

Cystic Fibrosis Facts

Name _____

Date of Birth _____ Identification number _____

- Cystic fibrosis (CF) is a common genetic disorder in Caucasians. It is present in other ethnic groups as well but is less common.
- CF causes lung problems. Children with cystic fibrosis get inflammation and infections in their lungs.
- CF causes digestive problems in about 85% of people. Lack of enzymes from the pancreas (pancreatic insufficiency), which aid digestion, can cause poor absorption of food.
- CF symptoms are highly variable, even in the same family. Some children may have very mild disease, some may have significant disease. Knowledge of specific mutations (gene changes) does not assist in predicting severity of disease.
- The median age of survival with current medical therapies is about 30 years although the median age of survival is longer in people without pancreatic insufficiency.
- CF occurs in a child only when both parents are carriers. This is called recessive inheritance. When both parents are carriers, the chance with each pregnancy to have a child CF is 25% (1 in 4 chance).
- CF carrier screening has been recommended by the American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) for:
 - Couples in whom at least one person is Caucasian and is pregnant or planning a pregnancy
 - Individuals with a family history of CF
 - Reproductive partners of an individual affected with CF
 - Screening should be made available to couples of non-Caucasian, lower risk, ethnic or racial groups
- Current CF carrier screening tests for the most common CF mutations (gene changes). Rare mutations are not analyzed.
- A negative screening result reduces the chance that someone is a carrier, but a small risk of being a n undetected carrier and a small possibility of having an affected child remain.
- A positive family history of CF will affect the estimation of carrier risk when a negative result is found and must be taken into account in the testing process. In some instances this is not straightforward, and genetic counseling is recommended.
- The estimate risk is couple-specific and does not apply to other pregnancies conceived with other partners.
- Some CF mutations and variants are associated with male infertility. Some couples may discover they are at increased risk of having an infertile son who is otherwise healthy.
- If a couple is found to be at risk of having a child with CF, genetic counseling and prenatal diagnosis are available. CVS (chorionic villis sampling) can be performed at 10 to 12 weeks, and amniocentesis can be performed at 15 to 20 weeks of gestation.
- Choosing prenatal diagnosis or other options is a private decision between a family and its health care provider.

This fact sheet is intended to highlight some key points in screening for cystic fibrosis. It is not intended to provide an in depth look at cystic fibrosis, informed consent, carrier screening, or prenatal testing options. This is not a test order; a test request form or prescription is necessary to obtain the test. Please discuss additional questions with your health care provider. *If your insurance does not cover this test you will be responsible to pay the lab fees of \$325.00.*

- Yes, I would like to have CF carrier screening.**
- No I do not want CF carrier screening.**

Printed Name _____

Signature _____ Date _____

Reviewed by _____

(Health care provider)