



INFORMATION ABOUT CYSTIC FIBROSIS

Cystic fibrosis (CF) is an inherited disease of the mucus and sweat glands. It affects mostly the lungs, pancreas, liver, intestines, sinuses and sex organs. CF causes the mucus to be thick and sticky. The mucus blocks air passage in the lungs, causing breathing problems and also making it easy for bacteria to grow. This can lead to problems such as repeated lung infections and lung damage.

The symptoms and severity of CF vary widely. Some people have serious problems from birth. Others have a milder version of the disease that doesn't show up until they are teens or young adults.

Although there is no cure for CF, treatments have improved greatly in recent years. Until the 1980s, most deaths from CF occurred in children and teenagers. Today, with improved treatments, people with CF live, on average, to be more than 35 years old.

Source: The National Institutes of Health

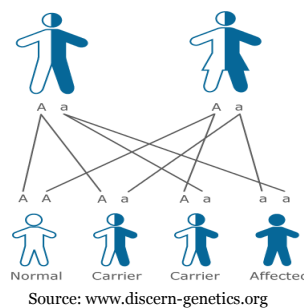
CYSTIC FIBROSIS CARRIER SCREENING

What is carrier screening?

The American College of Obstetricians and Gynecologists recommends that couples planning a pregnancy, or those already pregnant, be informed about genetic carrier screening. Specific ethnic groups are known to have an increased risk for certain genetic conditions, and carrier screening is available to find out if a couple is at risk for having a child with one of these conditions. If both members of a couple are carriers for the same condition, there is a 25% chance that the pregnancy will inherit the condition. *It is important to note that these conditions often occur in families with no previous history of the condition.*

How are these conditions inherited?

The conditions included in most carrier screening tests are inherited in an "autosomal recessive" manner. This means that both parents must be "carriers" for a condition in order to be at risk for a pregnancy to be affected with that condition. If both parents are carriers for the same condition, the chance of having an affected pregnancy is 1 in 4 (or 25%) for *each* pregnancy, as illustrated in the diagram below.



A "carrier" is an individual with no symptoms of a particular disease, but who possesses both a normal gene (inherited from one parent) and a non-working gene (inherited from the other parent) for that condition.

A "non-carrier" is an individual who inherits two normal genes, one from each parent.

An "affected" person has inherited two non-working genes, or mutations, one from each parent.

Who should be screened?

Carrier screening is based on an individual's ethnicity and/or country of origin. With every ethnicity there are certain genetic disorders that are more common. Genetic carrier screening is available for many conditions that are more common in certain populations.

Genetic carrier screening for cystic fibrosis is offered to all individuals, and is especially recommended for all Caucasian couples. Approximately 1/25 Caucasian individuals is expected to be a carrier of cystic fibrosis.

When should carrier screening be performed?

It is strongly recommended that you and your partner undergo genetic carrier screening prior to pregnancy, or as early in pregnancy as possible.

What is the carrier screening process?

- In our office, genetic counseling is provided before carrier screening is ordered. This ensures that you are offered the most appropriate carrier screening options.
- Carrier screening is performed through a simple blood test; no preparation is necessary.
- Test results are provided to you and your physician within 2-3 weeks.
- If both partners are determined to be carriers for the same condition, additional genetic counseling will be available to discuss the nature of the specific disorder as well as prenatal testing options.

UNDERSTANDING YOUR RESULTS

What does a “screen negative” result mean?

A “screen negative” carrier screen result means that your chance of being a carrier for cystic fibrosis has been greatly reduced. A negative result significantly lowers, but does not completely eliminate, the chance of being a carrier. Only the most common mutations in the CFTR gene are analyzed in carrier screening. The percentage of carriers detected by our cystic fibrosis carrier screen is >95%.

When your carrier screen is “screen negative”, we generally do not recommend that your partner pursue carrier screening, as the chance for an affected pregnancy is greatly reduced.

What does a “screen positive” result mean?

A “screen positive” carrier screen result means that you were found to be a carrier for cystic fibrosis. When the test determines that you are a carrier, the next step is for your partner to have carrier screening performed. Both parents must be carriers for the pregnancy to be at risk for cystic fibrosis. If testing determines that a couple is at risk, prenatal testing using chorionic villus sampling (CVS) or amniocentesis can be performed to determine whether the pregnancy has inherited the condition.

QUESTIONS?

If you have any questions concerning the above information, please do not hesitate to contact us at 312-981-4400.