CYSTIC FIBROSIS

Cystic fibrosis carrier testing is being made available to you on a voluntary basis. Testing can be right for some people and not right for others. Whether or not you are tested is a personal decision that belongs to you and your baby's father.

Cystic fibrosis (CF) is a life-long illness that is usually diagnosed in the first few years of life. The disorder causes problems with digestion and breathing. Cystic fibrosis does not affect intelligence or appearance.

The digestive problems can usually be treated by taking medicine daily. To treat lung problems, most children with CF need to have respiratory therapy for about a half hour every day; this helps clear mucus from the lungs. This is something that parents or other family members can do at home. Sometimes lung infections still develop. The children may need to be treated with antibiotics at home or in a hospital. However, the infections tend to become worse over time and more difficult to treat. Treatments are costly and may be burdensome without adequate health insurance.

Some individuals have milder or more severe symptoms than others for reasons that are not completely understood. It is not always possible to tell from a prenatal test how mild or severe a child's symptoms will be. In general, people with CF have a shortened life span, some die in childhood, and others live into their 40s or even longer. Although there is no cure for CF, research on more effective treatments is under way. Still, by adulthood, most people with CF will have some breathing and digestive problems. Despite these physical problems, there are many people with CF who attend school, have careers, and have fulfilling lives.

The purpose of CF carrier testing is to see if a couple is at increased risk for giving birth to a child who will have CF. Cystic fibrosis carrier testing is a laboratory test done on a sample of blood or saliva. If testing shows that a couple is at high risk, additional testing can be done on the developing baby to see whether or not it will have CF. However, most women's test results are normal.

Cystic fibrosis cannot be treated before birth. The purpose of having this information about your developing baby is so you can prepare yourself to care for a child with special health care needs or so you can terminate the pregnancy.

Cystic fibrosis is a genetic disorder. All genes come in pairs, so everyone has two copies of each gene. One copy comes from your mother and the other from your father. Some genes do not function properly because there is a mistake in them. If a gene has a mistake, it is said to be altered or changed. For some diseases-like CF-both genes of the pair have to be altered for a person to have the disease. If a person has one changed copy of a CF gene, that person is a carrier for CF. A carrier does not have CF. There are no known health problems associated with being a carrier. If a person has two changed copies of the CF gene, they will develop CF. When both partners in a couple are carriers, any child they have has a 1-in-4 (25%) chance to inherit a changed copy of the gene from each parent. A child with two changed copies of the CF gene will develop CF.

You could be a carrier of CF even if no one in your family has CF and even if you already have children without CF. About one of every 30 white people (about 3 in 100 or about 3%) carries the changed gene. If your family background is not white, your chance of being a carrier is less than 1 in 30. For example, some Asian-American groups have carrier rates of 1 in 90. Check the table below to see your chance of being a carrier according to your ethnic background. If a relative of yours has CF, or is known to be a carrier of CF, your chance of being a carrier is greater based on your family history than your ethnic background.

The Chance of Being a CF Carrier Depending on Race/Ethnicity

Ethnicity/Race	Chance of Being a CF Carrier	Chance Both Partners Are CF Carriers
European Caucasian, Ashkenazi Jewish	1 in 29	1 in 841
Hispanic American	1 in 46	1 in 2,116
African American	1 in 65	1 in 4,225
Asian American	1 in 90	1 in 8,100

NOTE: If your ethnic/racial group is not listed above, please ask your provider for this information. Also, you may want to mention your ethnic/racial group to your provider to learn about prenatal tests for diseases other than CF.

REMEMBER: Both parents must be carriers for the baby to develop CF.

There are some mutations in the CF gene that the current test cannot find. For this reason, you could be told your test result is normal and you could still be a carrier. Like most medical tests, this one has limitations because not all CF mutations are known. However, these unknown CF mutations are rare. The likelihood that you are a carrier even though you had a normal result is very small.

If the test shows that you are a carrier, the next step is to test the baby's father. Both parents must be carriers for the baby to have CF. If the father has a normal test result, the chance that your baby will have CF is very, very small. This remaining risk is because the test is not 100% accurate.

If two people who are both carriers have a child, that child may have CF. When two carriers have a child together, there is a 1-in-4 (25%) chance with each pregnancy that the child will have CF. This is true even if they already have other children with-or without-CF. If CF testing shows both parents are carriers, you might then see a provider for genetic counseling. This person could give you more information and help you decide if you want to test the baby for CF. This could be done around the 11th week of pregnancy using CVS (chorionic villus sampling). This involves removing a tiny piece of the placenta. Or it could be done around the 16th week of pregnancy using amniocentesis, a procedure where a needle is used to take fluid from around the baby for testing. *If either test shows that the baby will develop CF, you could choose to either terminate or continue the pregnancy. *CVS and amniocentesis may cause a miscarriage about 1 out of 200 test due to having the procedure done.

If the test shows you are a carrier, the result is definite and will not change. If you test negative now and become pregnant in the future, you should discuss CF carrier testing at that time with your provider, as test technology changes.

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 partner's family has CF or is a known carrier.
- If you and the baby's father would consider amniocentesis or 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 CF 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

Resources to learn more:

1. Cystic Fibrosis Foundation
   1-800-FIGHT CF (1-800-344-4823)
   www.cff.org
   E-mail: info@cff.org

2. National Society of Genetic Counselors
   1-610-872-7608 Press 7
   www.nsgc.org Click on ResourceLink

3. Genetic Alliance
   1-800-336-4363
   www.geneticalliance.org
   E-mail : info@geneticalliance.org

_____I want CF carrier testing
_____I do not want CF carrier testing

Signed:_____ Date:_____