

Newborn Screening Education for Parents and Families

MY BABY HAD AN ABNORMAL CYSTIC FIBROSIS NEWBORN SCREENING TEST

Before you took your baby home from the hospital, a small amount of blood was taken from his or her foot for newborn screening. Newborn screening is a statewide program to identify babies who may have certain health problems. Babies are screened for these problems because the earlier a child is diagnosed and treatment started, the healthier the child will be. You may have received a pamphlet from the hospital or your doctor describing the diseases that are screened for in your state.

WHY DOES MY BABY NEED A TEST FOR CYSTIC FIBROSIS?

One of these tests screened for is cystic fibrosis (CF). Your baby's screening test showed that your baby might have CF. However a second test called a sweat test or a genetic test is needed to diagnose CF.

The sweat test will measure how much salt is in your baby's sweat. This test is the best way to diagnose CF. Too much salt in the sweat means a person has CF. ***Most babies who have to have a sweat test because of an abnormal newborn screen do not have CF,*** but it is important to be sure that they do not. This should be done at a CF Foundation-accredited care center. You can find the center closest to you and more about sweat testing on the CF Foundation's Web site (www.cff.org).

CF causes people to have salty sweat, breathing and digestive problems. Those with CF need to be seen by special CF health care providers. When CF is diagnosed early, the baby can start to receive care to help keep them healthy.

Your baby may have a genetic test instead of a sweat test to find out whether or not he/she has CF. Genetic testing can be done by a blood test. A person has to have two abnormal CF genes or CF mutations in order to be diagnosed with CF.

WHAT CAN I EXPECT BEFORE AND DURING THE SWEAT TEST?

A CF sweat test should be done at a CF Foundation-accredited care center. This test can usually be done within a few weeks from the time the newborn screening test results are reported to your baby's doctor. If you have questions about or need help scheduling the sweat test, talk with your baby's doctor.

On the day of the sweat test, do not use any lotions, creams or moisturizing soaps on your baby's arms or legs. Bring an extra blanket or sweater and hat to keep the baby warm and so he/she will sweat during the test.

The sweat test takes about one hour from start to finish. A special gel is applied to the baby's skin on their arms or legs and small patches with wires are applied for about 5 minutes to cause the baby to sweat. The skin may feel warm and tingly while the patches are in place, but it is not painful. The sweat is then collected on a gauze pad or disk. After about 30 minutes, the gauze or disk is removed and the collected sweat will be tested in the lab.

WHAT DO THE RESULTS MEAN?

Results of the sweat test should be available to your doctor by the next day or two. There are four possible results:

Negative result: This means that a normal amount of salt was found in the baby's sweat. It is very rare for a person to have CF if the sweat test result is negative.

Positive result: A positive sweat test means that the baby probably has CF. A second sweat test should be done and an appointment should be made at a CF Foundation-accredited care center that specializes in treating people with CF. Your doctor can refer you to the closest CF center or you can find a care center on the CF Foundation's Web site (www.cff.org).

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Borderline result: Sometimes the sweat test result will be in between positive and negative. The baby should have another sweat test or a genetic test and should be seen at a CF Foundation-accredited care center.

Quantity Not Sufficient (QNS): This means that there was not enough sweat on the gauze to measure the amount of salt. The baby will need to come back on another day to repeat the sweat test.

MY BABY WAS FOUND TO HAVE ONE CF GENE OR TO BE A CF CARRIER. WHAT DOES THIS MEAN?

Some babies whose newborn screen is positive or abnormal for CF do not have the disease, but they are carriers of one CF gene mutation and do not need special medical care. However, if your baby is a CF carrier, then either you and/or your partner are also carriers of the CF gene mutation. If both of you are CF carriers, then you could have a baby with CF in the future. This fact sheet *My Baby is a CF Carrier: What does that mean for my baby? What does that mean for me?* talks about what it means to be a carrier of a CF gene mutation. It is available from the CF Foundation (www.cff.org). If you are not sure if your baby is a CF carrier, ask your baby's health care provider.

I HAD A NEGATIVE CF BLOOD TEST WHEN I WAS PREGNANT. DOES MY BABY STILL NEED TO BE TESTED FOR CF?

Yes. Any baby who had a positive or an abnormal newborn screening result should be tested for CF either by a sweat test or genetic test. A genetic test for CF looks for only the most common mutations in the CF gene. There are over 1,300 CF gene mutations so you may carry a mutation that was not tested for when you were pregnant.

RESOURCES FOR PARENTS AND FAMILIES

To learn more about testing for cystic fibrosis and Newborn Screening for CF, visit the CF Foundation's Web site (www.cff.org) or you can contact the CF Foundation at 800 FIGHT CF (800-344-4823).