When Baby Needs A Second Test for Galactosemia
Elevated Total Galactose with Normal GALT

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 these 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 galactose was high in your baby's first test, he or she could possibly have a form of galactosemia. It is NOT likely that he or she has classical galactosemia (GALT deficiency).

What is galactosemia?

When a baby has galactosemia, he or she cannot break down galactose, a part of the sugar lactose that is found in breast milk and cow's milk-based baby formula. Galactose builds up in the baby's blood and damages the baby's body. There are three main forms of galactosemia. They can be identified when the compound galactose is measured in a baby's blood. When the galactose is high, another test is also done. It measures an enzyme called galactose-1-phosphate uridyl transferase or GALT. **Your baby's GALT test was normal, so it is not likely that he or she has classical galactosemia, the most serious form.**

One of these other forms is called galactokinase (GALK) deficiency. A baby with GALK deficiency will have cataracts in the eyes.

Another form is called UDP-galactose-4-epimerase (GALE) deficiency. A baby with GALE deficiency may not ever have any problems. However, a very few babies with GALE deficiency may have serious problems like swollen liver, weight loss and weakness.

Very few babies have either GALK or GALE deficiency. Your baby needs to be retested quickly, though, as a precaution.

How will I know if my baby really has one of these forms of galactosemia?

The newborn screening test will be repeated and additional tests may be done to help the doctors figure out if your baby has galactosemia. Usually the results of these tests take a few days to come back. You may also be referred to a doctor who specializes in these kinds of disorders.

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. Your doctor will decide whether your baby needs any treatment right now.

How is galactosemia treated?

All forms of galactosemia are treated with a special diet. At first, babies with galactosemia must be fed a soy-based baby formula. When they begin to eat solids, the parents will have to be careful about which foods are given to the baby. The baby must not eat foods that contain any milk or dairy products, including all animals' milk like goat's milk. A dietitian will help the family learn which foods the baby can eat.

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

Don't forget to keep all of your well baby check-ups! Seeing the doctors regularly and following your baby's diet plan carefully are the best things you can do to help your baby grow up healthy.