

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 and 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 in the United States is born with PKU. 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%) 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 that child a carrier like the parents. These chances are the same in each pregnancy with the same parents.

Is There a Test for PKU?

Yes. Babies are tested through 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 specialize in this condition. Phenylalanine content in foods is different, so an experienced dietician or nutritionist will recommend a special diet that includes certain vegetables, fruits, grains, and a metabolic formula (medical replacement

food) that provides protein without phenylalanine. Other medical replacements, such as a tyrosine supplement, may be needed. In addition, regular blood tests are used to monitor phenylalanine and tyrosine levels.

What is Maternal PKU?

Maternal PKU happens 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. It is important that women with PKU 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 before 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:



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
www.pkunetwork.org



PO Box 1244
Mansfield, MA 02048
Phone: 877-996-2723
www.pku-allieddisorders.org/home.htm



Virginia Schuett, Editor/Dietician
Email: schuett@pkunews.org
www.pkunews.org

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