

## Cystic Fibrosis – Prenatal Screening and Diagnosis

### What is cystic fibrosis (CF)?

Cystic fibrosis (CF) is a genetic disorder.

- ❖ It is caused by an abnormal gene that is passed from parent to child and is a lifelong illness that affects all of the organs of the body and often causes problems with digestion and breathing.
- ❖ It does not affect a person's looks or mental ability.
- ❖ In some cases, CF poses a serious risk to a person's health and shortens the life span.
- ❖ Despite their physical problems, many people with CF attend school, have careers, and lead full lives.

### What causes CF?

Cystic fibrosis is a recessive disorder.

- ❖ In a recessive disorder, **both parents** must carry a copy of the abnormal gene for the problem to occur in their child.
- ❖ A person who has one copy of an abnormal gene for a recessive disorder is a **carrier** for that disorder, even though he or she may show no signs of it.
- ❖ If both parents are carriers, each of their children has a 25% chance of having the disorder.
- ❖ Put another way, this couple has a 1-in-4 chance of having a child with CF.

### What are the symptoms of CF?

The symptoms of CF can vary in type and severity.

- Many people with CF produce a thick, sticky mucus in their bodies.
- This mucus builds up and clogs the lungs, which makes it hard to breathe, and can lead to infection.
- It also can affect the digestive organs, making it hard for the body to break down and absorb food.

### Is treatment available for CF?

New drugs and treatments have improved the outlook for people with CF, but it is still a lifelong disease. Your pediatrician will guide you and there is a large CF community for

information and support.

## What are risk factors for CF?

The risk of being a CF carrier is higher in certain races and ethnic groups. It occurs more often in white people than in other racial groups. The risk also is increased in families with a history of CF.

## Can I be tested to assess whether I am a CF carrier?

Carrier testing can be done for couples planning a pregnancy or during pregnancy to assess their risk. The test is done on a blood sample and carrier testing is available to all pregnant women. If testing shows that a couple is at high risk, more testing can be done during pregnancy to see whether their fetus has CF.

## What does it mean if test results for one partner are negative?

If your test results are negative, the chance that you are a CF carrier is small. There are some rare CF gene defects that the test does not detect. For this reason, you could be told your test result is negative, and you could still be a carrier. Although this is very rare.

## What does it mean if test results for one partner are positive?

If the test results show that one partner is a carrier, the next step is to test the other partner. Both parents must be CF carriers for the baby to have CF.

If one parent has a negative test result, the chance that the baby will have CF is small. Because the risk is small, if one partner is a carrier but the other has a negative result, no further testing is recommended.

If the father is not available for a carrier test, a genetic counselor may be able to help you decide whether to have prenatal testing of the fetus to see if it has CF.

## What does it mean if test results for both partners are positive?

If two people who are both CF carriers have a baby, there is a 25% chance that the baby will have CF. However, it is more likely that the baby will be a carrier, like the parents, and will have the gene but will not have the disease. It also is possible that although the parents are both carriers, the baby will not be a CF carrier.

## If both partners are positive, what follow-up tests are appropriate and what do they assess?

If both partners are CF carriers, further prenatal testing can be done to see if the baby has CF. This testing is not recommended when only one partner is a carrier. Parents may want to know if the baby will have CF so that they can prepare for the care of a child with special health care needs, or they may choose to end the pregnancy.

## What prenatal tests can be done to detect CF and other disorders?

Prenatal tests done to detect CF and other disorders are chorionic villus sampling (CVS) and amniocentesis. CVS can be performed after 9 completed weeks of pregnancy.

Amniocentesis can be performed between 15 weeks and 20 weeks of pregnancy.

## What should partners who are CF carriers be aware of when thinking about future pregnancies?

If a test result shows that you are a CF carrier, the result is definite and will not change. If both partners are carriers, it means that in each pregnancy the baby will have a 25% (1-in-4) chance of having CF. If you want to know whether your baby will have CF, you will need to have amniocentesis or CVS in each pregnancy. Other options include the following:

- Adoption
- Using donor sperm or donor eggs (but the donor should be tested for CF carrier status)
- Using in vitro fertilization with your own sperm and eggs, and then using preimplantation genetic diagnosis to see if the fertilized egg has CF or is a CF carrier.