

# **PARENT INFORMATION for POSITIVE Newborn Screening Results for PHENYLKETONURIA (PKU)**

When your baby was born a small sample of blood was taken from your baby's heel to test for certain diseases. These diseases usually cannot be found simply by looking at your baby, so that is why the blood test is so important.

The results of your baby's screening test were positive for a disorder called Phenylketonuria (FEE-nil-KEY-tone-u-ree-ah), or PKU. It is important to remember this is only a screening test. A new blood sample will be collected from your baby and a new test will be done to help to determine if your baby has PKU or not.

## **What does a positive screen mean?**

A positive test means that your baby needs to have a new blood test to check for PKU. A small percent of all babies screened will be identified as positive on screening. Some are later found not to have PKU. This is just a screening test and further testing needs to be done to determine if your child has this disorder or not. Keep in mind that the new test could show that your baby does not have PKU.

## **What is the next step?**

Be sure to follow your baby's doctor's instructions for getting a new blood sample collected for further testing. PKU is a rare metabolic disease, so your baby's doctor might consult with or refer you to a specialist.

## **What exactly is PKU?**

PKU is an inherited disease. An enzyme that would normally help break down the amino acid phenylalanine is missing. The phenylalanine or phe, then builds up in the baby's body. When the phe levels get too high, the nerve and brain cells are damaged. If it is not treated, it can result in mental retardation.

Phenylalanine is one of the building blocks of protein. It is found in protein foods like milk, meat, eggs, and cheese. After eating these foods, the phe levels in a person with PKU can

get too high. Finding out early if your baby has PKU, means you and your baby's doctor can take steps to prevent these effects with a special diet for your baby.

## **What should I do?**

**The most important thing that you can do is to take your baby to get a new blood sample collected and tested immediately.** If your baby does have PKU, there are very effective treatments available. The pediatric metabolic specialists and nutritionists will be able to help you manage your baby's special diet.

Treatment should NOT be started before the results from the new blood tests are received. NO treatment should be done unless a doctor recommends it. In an infant, PKU is treated by giving a formula with reduced or absent phenylalanine. Regular checks by a nutritionist and a metabolic physician are necessary. The Nebraska Department of Health and Human Services contracts with the University of Nebraska Medical Center in Omaha to provide formula, and medical, nutritional and consultative services to all PKU affected individuals born in Nebraska.

You have probably already done this, but now is also a good time to be sure your newborn is covered with your health insurance provider.

For more information, talk with your baby's doctor. You can contact the Nebraska Newborn Screening Program at (402) 471-0374 for information on specialists available in Nebraska.

Department of Health & Human Services

