

# PHENYLKETONURIA

## FACT SHEET



Ellis, diagnosed with PKU

*This fact sheet is for educational purposes only. An experienced healthcare professional should be consulted for the management of phenylketonuria.*

### What Is Phenylketonuria?

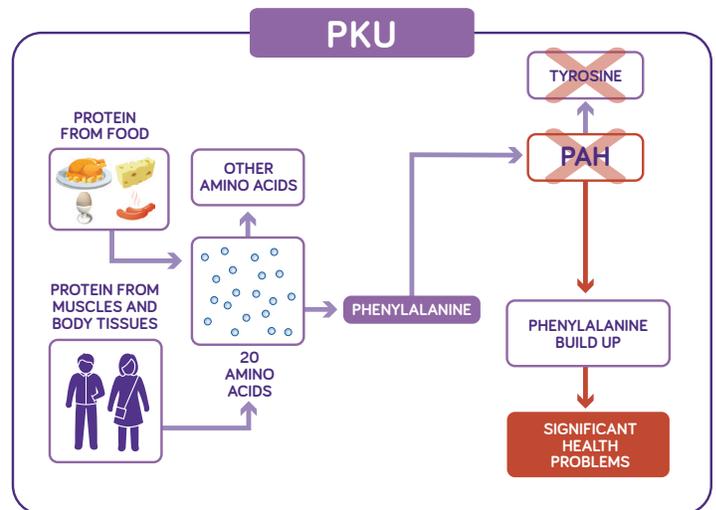
Phenylketonuria (PKU) is an inherited genetic disorder caused by a deficiency of the enzyme phenylalanine hydroxylase (PAH). PAH helps the body break down the amino acid phenylalanine (PHE) into tyrosine (TYR), another amino acid.

Protein is made up of smaller units called amino acids. Amino acids are important to build muscle and other body tissues for normal growth. Any extra amino acids are normally broken down to produce energy. In PKU, PHE cannot be broken down in the body. Consuming too much PHE from food can lead to a toxic buildup of PHE in the blood and brain of a person with PKU.

Finding out that your child has PKU can be overwhelming. Fortunately, nutrition management is available to help prevent many of these problems. Take comfort in knowing that your metabolic healthcare team can provide you with the best advice to help manage PKU.

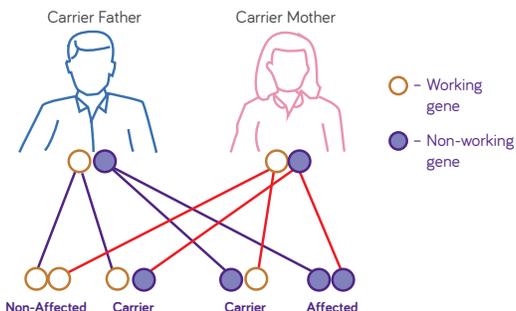
There are two classifications of PKU: classic PKU and hyperphenylalaninemia (hyperphe). Both classic PKU and hyperphe are characterized by a mutation of the PAH enzyme. Classic PKU is the result of severe mutations of this enzyme and requires a strict low-PHE diet. In the case of hyperphe, the defect in the enzyme is less severe, and blood PHE levels are usually not as high as in a person with classic PKU. However, blood PHE levels are still above the normal range, which may require some dietary restriction.

Persons with hyperphe tend to tolerate higher amounts of PHE in their diet. Your metabolic healthcare team can provide you with the best advice to help manage these dietary restrictions.



### Autosomal Recessive Inheritance

Chances a child will be affected when both parents are carriers



### Incidence, Genetics and Newborn Screening

It is estimated that one out of 10,000 live births has decreased PAH activity. There is a higher incidence in Caucasians and Native Americans, and a lower incidence among African Americans, Hispanics, and Asians.

PKU is inherited as a “recessive disorder.” In recessive disorders, both the mother and father are “carriers,” and do not have any symptoms of the disorder (see diagram). With each pregnancy there is a 1 in 4 (25%) chance that the infant will be affected. A genetic counselor can help explain the inheritance of PKU and the risks to future infants.

PKU can be screened for at birth through a simple blood test, referred to as newborn screening (NBS). NBS for PKU began in the US in the 1960s. It is important that an infant is diagnosed and managed with diet within days after birth to prevent permanent negative effects from PKU. For more details on NBS in the US, visit [www.newbornscreening.hrsa.gov/your-state](http://www.newbornscreening.hrsa.gov/your-state); for information on NBS in Canada, visit [www.raredisorders.ca](http://www.raredisorders.ca).

## Management of Phenylketonuria

There is currently no cure for PKU, but it can be managed with a special low-PHE diet consisting of foods low in protein and a special PHE-free formula\*. Since all foods with protein contain PHE, persons with PKU must restrict the amount and types of foods containing protein. The main sources of foods with protein allowed on the low-PHE diet are vegetables and fruits and a limited amount of cereals and grains. In addition, individuals with PKU must drink a special PHE-free formula\* that supplies additional protein the body needs for growth and maintenance, without the addition of PHE. There is also a wide range of commercially available low protein food products for today's PKU population.

Nutricia North America provides a range of PKU formulas. Please contact us for more information. Your dietitian will help you decide which products are best for you.

Nutrition management of PKU includes frequent monitoring of blood PHE levels and regular clinic visits. Blood PHE levels can be measured either at the clinic or hospital, or collected at home and mailed in for analysis. The recommendation is to maintain blood PHE levels between 120-360 µmol/L (2-6 mg/dL).

## Symptoms of Elevated Blood PHE Levels

An infant with PKU who is not managed immediately will begin to build up PHE in the blood and brain. In an infant with PKU, increased PHE levels in the blood and brain over time will result in irreversible brain damage. Therefore, it is essential that each infant who is diagnosed with PKU be started on the low-PHE diet as soon as possible after birth.

If a child or adult previously diagnosed with PKU and on diet decides to discontinue the diet, he or she will begin to build up PHE in the blood, which increases PHE levels in the brain. Over time, toxic levels of PHE in the brain can harm a person's nervous system. A person with PKU who discontinues the low-PHE diet might experience mood changes and become easily aggravated. Thinking skills, memory, social behavior and concentration can all decline with high PHE levels. Also, a person might feel shaky and clumsy. For these reasons, it is important that persons with PKU stay on the low-PHE diet as advised by their healthcare provider.

*\*With the exception of glycomacropeptide (GMP)-based formulas, which naturally contain some PHE.*



Contains GMP, made from a whole protein source!

## Resources

- Screening Technologies and Research in Genetics (STAR-G): [newbornscreening.info](http://newbornscreening.info)
- Newborn Screening in Your State (US): [newbornscreening.hrsa.gov/your-state](http://newbornscreening.hrsa.gov/your-state)
- National PKU Alliance (NPKUA): [npkua.org](http://npkua.org)
- Canadian Organization for Rare Disorders: [raredisorders.ca](http://raredisorders.ca)
- Canadian PKU & Allied Disorders: [canpku.org](http://canpku.org)

*Nutricia North America would like to thank Laurie Bernstein, MS, RD, FADA, FAND, Children's Hospital Colorado for her consultation.*

Questions about PKU  
Formula Coverage?



1 800-365-7354

Coverage@Nutricia.com

For more information or product samples, please visit [NutriciaMetabolics.com](http://NutriciaMetabolics.com)  
or call US: (800) 365-7354

All products shown are medical foods in the US and foods for special dietary use in Canada for the dietary management of phenylketonuria (PKU) and must be used under medical supervision.