

## Phenylketonuria (PKU)

### What is PKU?

PKU is a type of amino acid disorder. People with PKU have problems breaking down an amino acid called phenylalanine (Phe) from the protein in food they eat.

### What Causes PKU?

Enzymes help start chemical reactions in the body. PKU happens when an enzyme called “phenylalanine hydroxylase” (PAH) is either missing or not working right. This enzyme breaks down the amino acid Phe. When a child with PKU eats food containing Phe, it builds up in the blood and causes problems. Phe is found in almost every food, except pure fat and sugar. PKU is an inherited disorder where both parents carry a gene for PKU.

### What Symptoms or Problems Occur with PKU?

*[Symptoms are something out of the ordinary that a parent notices.]*

Babies with PKU seem perfectly normal at birth. The first symptoms are usually seen around 6 months of age. Untreated infants may be late in learning to sit, crawl, and stand. They may pay less attention to things around them. A child with PKU who doesn't get treatment will become intellectually disabled.

Some of the things caused by untreated PKU include:

- intellectual disability
- behavior problems (such as hitting, biting)
- hyperactivity (over-active)
- restlessness or irritable mood
- seizures
- eczema (itchy areas of skin that become flaky or hard)
- a “musty” or “mousy” body odor
- light hair and skin

### What is the Treatment for PKU?

The following treatments are often used for children with PKU:

**1. Medical formula with low-Phe** – Even though they need less Phe, children with PKU still need a certain amount of protein. A special low-Phe medical formula gives babies and children with PKU the nutrients and protein they need. It helps keep their Phe at a safe level.

**2. Low-Phe food plan** – The right diet is made up of foods that are very low in Phe. This means your child must not have cow's milk, regular formula, meat, fish, eggs, or cheese. Regular flour, dried beans, nuts, and peanut butter also have Phe. They must be highly limited or not eaten at all. This diet is needed for life. A dietician can help plan the proper diet for the child.

**3. Medication** – Additional treatment may include BH4. There are some newer medications that may be indicated.

### Things to Remember

Children with PKU who start treatment soon after birth usually have normal growth and intelligence. Even when treated, some children have problems with schoolwork and may need extra help.