

## When Baby Needs A Second Test for Phenylketonuria (PKU) Elevated PHE

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 PHE (phenylalanine) was high in your baby's first test, he or she could possibly have a disorder called Phenylketonuria.*

### What is Phenylketonuria (PKU)?

PKU is a genetic disorder that is found in a few babies born each year. PKU can 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 mental retardation.

### 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 probably has 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.

### 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 PKU treated?

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 will still need to drink their special formula 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.