

# PRENATAL DETECTION OF CYSTIC FIBROSIS

## What is Cystic Fibrosis (CF)?

Cystic Fibrosis is a genetic disorder that mainly causes problems with digestion and breathing. It does not affect intelligence or appearance. Not all people with CF have the same symptoms. Some have more severe symptoms than others. It is not always possible to tell how severe the symptoms will be just by a genetic test before birth. While most people with CF have a shortened life span, it is not easy to tell who will die in early childhood and who will live into their 40's or 50's.

## Is CF common?

It is the most common autosomal recessive disease among Caucasians with a disease frequency of 1 in 3,300 people.

## How can my baby get CF?

CF is genetic and is autosomal recessive. This means that both parents have to be carriers of a mutated gene in order to have a baby with CF. When both partners are carriers, there is still only a 1 in 4 chance or 25% chance that their baby will have CF.

## What does being a carrier mean?

A carrier does not have CF and probably doesn't even know that he/she is a carrier. It just means that they have one mutated copy of the gene that causes CF. You need two copies to have the disease.

## Are there some people who are at higher risk than others?

The chance of being a carrier mainly depends on your ethnicity or background. European Caucasians/Ashkenazi Jewish people are at the highest risk of being carriers. Their risk is 1 in 29. The chance that both partners are carriers is 1 in 841. For other groups such as Hispanic Americans, their risk is 1 in 46 for one partner and for two: 1 in 2,116. African Americans risk is 1 in 65 and 1 in 4,225 for both partners. Asian Americans have the lowest risk at 1 in 90 for one and 1 in 8,100 for both partners.

## How do I know if my partner and I are carriers?

A blood test can be done on both you and your partner to determine if you are both carriers. Since you both need to be carriers to pass it along to your baby, you may want to test only one of you first and if that partner is negative then the risk that your baby could have CF is very low.

## How accurate is the screening test?

The test screens for 25 of the most common mutations of the gene, which picks up 80% of European Caucasians, 90% of Northern European Caucasians and 97% of Ashkenazi Jews. The test has a lower detection rate for other ethnic groups like African Americans and Hispanic Americans because of the decreased frequency of the disease in these groups. This means that some people who are truly carriers might test negative with this screening test. But the risk of having a baby with CF is still low. For example, in the Ashkenazi Jewish population, if one partner is screened and is negative, the risk of having a baby with CF drops from 1 in 3,300 to 1 in 100,000; in people of European Caucasian descent, if only one partner is screened and is negative, the risk of having a baby with CF drops from 1 in 3,300 to 1 in 16,000.

## What if my partner and I are both carriers...then what?

Just because you and your partner are both carriers does not mean that your baby will automatically have CF. There is a 25% chance that your baby will have CF. It is possible to tell if your baby has CF by doing more extensive testing such as an amniocentesis or CVS (chorionic villus sampling). However, the severity of the disease cannot be predicted by these methods.

Remember, there is no easy answer as to whether or not to undergo genetic testing and no one can make the decision for you. A well-informed patient makes the best patient and we just want you to have all of the available options to you to ensure that this pregnancy has the best outcome possible. This screening test is not for everyone and you may decide not to have it done. Just read the information, ask questions and make the right decision for you, your partner and your baby.