

## Phenylketonuria (PKU)

### What is Phenylketonuria?

Phenylketonuria (also known as PKU) is an inherited condition that affects the way a person's body uses [protein](#). A person with PKU cannot use a component of [protein](#) called phenylalanine. Phenylalanine is an [amino acid](#) needed for proper growth and development, but too much can cause serious health problems. In the case of PKU, too much phenylalanine builds up in the blood, penetrates and damages the brain. The high levels of phenylalanine ultimately cause mental retardation and other serious health problems.

About one baby in 14,000 is born with PKU in the United States. The condition occurs in all ethnic groups, but it is most common in individuals of Northern European ancestry.

### How does PKU affect a child?

The symptoms of this condition vary from mild to very severe. The most severe form of this condition is known as Classic PKU. Infants with Classic PKU appear normal until they are a few months old. Without treatment, these children develop permanent mental retardation and behavioral problems. Seizures, delayed development and movement disorders are also common. These children may have a musty odor about them, and may have dry skin, rashes or convulsions. Less severe forms of PKU (sometimes called Mild PKU or hyperphenylalaninemia) also require treatment. These rare variants also have high phenylalanine that is not PKU and require different treatment.

### What causes PKU?

PKU is a genetic condition caused by a change in the PAH (Phenylalanine Hydroxylase) [gene](#). The PAH [gene](#) is responsible for making an [enzyme](#) called phenylalanine hydroxylase. Phenylalanine hydroxylase changes phenylalanine to other needed compounds in the body. When there is an alteration in the PAH [gene](#), phenylalanine hydroxylase levels go down; therefore, phenylalanine builds up in the blood stream.

PKU is inherited in an [autosomal recessive](#) pattern, which means two copies of the PAH [gene](#) must be changed for a person to be affected with PKU. Most often, the parents of a child with an autosomal recessive condition are not affected because they are "[carriers](#)", with one copy of the changed [gene](#) and one copy of the normal [gene](#).

When both parents are [carriers](#), there is a one-in-four (or 25 percent) chance that both will pass the changed PAH [gene](#) on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal PAH [gene](#), and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed PAH [gene](#) from one parent and a normal PAH [gene](#) from the other, making it a [carrier](#) like its parents. These chances are the same in each pregnancy with the same parents.

### Is there a test for PKU?

Yes. Babies are tested (newborn screening) for PKU before they leave the hospital. The baby's heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has more than a normal amount of phenylalanine. All states screen newborns for PKU.

### **Can PKU symptoms be prevented?**

Yes. In most cases, the symptoms of PKU can be prevented by a diet very low in phenylalanine. This diet should begin as soon as possible following a diagnosis. Children and adults with PKU require follow-up care at a medical center or clinic that specializes in this condition.

Phenylalanine content in foods are different, so an experienced dietician or nutritionist will recommend a special diet that includes certain vegetables, fruits, grains, and a metabolic formula (food) that provides protein without phenylalanine. Other medical replacements, such as tyrosine may be needed. In addition, regular blood tests are used to monitor phenylalanine levels.

### **What is maternal PKU?**

Maternal PKU is when there are high levels of phenylalanine in a woman's blood during pregnancy. This circulates to the growing fetus. These high levels greatly increase the risk for a baby to be born with a small head size (microcephaly), mental retardation, growth delay, heart defects, characteristic facial features and behavioral problems. For women with PKU, it is important that they follow a low phenylalanine diet if they plan to become pregnant or are pregnant. The bad effects of high levels of phenylalanine can be prevented if this diet is followed prior to conception and during the pregnancy.

**DISCLAIMER:** The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

### **Resources:**

Children's PKU Network  
3790 Via De La Valle, Suite 120  
Del Mar, CA 92014  
Phone: 800-377-6677 / 858-509-0767  
Fax: 858-509-0768  
Email: [pkunetwork@aol.com](mailto:pkunetwork@aol.com)  
[www.pkunetwork.org](http://www.pkunetwork.org)

National Coalition for PKU and Allied Disorders  
PO Box 1244  
Mansfield, MA 02048  
Phone: 877-996-2723  
[www.pku-allieddisorders.org/home.htm](http://www.pku-allieddisorders.org/home.htm)

National PKU News  
Virginia Schuett, Editor/Dietician  
Email: [schuett@pkunews.org](mailto:schuett@pkunews.org)  
[www.pkunews.org](http://www.pkunews.org)

### **References:**

- Cedaerbaum, S.D., Scott, C.R., & Wilcox, W.R. (1997) Amino Acid Metabolism In; Rimoin, D.L., Connor, J.M., Pyeritz, R.E. (eds) Emery and Romoin's Principles and Practice of Medical Genetics, 3rd ed. Churchill Livingstone, New York, 1867-1870.
- GeneTests (Phenylalanine Hydroxylase Deficiency) <http://www.geneclinics.org>

- National Institute of Health Consensus Development Conference Statement:  
Phenylketonuria: Screening and Management, October 16-18, 2000. *Pediatrics* 108:972-82
- Online Mendelian Inheritance in Man (OMIM topic 261600)  
<http://www.ncbi.nlm.nih.gov/Omim>
- Scriver, C.R. and Kaufman, S (2001) The Hyperphenylalaninurias. In: Scriver, C.R., Kaufman, S., Eisensmith, E., Woo S.L.C., Vogelstein, B. Childs, B. (eds) *The Metabolic and Molecular Bases of Inherited Disease*, 8th ed. McGraw-Hill, New York, Ch.27.