When Baby Needs a Second Test for Elevated PHE
Benign Hyperphenylalaninemia (H-PHE) or Phenylketonuria (PKU)

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 biochemical disorders. Sometimes, 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 a newborn screening disorder, early treatment will give him or her the best chance to grow up healthy.

Because a compound called phenylalanine (PHE) was high in your baby's first test, he or she could possibly have a condition called benign hyperphenylalaninemia (H-PHE) or Phenylketonuria (PKU).

What is Benign Hyperphenylalaninemia (H-PHE)?

Benign hyperphenylalaninemia (H-PHE) is a mild form of phenylketonuria. It is considered an amino acid condition because people with H-PHE have problems breaking down an amino acid, a building block of proteins, known as phenylalanine. Most people with this condition experience mild symptoms or no symptoms.

Children with H-PHE have more phenylalanine in their bodies than is typical. But they have a lower amount of phenylalanine in their bodies than do children with the condition known as classic phenylketonuria. Measuring the amount of phenylalanine in your baby's body can help doctors determine if your baby has this condition.

What is Phenylketonuria (PKU)?

PKU is a genetic disorder that is found in a few babies born each year. PKU may be identified when compounds called amino acids are measured in a baby's blood. Amino acids are the "building blocks" of protein. When a baby has PKU, he or she cannot use one of the amino acids that are found in foods including breast milk and infant formula. This amino acid, PHE, builds up in the baby's body. Over time, very high amounts of PHE can hurt the baby's brain and cause severe intellectual disability.

How will I know if my baby really has PKU?

If your baby's newborn screening result showed very high PHE levels, he or she may have PKU. The newborn screening test will be repeated, and additional tests will be done to help the doctors figure out if your baby has PKU. Usually, the results of these tests take a few days to come back. You will also be referred to a doctor who specializes in these kinds of disorders called a metabolic specialist.

What do I need to do until I know the final results? How is PKU treated?

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.

PKU is treated with a special diet. At first, babies must be fed a special formula. When they begin to eat solids, the protein in their diet will need to be limited very carefully. They may still need to drink special metabolic formulas throughout their lives to make sure they get everything they need to grow properly. 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 and develop normally.

Internet Resources: http://www.babysfirsttest.org/