

# Cystic Fibrosis Testing

*Sarah and Richard Anderson were planning to have three or four children. In April, after 9 trouble-free months, their second child, Mark, was born weighing 7 lbs 9 oz. Sarah had seen her obstetrician regularly, and there had been no complications at all. A few hours after the birth, the pediatrician examined Mark and pronounced him healthy. The Andersons were very happy and had no reason to think there were any problems. Their first child, a healthy, active 3-year-old named Ryan, was a little worried about becoming a big brother.*

*From the beginning, Mark was cranky. He had a great appetite, but wasn't growing well. At first the pediatrician was not concerned. Soon, Sarah and Richard began to feel that something was seriously wrong. Sarah tried breast-feeding and different formulas, but Mark still didn't gain much weight. Sarah was planning to go back to work, but she was exhausted.*

*Nothing she did seemed to comfort Mark. She felt like she couldn't do anything right. Richard talked with a friend at work about how worried he was. It just didn't make sense. Mark was eating constantly, but he was not growing. Their older son, Ryan, had been colicky, but he had grown fast. And, he did have some good times of the day. "It's strange," Richard told his friend, "when I kiss Mark he tastes salty." Finally, in July the pediatrician ordered some tests. The results showed that Mark had cystic fibrosis.*

## **What is cystic fibrosis?**

Cystic fibrosis (CF) is a common genetic disease. This means that a baby with CF has the disease from the moment of conception. Boys and girls have the same chance of being born with CF. It is not contagious or caused by an infection. Nothing the parents do during the pregnancy causes the baby to have CF.

## **What happens when a person has CF?**

CF affects certain cells in the body. Some of these cells are in the glands that produce the mucus in the lungs and in the digestive tract. When a person has CF, these glands do not work correctly. People with CF have frequent lung infections that cause permanent damage to the lungs.

Problems in the digestive tract may keep the body from digesting fat and protein. As a result, many people with CF have trouble gaining weight and growing. CF also affects the sweat glands. People with CF have saltier sweat than other people. The skin of a baby with CF may taste salty.

People with CF look and behave like other people. CF does not affect a person's intelligence, so many people with CF complete high school and go on to college. CF does not affect a person's appearance, except that people with CF are often thin. They may also be shorter than expected. People with CF generally live for 25 to 30 years. While some people with CF live into their 40's, about half of people with CF die before the age of 28. Almost all people with CF die from lung disease.

*The Andersons were shocked by the news from their pediatrician. They had barely heard of CF. They talked with their parents and relatives. No one on either side of the family had ever had CF. Sarah and Richard didn't understand how they could have one healthy child and one child with CF. They thought that if it was genetic, all of their children would have CF.*

*The pediatrician explained that their situation was typical. Most children with CF are born to families with no history of the disease. It is a matter of risk. Both Sarah and Richard are carriers of a gene that could cause CF. As a result, each of their children has 1 chance in 4 of having CF. He explained that many diseases and disabilities are probably genetic, but scientists don't always understand how these genetic differences are passed on. CF is one of the diseases that we do understand.*

## **What is a genetic disease?**

Just as our appearance is determined by our genes, many diseases can be caused by defects in our genes. Genes are very tiny, but each one contains important information. Genes provide the instructions that tell our bodies how to develop and how to function. When a person has CF, their genes send a wrong instruction to certain cells in the lung and digestive tract.

## **How does a person get CF?**

Many people find it hard to understand how parents can have a baby with CF when no one in their families has ever had the disease. The key is to understand that every person gets two copies of the gene involved in CF. One copy comes from each parent. For a person to have CF, both copies of the gene must have defects. The person must get an abnormal gene from each parent. If a person has one normal gene and one abnormal gene, he or she is a CF carrier. A carrier does not have CF. The CF carrier's normal gene provides the correct instruction to cells in the lungs and digestive tract. Of course, a person can also have two normal genes. That person does not have CF and is not a carrier. Every time a CF carrier has a baby, the baby can get the abnormal gene. When two CF carriers have a baby, these are the possible results.

## **Who are CF carriers?**

About 1 in every 25 white people in the United States is a carrier of an abnormal CF gene. A person who has a family member or relative with CF is much more likely to be a CF carrier. A carrier has no signs of CF and will never develop the disease. However, every time a carrier has a baby, there is a chance that the baby will get the abnormal gene. As long as the baby's other gene is normal, the baby will not have CF. In this way, the CF gene is passed on from parent to child for generations without any baby being born with CF.

## **Who is at greatest risk for CF?**

CF is one of the most common genetic diseases among white people. One out of every 2,500 white newborn babies has CF. You do not have to be white to have CF; however, it is most common in the white population. CF is less common, but it does occur, in people of African and Asian ancestry.

*Sarah and Richard worked with their CF doctor to find the best treatments for Mark. By giving him enzymes with his food, they were able to help him digest food better and gain weight. His mood improved and his parents began to enjoy him. Two months after the diagnosis was made, Sarah was able to go back to work. The Andersons learned to give Mark the breathing treatments he needed to help clear mucus from his lungs. Slowly Mark's CF treatments became routine, but it took Richard and Sarah many months to accept the fact that there was no cure for their son's illness.*

## **Is there a cure for CF?**

A great deal of research is being done to find better treatments for people with CF. CF is a chronic illness, which means it lasts for a lifetime. CF is not a predictable disease. For some children the symptoms are severe from the beginning, for others there may be periods of time when the symptoms are very mild. The goals of the treatments are to lessen the symptoms and prevent complications.

## **What treatments are there for CF?**

Most people with CF need antibiotics to treat lung infections. Different kinds of breathing treatments may help to clear the mucus from the lungs. Usually, parents learn to do the treatments at home. Children with CF may be hospitalized when a lung infection is severe. Most people with CF also have digestive problems. In order to digest fat and protein, they must take enzymes with their food. As a result, they can gain weight and grow. Better nutrition also gives them more energy so they can lead more active lives.

*Four years later, Mark is doing well. He was hospitalized once for a severe case of bronchitis and pneumonia. Sarah and Richard are finding it easier to cope and are trying to decide whether to have another child.*

*Many of Sarah and Richard's relatives have had CF carrier testing. Sarah's brother tested negative so it's very unlikely that he is a CF carrier. Richard's niece and one cousin both tested positive, but their partners had negative results on the CF carrier test.*

*The Andersons' close neighbors, Steve and Fran, were thinking about starting a family. They talked about CF carrier screening with Sarah and Richard. Fran said she was worried about having a child with CF, but she didn't think Steve realized what it would mean. Steve laughed and said he thought it was hard to imagine what it would be like to have any child. Then, he surprised Fran by saying he was in favor of carrier testing, "There are plenty of things in life we don't know about. It makes sense to me to learn what we can." Fran was glad. She said, "Hopefully we'll both test negative and we can pretty much stop worrying about CF. If it turns out we're both carriers then we can look at our options."*

## **What is CF carrier testing?**

A genetic test is now available to determine if a person is a CF carrier. Baylor College of Medicine is offering the test to individuals who would like to know if they are CF carriers. Couples can use the results of the test to make decisions about

planning their families.

### **What will the test tell me?**

If you have a positive test result, you will know that you are a CF carrier. If your test result is negative, there is still a small chance that you are a CF carrier. The test can detect approximately 90% of CF carriers. The percentage is even higher for people of Ashkenazic Jewish ancestry. Information on the detection rate for people of Hispanic, African and Asian ancestry is limited. If either you or your partner tests positive, many relatives will also be CF carriers. Their risk of having a baby with CF will be higher than the general population. We recommend that relatives of CF carriers have genetic counseling and testing before they plan to have children. What if my partner and I are both CF carriers?

When both partners are CF carriers, each baby has 1 chance in 4 of being born with CF. When both partners are carriers, they may choose to have a fetus tested for CF during the early stages of pregnancy.

There is no way for parents to control which gene they pass on. It is possible for two carriers to have several healthy children. It is also possible for two carriers to have several children with CF and no healthy children. Carriers who have one or more healthy children can still have a child with CF.

If you and your partner are both CF carriers, we will provide as much information as you would like about the disease. Many families find it helpful to talk with doctors and other staff members at the Cystic Fibrosis Center.

### **What do negative results mean?**

When one or both partners has a negative result, there is still a small chance of having a baby with CF. Because the test is not 100% accurate, a few people who have negative results are CF carriers. The best way to understand the risk of having a baby with CF is to look at the possible results. Remember, a white couple with no history of CF in either family has 1 chance in 2,500 of having a baby with CF. This is the risk for the general population.

If one partner tests positive and the other tests negative, their chance of having a baby with CF goes up. About 1 child in 880 born to these couples will have CF. This is a greater risk than the general population. However, it is still a fairly low risk.

If both partners have negative results on their CF carrier tests, their chance of having a baby with CF is very small (only 1 in 190,000 babies born to these couples has CF). Most couples have negative test results and find this information very reassuring.

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