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 laboratory for testing. This testing is called Newborn Screening. In SC, newborns are tested for several genetic and chemical disorders. Some times, 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 a genetic disorder that is found in around 10 to 15 babies born in SC each year. When a baby has CF, some fluids in the body that should be thin and slippery are thick and sticky. These fluids can plug up some of the "tubes" or "channels" in the body. This 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 drugs. They also may not digest their food well, often in spite of 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 their milk. Poor digestion can cause diarrhea, really smelly diapers and poor growth. Babies found through screening often have not yet started to show much of these problems or to "act sick".

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

The IRT test will be repeated and if the IRT is still high in a second blood sample, your doctor will do other tests to figure out if you baby really has CF. Because children with CF also have extra salt in their sweat, a special test called a "sweat test" is usually done. This test needs to be done at a 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 about five minutes, the skin is cleaned and, over the next half hour, the 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 does have CF.

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

Your baby will probably 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. There the family is fully told about CF. Each child is different, but they are usually given medicines and treatments to help them avoid lung infections. Most babies with CF need enzyme medicines to help them digest their food. These 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 are given supplements with extra calories. Once they start eating solid food, it is important that they get plenty of calories and vitamins to help them grow. Certified CF Care Centers have registered dietitians (RD) 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 the 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 difficult time.