When Baby Needs a Second Test for Cystic Fibrosis

A small sample of your baby's blood was collected soon after birth and sent to the DHEC Public Health Laboratory for testing. This testing is called Newborn Screening. In South Carolina, newborns are tested for several genetic and chemical disorders. Sometimes, a second test is needed to help your doctor decide if your baby has one of the disorders. In many cases, the second test will be normal. However, if your baby does have one of the newborn screening disorders, early treatment will give him or her the best chance to grow up healthy. Because a compound called immunoreactive trypsinogen (IRT) was high in your baby's first test, he or she could possibly have Cystic Fibrosis (CF).

What is Cystic Fibrosis?

Cystic fibrosis is an inherited genetic disorder of the mucus glands. It is found in roughly 10 to 15 babies born in SC each year. Mucus is a slippery substance the body secretes to cover and protect many organs and tissues. CF causes the body to produce excess mucus that is abnormally thick and sticky, which can lead to a variety of health problems.

CF affects the lungs and digestive system the most. Babies with CF often cough or wheeze and can get lung infections that need treatment with strong medications. Also, they may not digest their food well, despite a huge appetite. One of our organs, the pancreas, makes digestive juices or "enzymes" that break down the food we eat. In babies with CF, these enzymes cannot mix with food, making it hard to digest milk. Poor digestion can cause diarrhea, very smelly diapers and poor growth.

Babies with CF often have not started to show any of these problems or to "act sick". However, if the condition is identified early and proper treatment is begun, many of the symptoms of CF can be controlled and children can live longer, healthier lives.

How will I know if my baby really has Cystic Fibrosis?

A portion of your baby's blood that was already sent to DHEC will be tested again for changes in their CF gene. Because children with CF also have extra salt in their sweat, a special test called a "sweat chloride test" is usually done. This test needs to be done at a certified CF Center, so it may not be available at your local hospital. To get the baby to sweat, a special chemical is put on a small part of the baby's arm or leg. The skin is then slightly warmed. After five minutes or so, the skin is cleaned. Over the next half hour, sweat is collected on special paper or in a plastic coil. The amount of salt (chloride) in the baby's sweat tells the doctor if the baby really has CF.

What do I need to do until I know the test results?

Your baby may not have any symptoms at first. But you will need to follow your doctor's instructions very carefully. If your baby seems to be getting sick, call your doctor right away.

How is Cystic Fibrosis treated?

Babies with CF are usually referred to a certified CF Care Center, where the family is told about CF by a counselor. Each baby is different, but they are usually given medications and treatments to help them avoid lung infections. Most babies with CF also need enzyme medications to help digest their food. Medicines are given before each feeding. Babies with CF can be breastfed. Babies with CF who aren't growing well are sometimes fed special formulas or given supplements with extra calories. Once they start eating solid food, it is important they get plenty of calories and vitamins to help them grow. Certified CF Care Centers have registered dietitians (RD) and nutritionists who work closely with families, so the baby will gain weight but won't have diarrhea.

What else should I do to keep my baby as healthy as possible?

CF is a serious disease. But following the doctor's instructions closely will give a baby with cystic fibrosis the best chance to stay healthy. If you have question or concerns now, before the testing is completed, talk to your primary doctor, who will guide you through this process.

Where can I find additional information?

https://www.babysfirsttest.org/newborn-screening/conditions/cystic-fibrosis-cf

https://www.cff.org/ http://www.newbornscreening.info/Parents/otherdisorders/CF.htm