

# Introduction to Phenylketonuria

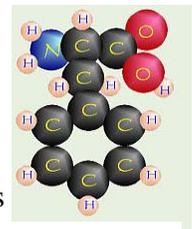
Your child has been diagnosed with a condition called phenylketonuria, or PKU for short. There is no cure for PKU, however, with proper treatment, those with PKU can live a normal, healthy life.

Children with PKU are unable to break down (metabolize) the amino acid phenylalanine (Phe) in a normal way. Amino acids, including Phe, are part of proteins in the food we eat, therefore children with PKU must limit their intake of protein containing foods. Your child will need to eat all the foods necessary for normal growth and development but within the limits of phenylalanine he or she can safely use.

As you read the following information, if you have any questions, be sure to write them down and ask the nutritionist (dietician), nurse, or doctor at the metabolic clinic.

## What is PKU?

PKU is a genetic inborn error of protein metabolism. Proteins, which are made up of many different amino acids linked together, are found in many parts of the human body including hair, blood, skin and muscles. Most foods also contain proteins, which are important for building and repairing the body's tissues. When we eat foods containing protein, the protein is split into amino acids during digestion by means of an enzyme. Enzymes are substances that cause a chemical change, breaking down the protein into its "building blocks" of amino acids. The amino acids are later put back together in a different way to form a new protein which is then used by the body.



Phenylalanine

PKU is characterized by the absence or deficiency of the enzyme responsible for processing the essential amino acid phenylalanine, or Phe, which is found in all foods containing protein. All children need a certain amount of Phe for normal growth and tissue repair. With normal enzyme activity, most of the Phe is converted to another amino acid (tyrosine), which is then utilized by the body. However, when the phenylalanine hydroxylase (PAH) enzyme is absent or deficient, some of the phenylalanine takes an alternate route and is converted to phenylpyruvic acid and other products. These products and the unused Phe accumulate in abnormally high amounts in the blood and are toxic to brain tissue.

Without treatment, most infants with PKU develop mental retardation, or other neurological symptoms. To prevent these problems, treatment consisting of a Phe-restricted diet is begun during the first days or weeks of life. Since Phe is found in all foods that contain protein, children with PKU must limit their intake of protein containing foods. Foods that contain high amounts of protein are high in Phe and therefore should not be eaten. Foods with small amounts of protein and Phe can be eaten in controlled amounts. Some foods are "free" foods because they contain no protein and are free of Phe. "Free" foods are eaten to help boost the child's intake of calories needed for energy.

A child with PKU also has a special drink or medical formula that has most of the protein, vitamins and minerals that a child needs for growth with little or no Phe. The formula provides almost all of the nutrients that other children get from their food.