



## Phenylketonuria (PKU) Information for Parents

### ➤ Overview

Phenylketonuria (PKU) is an inherited metabolic disorder. This specific disorder affects the breakdown of protein by the body.

### ➤ What is PKU?

Phenylketonuria (PKU) is a disorder of amino acid metabolism that results in excess levels of phenylalanine in body fluids. Babies with PKU are unable to use a certain part of protein (phenylalanine) found in food and milk. Without treatment, phenylalanine will build up in a baby's body and intellectual disabilities will result.

### ➤ Why is newborn screening done for PKU?

Newborn screening is done for PKU so that babies with this condition can be diagnosed quickly. **Early diagnosis and treatment is essential to prevent developmental delays.**

### ➤ Does a presumptive positive result from the Kansas Newborn Screening Lab mean that my baby has PKU?

No, not necessarily. Newborn screening tests the baby's level of phenylalanine. The level was outside of the normal range in your baby. Prompt follow up is important. Additional tests will need to be done to determine if your baby has PKU or not.

### ➤ How common is PKU?

In the United States, PKU occurs in 1 in 25,000 newborns. The incidence varies according to ethnic background of the child, with a higher incidence in Caucasians and Native American populations and lower incidence in African American, Hispanic, and Asian populations.

### ➤ How is PKU inherited?

PKU is inherited in an autosomal recessive pattern. Parents of a child diagnosed with PKU are unaffected. These individuals are carriers of the condition. Each pregnancy between carrier parents has a 25% chance of producing a child affected with PKU, a 50% chance of producing an unaffected carrier child, and a 25% chance of producing a child who is unaffected and is not a carrier.

➤ **What are the signs and symptoms of PKU?**

Although infants with PKU usually appear normal at birth, early symptoms may include skin rash, seizures, excessive restlessness, irritable behavior and a musty odor of the body or urine. Later signs include developmental delays, problems with walking and balance and intellectual disabilities.

➤ **How is PKU diagnosed?**

Diagnostic tests include plasma amino acids. Additional tests may be recommended by a specialist.

➤ **Is there a cure for PKU?**

There's no cure for PKU, but starting a low-phenylalanine diet as soon as possible after birth can help to prevent the harmful effects. Phenylalanine is found in many proteins, so protein-rich foods, such as meat, must be avoided and replaced with 'safe' proteins, usually in the form of specially formulated substitutes. This diet should be followed for the long term.

➤ **How is PKU treated?**

Early diagnosis and treatment is essential to prevent developmental delays. Treatment is life long and consists of a low phenylalanine diet, which includes a specialized medical formula, in combination with regular foods that are low in phenylalanine.

A subset of children with PKU have a different form of the disorder, due to pterin defects, and may require specific therapy.

Babies born to mothers with poorly controlled PKU have a significant risk of intellectual disabilities due to their exposure to very high levels of phenylalanine before birth. These infants may have a low birth weight and grow more slowly than other children. Other characteristic medical problems include heart defects, small head size, and behavioral problems. Women with poorly controlled PKU also have an increased risk of miscarriage. Strict dietary control prior to conception and through out pregnancy is essential.

➤ **Where can I get additional information?**

1. For more information about newborn screening in general and about PKU specifically, contact the national Newborn Screening and Genetics Resource Center, 1912 W. Anderson Lane, Suite 210, Austin, TX 78757; telephone 512-454-6419; fax 512-454-6509.
2. National Coalition for PKU and Allied Disorders [www.pku-allieddisorders.org](http://www.pku-allieddisorders.org)
3. National Institutes of Health [www.nih.gov](http://www.nih.gov)