Palmetto Health USC
MEDICAL GROUP
Department of Obstetrics and Gynecology
USC Genetic Counseling

CARRIER SCREENING FOR CYSTIC FIBROSIS

WHAT IS CYSTIC FIBROSIS (CF)?

CF is a genetic condition that causes buildup of mucus in the lungs and other organ linings leading to respiratory failure, frequent infections, pancreatic problems, and male infertility. People with CF typically have normal intelligence, but their lifespan is often shortened due to complications of the disease. However, there is a wide range of clinical symptoms for people with CF, from very mild symptoms and infertility to life threatening problems.

WHO IS AT RISK FOR CF?

In the general population, individuals of Caucasian ancestry and Ashkenazi Jewish ancestry have a higher chance to be carriers of CF than individuals of other ancestries. About 1 in every 29 Caucasian individuals carries a change in the CF gene, otherwise known as a CF trait. A person who has only one CF trait does not have symptoms of CF. However, a person who has two CF traits will have symptoms of the condition. In order for a pregnancy to have an increased chance for CF, both members of the couple must be carriers of CF traits. The chance that a Caucasian couple will have a child with CF is approximately 1 in 3,300 (0.04%).

People of other ethnic backgrounds can also be carriers of CF, but the chance is smaller. For example, people who are Hispanic have about a 1 in 46 risk to be carriers (1 in 8,400 for an affected child), while people who are African American have about a 1 in 60 risk (1 in 14,400 for an affected child). Please see the chart below for more information on your ethnic group.

Ethnic Background	Chance to have a child with CF	Chance to be a carrier	Chance a carrier will be detected by screening	Number of couples at risk for a child with CF missed by current screening	Remaining risk if one member of a couple is found to be a carrier and the other member is not
Caucasian, Nothern European (English, Irish, French, German)	1 in 3,300	1 in 29	90%	20%	1 in 1,000
Caucasian, General European			80%	35%	1 in 560
Caucasian, Southern European (Slovackian, Italian, Greek)			70%	50%	1 in 325
Ashkenazi Jewish	1 in 3,300	1 in 29	97%	5%	1 in 3,700
Hispanic	1 in 8,400	1 in 46	57%	68%	1 in 420
African American	1 in 14,400	1 in 60	72%	50%	1 in 780
Asian	1 in 32,400	1 in 90	30%	90%	1 in 500

WHAT IF I HAVE A FAMILY HISTORY OF CF?

If you have a family history of CF such as a brother, sister, or cousin with CF, then you may have a higher chance of being a carrier. Your specific carrier chance is determined based on how you are related to the person in your family with CF. Testing for CF may be more reliable than indicated based on your ethnic background alone. If the changes in the CF gene or CF traits are known for your family member, then the testing can look for those specific changes and rule our whether or not you have inherited them. Genetic counseling is recommended for people who have a family history of CF to discuss these issues in further detail prior to drawing blood for CF screening.

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HOW DOES THE TESTING WORK?

A blood sample is taken and the DNA that codes the CF gene is analyzed for the most common changes. Over 1,000 different changes in the CF gene have been described, but many of these changes are "private" mutations, meaning they only happen in a few families. Since only the common mutations can be tested for, not everyone who is a carrier will have a "positive" carrier screen. For example, in people who are Caucasian, 80% of carriers will be detected, whereas only 70% of African American carriers will be detected. This means that sometimes a couple who is at risk to have a child with CF will not be detected using current carrier screening technology.

WHAT HAPPENS IF I AM A CARRIER?

When a person tests positive for CF trait, carrier screening for their partner is recommended. Remember, a couple is only at increased chance to have a child with CF if both members are carriers. If one member of the couple is a carrier, but the other member is not a carrier of one of the common mutations, the chance for to this couple to have a child with CF is higher when compared to a couple where neither member is found to be a carrier of the common mutations. However, in most cases, this chance is small. Each couple's specific risk will be determined by their ethnic background and corresponding carrier detection rate. For example, a Caucasian couple where one member of the couple is found to be a carrier and one is found not to be a carrier will have a 1 in 560 risk to have a child with CF. If neither member of the couple is found to be a carrier of a common mutation, then the risk is reduced to 1 in 78,400.

If BOTH members of the couple are carriers, then the couple has a 25% risk (1 in 4) with each pregnancy to have a child with CF. This couple may consider prenatal diagnosis by CVS or amniocentesis to learn a fetus's CF status or may have their child tested at birth. Couples may consider alternative parenting options such as adoption, egg or sperm donation, or preimplantation genetic diagnosis.

Genetic counseling is recommended for every couple where at least one member of the couple is found to be a carrier. During a genetic counseling session many questions can be answered, such as explaining the inheritance of these conditions, teaching more about the diseases, and helping a couple understand their risks and reproductive options.

SHOULD I HAVE A CARRIER SCREENING FOR CF?

The American College of Obstetricians and Gynecologists (ACOG) published guidelines for offering CF carrier screening. They suggest offering screening to all women who are pregnant or planning a pregnancy, individuals with a family history of CF, and partners of individuals with CF or CF trait.

The decision to pursue screening is a personal one. Some women/couples want to know if their risk to have a child with CF is increased prior to or during a pregnancy. Other women/couples do not feel like the risk of CF is high enough for them to consider screening. Talk with your doctor and/or arrange to speak with a genetic counselor for more information.

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