Arkansas Department of Health
Newborn Screening Result: Elevated Phenylalanine
Infant may have Phenylketonuria
Information Sheet for Parents

You have just heard that your infant may have Phenylketonuria (FEE-nil-KEY-tone-u-ree-ah), or PKU. Please understand that the newborn screening is just that – a screening test. Not all cases that are screened positive will be confirmed to have the diagnosis. The below information is meant to keep you informed while further testing is done to evaluate the newborn screening result.

Overview
Phenylketonuria (FEE-nil-KEY-tone-u-ree-ah), or PKU is an inherited disease that causes the body to have problems breaking down certain proteins. People with PKU cannot break down a substance in proteins called phenylalanine.

What is PKU?
People normally break down extra phenylalanine that the body doesn't need. However, people with PKU are unable do this, so the extra phenylalanine builds up and becomes toxic to the organs and the brain. Phenylalanine is broken down by an enzyme that is either missing or not working correctly in people with PKU. Untreated PKU can cause health concerns, behavior problems, and mental retardation. A special diet that is low in phenylalanine usually prevents most of the symptoms of PKU.

Why is newborn screening done for PKU?
Newborn screening for PKU offers earlier detection so that treatment can begin earlier. Early detection of PKU is key to preventing many of the complications that may arise should the disorder go untreated during a child's infancy. Specialists will be able to improve the quality of life for individuals with PKU.

Does a positive newborn screening result mean that a baby has PKU?

Not always. Some babies who are screened "positive" are found not to have the disorder after further testing. This is because the screening test is not a diagnostic test. A screening test is designed to identify all infants with the disorder, but sometimes picks up infants who don't have the disorder. Therefore further testing needs to be done in order to determine if the baby has the disorder, or to rule it out as a "false positive."

How common is PKU?
PKU affects one in every 15,000 babies.

What are the signs and symptoms of PKU?
Babies with PKU may appear normal at birth; however, later they may show signs of skin rash, seizures, excessive restlessness, irritable behavior, and a musty odor of the body or urine.

How is PKU diagnosed?
After receiving a positive newborn screen, the most important thing parents can do is be sure that their baby goes in for a new specimen to be collected and tested as soon as possible. The test results should be available within 3 days.

Is there a cure for PKU?
There is not currently a cure for PKU. However, treatment programs are allowing most people with PKU to live fairly normal lives.

How is PKU treated?
If a diagnosis is confirmed, a pediatric metabolic specialist and nutritionist will be able to help parents manage PKU. No therapy will be started until results come back from the confirmatory test. PKU can be treated by giving a formula with reduced levels of phenylalanine along with regular monitoring by the nutritionist and metabolic physician. Treatment is lifelong.

Where can I get additional information?
http://www.arkansas.gov/newborn_screening/index.html