Phenylketonuria (PKU)

What is phenylketonuria?

Phenylketonuria (PKU) is a rare birth defect. A person who has PKU is missing an enzyme needed to process an amino acid called phenylalanine. Amino acids are the building blocks for protein. For children with PKU, too much of this amino acid can be dangerous.

What is the cause?

For a child to be born with PKU, both parents must have the defective gene. If only one parent has the PKU gene, there's no risk of passing PKU to a child. A parent can have the defective gene, but not have the disease. This is called being a "carrier." PKU occurs mainly in white people. It's much less common in black or Asian people.

Children of mothers who have PKU but who didn't follow the PKU diet during pregnancy may have problems because of the high level of phenylalanine in the mother's blood. These babies are at risk of being born with mental retardation or an abnormally small head (microcephaly). They may also have heart defects, low birth weights, and behavioral problems.

What are the symptoms?

Newborns who have PKU do not have symptoms. Without treatment, though, babies usually show signs of PKU within a few months. Symptoms can be mild or severe and may include:

- A musty odor in the child's breath, skin or urine
- Fair skin and blue eyes, because the body produces less melanin (the chemical responsible for hair and skin tone)
- Rocking
- Tremors or jerking movements in the arms and legs
- Skin rashes
- Small head size
- Stunted growth
- Vomiting.

A child with PKU may be irritable, restless, and destructive. Untreated PKU leads to brain damage and mental retardation within the first few months of life. Older children with untreated PKU may also develop behavioral problems and seizures.

How is it diagnosed?

Newborn screening identifies almost all cases of PKU. All 50 states in the United States require newborns to be tested for PKU.
Tests for PKU are done between 24 hours and 7 days after birth, They are usually done before a new baby leaves the hospital. If you don't deliver your baby in a hospital or are discharged soon after the birth, you may need to schedule a newborn screening with your health care provider.

How is it treated?

The main treatment for PKU is a strict diet with very limited amounts of phenylalanine, which is mostly found in protein. People with PKU need to follow the diet for life.

Because regular infant formula and breast milk contain phenylalanine, babies with PKU are put on a special infant formula. A small amount of breast milk may be okay for some babies. However, your child’s health care provider must determine a safe amount.

A special nutritional drink or supplement is available for people with PKU. The formula contains protein substitutes and essential nutrients but little or no phenylalanine. Older children continue to drink several glasses of formula each day, as directed by a health care provider or dietitian.
You'll need to keep records of what your child eats every day. It helps to use a food diary or computer program that lists the amount of phenylalanine in baby foods, PKU formulas, and other foods.

Children should avoid foods and medicines made with aspartame (NutraSweet, Equal). Aspartame, found in many artificial sweeteners, releases phenylalanine when digested. You'll find low-protein rice, pasta, pizza crusts, tortillas, bagels, breads, cookie dough, and baking mixes, as well as egg substitutes and imitation cheeses. These products allow children with PKU to eat lunches and dinners that are similar to what everyone else is eating.

A safe amount of phenylalanine differs for each person. Your health care provider will determine a safe amount through regular review of diet records, growth charts, and blood tests. Talk to your health care provider or dietitian if you have any questions.

Can it be prevented?

If either parent has a family history of PKU, your health care provider may suggest screening tests before pregnancy or birth. A blood test can tell if you are a PKU carrier.

Women with PKU can prevent birth defects by sticking to a low-phenylalanine diet before becoming pregnant. Even women with mild PKU should follow the special PKU diet while pregnant.

*NOTE: This information is provided as a public educational service. The information does not replace any of the instructions your physician gives you. If you have a medical emergency please call 911 or call the Hospital at (208) 529-6111. If you have questions about your child's care, please call Idaho Falls Pediatrics at (208) 522-4600.