FERTILITY ASSOCIATES of MEMPHIS
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Genetic Testing for Cystic Fibrosis

Cystic fibrosis (CF) is a common inherited disorder that affects approximately 1 in 2500 Americans. In this disease, regulation of ion balance in the body's secretory glands is affected resulting in chronic respiratory, digestive, endocrine, and infertility problems. CF does not cause mental impairment or affect appearance. This chronic illness requires multidisciplinary medical management, and symptoms range from mild to life threatening.

Because cystic fibrosis (CF) is one of the most common genetic disorders, the American College of Obstetrics and Gynecologists (ACOG) recommends health care providers offer genetic screening to all couples who are pregnant or who are interested in pregnancy. Testing is available for couples of all ethnic backgrounds; however, Caucasians, especially those of European or Ashkenazi Jewish descent, are the groups who are considered at higher risk. Also, men who have absence of sperm should be offered screening as this may be their only manifestation of CF. A person can be a carrier of CF even if he/she does not have the disease or has no family history of CF. Approximately 1 in 29 Caucasians is a genetic carrier, which means they have one copy of abnormal genetic mutation. When both parents are carriers of CF, there is a 25% chance with each pregnancy that the child will inherit both copies of the abnormal mutation and develop CF.

The CF carrier test is a blood test that assesses the most common genetic mutations in the CF gene (CFTR). A negative CF screening test confirms that one is not a carrier of these most common mutations, although it does not completely rule out the possibility of being a carrier of a rare mutation. A positive screen means the person is a carrier of CF, and we will recommend testing the partner. If the partner has a negative test result, the chance of having a baby with CF is less than 1/100. If both partners are carriers, the couple has a 25% chance (1 in 4) of having an affected child with CF. Genetic counseling will be recommended for at-risk couples to further discuss options such as preimplantation genetic diagnosis at time of in vitro fertilization or prenatal diagnosis during pregnancy.

Cystic fibrosis screening is optional. The decision whether to be tested or not is a personal decision that belongs to you and your partner. **Please check with your health insurance company to know that the cost of the CF test is covered.** If you have questions about testing, please discuss them with your provider.

References

1. American College of Obstetricians and Gynecologists. News Release. 2003. www.acog.org
2. Brown, T. and Pratt, VM. "Cystic Fibrosis: An Update." Advance/Laboratory. March 2002.
3. "Cystic Fibrosis." Laboratory Corporation of America Holdings. 2002.
4. "Cystic Fibrosis Carrier Screening Test: Helping You Make an Informed Decision." Quest Diagnostics. 2001.

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