Chorionic villi sampling (CVS)-to help you decide about continuing the pregnancy or to help you prepare for the birth of a baby with CF, if you were both found to be carriers

◆Possible reasons not to be tested

- If CF does not seem like a very serious disorder to you
- If the chance of being a carrier seems low to you; this may be especially likely if you are Asian American or African American
- If you and the baby's father would never consider having amniocentesis or CVS-to help you decide about terminating the pregnancy or preparing for the birth of a baby with CF, even if you were both found to be carriers

- Because the test is not perfect and will not identify all carriers
- Because the cost of testing may not be covered by your insurance company

◆ In Summary...

Cystic Fibrosis carrier screening

- Determines your risk for carrying an altered gene that can cause CF
- Determines your risk for passing that gene on to your child
- Determines your child's risk of having CF if both you and your partner are screened

A negative screen means that

- None of the common CF gene changes were found in your blood sample
- You are much less likely to pass an altered CF gene onto your child
- Remember that a negative screen does not guarantee you are not a carrier and cannot pass on an altered CF gene to your child.

A positive screen means that

- You are a CF carrier
- You might pass the altered CF gene onto your child
- Your child might have CF if your partner is also a carrier
- CF carrier screening cannot tell you for sure if you will, or will not, have a child with Cystic Fibrosis. Carrier screening will, however, give you important information that will help you make the best possible decisions for you and your family.