

## Overview of Newborn Screening for Phenylketonuria – For Parents

### What is newborn screening?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to test for a group of conditions. One of these conditions is **phenylketonuria** (also called **PKU**). Babies who screen positive for phenylketonuria need a second test done to confirm they have PKU. **Not all babies with a positive newborn screen will have phenylketonuria.**

### What is phenylketonuria?

When a person has phenylketonuria, his/her body is not able to break down **phenylalanine** (also called **phe**—pronounced —feel). Phe is an **amino acid** (one of the —building blocks used to make proteins). Phe is found in many of the foods that we eat. Most people with PKU are missing an **enzyme** (a protein that helps our bodies function) called **phenylalanine hydroxylase** (also called **PAH**). When a person with PKU eats food containing phe, his/her body cannot break down the phe. Instead, the phe builds up in the blood. There is a milder (less severe) type of PKU called **hyperphenylalaninemia** (also called **hyperphe**). People with hyperphe may not need treatment.

### What causes phenylketonuria?

Phenylketonuria is an **inherited** (passed from parent to child) condition. Everyone inherits two copies of the gene for PAH. We inherit one copy of the PAH gene from our fathers and one copy from our mothers. Sometimes these genes have changes (also called mutations) that prevent the gene from working correctly. In order for a person to have PKU, he or she must have two PAH gene changes. People with one PAH gene change do not have PKU.

### What are the symptoms of phenylketonuria?

Every child with phenylketonuria is different. Most babies with PKU will look normal at birth. Symptoms of PKU can appear in the first six months of life if a baby with PKU does not receive treatment. Some of the symptoms of untreated phenylketonuria include:

- Mental retardation
- Behavior problems
- Seizures
- A skin condition called **eczema**
- A —mousy or —musty body odor
- Fair (light) skin & hair

### What is the treatment for phenylketonuria?

There is no cure for phenylketonuria. However, there are treatments that can help with the symptoms. Babies with PKU need to be on treatment as soon as possible to prevent the symptoms of PKU. Babies with PKU should drink a special medical formula that does not contain phe. People with phenylketonuria need to follow a special diet that contains low levels of phenylalanine. When a child with PKU gets older, medication may be added to his/her diet to help control phe levels. A person with phenylketonuria will need treatment for his/her entire life.

### What happens next?

Although there is no cure for phenylketonuria, good medical care makes a difference. Children with phenylketonuria should see a metabolic geneticist (a doctor who specializes in PKU and other related conditions) as well as their pediatrician. Your child's doctor will work with the metabolic geneticist to coordinate any treatment, tests, or appointments that your child needs.

### Where are Indiana's metabolic genetics clinics?

Indiana's metabolic genetics clinics are located at Riley Hospital for Children at IU Health, Indianapolis, IN, (317) 274-3966 and The Community Health Clinic, Topeka, IN, (260) 593-0108.

### Where can I get more information about phenylketonuria?

- **STAR-G** – <http://newbornscreening.info/Parents/aminoaciddisorders/PKU.html>
- **Region 4 Genetics Collaborative** – <https://www.region4genetics.org/education/families/>