Understanding Newborn Screening for Cystic Fibrosis

Video Transcript

Narrator: Having a baby is an exciting, but stressful time in every parent's life. As parents, you're coping with the many challenges of caring for a newborn baby. You're learning how to care for a new person, with his or her own unique needs and personality.

At the same time, medical professionals are giving you information about the care and health of your new baby. It can be a bit overwhelming, and just when you're getting the hang of being a new parent, your child's doctor gives you some unexpected news. It's about the newborn screening test results for Cystic Fibrosis, done when your baby was just born.

Mother 1: When my first child was born, I didn't know anything about newborn screening, and I was surprised at the hospital they asked to have blood work done and I didn't really pay that much attention to it.

Narrator: This video will provide answers to some of parents' commonly asked questions about the newborn Cystic Fibrosis screening process.

The doctor has told you that the newborn screening test for Cystic Fibrosis came back positive. This doesn't mean your baby definitely has Cystic Fibrosis, it only suggests that the possibility for Cystic Fibrosis is higher, and that more tests will be needed.

Mother 2: When I got the initial call from my doctor, who told me that both boys needed to be rechecked because they did test positive for Cystic Fibrosis, I was extremely nervous. I didn't know what this disease was, if there was a cure, what part of the body it affects, or anything.

Narrator: You might be wondering: "Does my baby have Cystic Fibrosis? Will my baby get sick? Is there a cure? How did this happen? And, should other family members get tested?"

When your baby was born, he or she had blood drawn and placed on a card. The card was then sent to the Newborn Screening Lab of Hygiene in Madison, where your baby's blood was tested for several genetic conditions. Your baby tested positive for Cystic Fibrosis. What does that mean? First, let's consider what the test itself tells us.

Technicians at the lab tested your baby's blood for immunoreactive trypsinogen. This chemical, known as IRT, is made in a baby's body. Your baby's blood level of IRT may be high for many reasons. Perhaps labor and delivery were stressful, or you might have been given Pitocin to help you with the delivery process, or the IRT level might be high for unknown reasons. The IRT level does not diagnose Cystic Fibrosis. It simply identifies the babies that have a higher chance to have Cystic Fibrosis.

The next step done on babies with levels of high IRT in their blood is genetic testing. Lab technicians will test the same spot of blood for the presence of the most common CF genes. This most common gene is called Delta F 508. We will talk more about genes later in this video. Doctors of babies testing positive for the presence of a CF gene are notified of abnormal newborn screen result. They are encouraged to have these children tested in a third way for Cystic Fibrosis. This is called a sweat test.

Mother 2: To have the test done, it was nothing at all. I thought they were going to hook them to all these devices and everything. They did not. It's just little electrodes they put on their arms to make them sweat. And then from there, then the test goes. I got the phone call that same day to let me know that the latest carrier trait is all they do.

Narrator: Children with Cystic Fibrosis have a higher amount of salt in their sweat. In most cases, testing the amount of salt in a baby's sweat indicates whether or not your baby has Cystic Fibrosis.

The sweat test is a simple, inexpensive, and very accurate method of diagnosing Cystic Fibrosis. A mild, safe medication that induces sweating is applied near the sweat glands on the child's forearm. The area is then stimulated by a very slight, painless electrical current. After a few minutes, the electrodes are removed and an absorbent material is placed over the same area. The sweat that is collected is then analyzed in a laboratory.

If your baby has a normal sweat test result, he or she does not have CF. This means your baby will never develop Cystic Fibrosis. However, you baby does still carry a copy of the CF gene. Why does this matter? Genes are the instruction booklets that tell our bodies how to work. For example, genes determine our height, hair color, and eye coloring.

Genes come in pairs. We get our genes from our parents. We get half our genes from our mother and half from our father. Therefore, a child who has CF then gets their CF genes from their parents. Likewise, there is a same chance a child can receive no CF genes. The parents of a child with CF each have one copy of the CF gene. We call a person who has one copy of a CF gene a carrier. A carrier does not have CF, and usually has no knowledge that they carry the CF gene. However, parents can unknowingly pass the CF gene onto their children. The chance of this happening is the same with each pregnancy.

Brothers and sisters of children with CF can also be carriers of the CF gene. This matters when carriers of CF are old enough to think about having their own children. They should consider having their partner tested to see if he or she is also a CF gene carrier, because parents who are both CF carriers can have children with CF.

Genetic testing is available if parents wish to determine their CF gene status. Other, more distant family members may also carry a gene for CF and may wish to find out about testing. Many center have free genetic counseling services provided through state funding. You do not need to have a completed sweat test to call these resources. Your doctor can provide a list of genetic resources in your area. Feel free to call if you have questions or would like to schedule testing for CF.

Many centers, like Children's Hospital of Wisconsin, will also provide free nursing and/or genetic counseling on the day of a sweat test to answer any questions parents might have.

Mother 1: My first thought for parents who are coming in for the screening is, be careful who you ask information from, because there are a lot of people who don't know a lot about CF or if they do, it's kind of the old information. Make sure that you are asking people that really know what they are talking about to explain it.

Narrator: Your health care provider will contact you with the results of a sweat test.

Though the effects of CF vary with each person, it usually affects the lungs and digestive track of people with this condition.

Why is it important to test newborns for CF? The necessary medical care and nutritional support can be started at an early age. The hope is that early diagnosis and treatment will give children a healthier start. You can be encouraged by the fact that treatment and the health of children with Cystic Fibrosis has significantly improved in recent years.

We have not gone into great detail about Cystic Fibrosis in this program. We encourage you to find out more by contacting the Cystic Fibrosis Foundation.