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Cystic Fibrosis Testing

What is Cystic Fibrosis?

Cystic fibrosis 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.

What is the Purpose of Cystic Fibrosis Carrier Testing?

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.

If My Test Result Is Normal, Could I Still Be a Carrier?

Yes. 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 I Am a Carrier, What Should I Do?

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 percent accurate, as mentioned in the previous section.

However, since this is a very rare occurrence, if you are a carrier but the father has a normal result, no further testing would be recommended.

What if Both My Partner and I Are Cystic Fibrosis Carriers?

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 percent) 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.

How Do I Decide Whether or Not to Have Carrier Testing?

After learning about CF carrier testing, some people decide to have testing, and others decide against it. The cost of testing is covered by some insurance and not by others. You may want to check with your insurance company before deciding if you want testing.

Listed as follows are some reasons other people have given for having or not having CF testing.

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
- Because test results are usually reassuring
- Because the cost of testing is covered by your insurance company

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

FOR MORE INFORMATION, CONSULT YOUR PHYSICIAN.