

PKU PRIMER



For Community Health Care Providers
Treating Adolescents and Adults with PKU

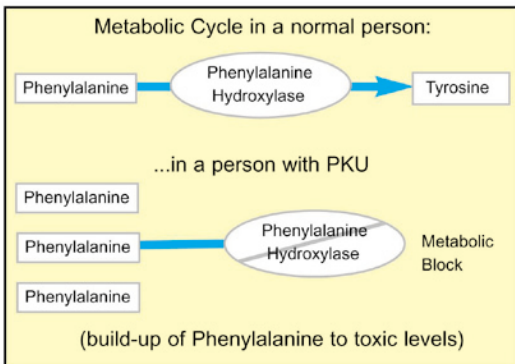
The Children's Hospital
of Boston



BACKGROUND

· What is PKU? What is Maternal PKU?

PKU or *phenylketonuria* is an inherited biochemical disorder that prevents a person from metabolizing the essential amino acid, *phenylalanine*. This problem occurs because a person with PKU lacks the enzyme, *phenylalanine hydroxylase*, which is required to convert phenylalanine into another amino acid, tyrosine. Because of this metabolic defect, phenylalanine rises to toxic levels inside the body and can cause mental retardation, seizures, learning disabilities, and emotional problems when the infant or adult is untreated or inadequately treated. PKU is rare, with a frequency of approximately 1 in 11,000 in the U.S. Fortunately, all newborn babies are screened for PKU before leaving the nursery. This allows the baby with PKU to be diagnosed within the first two weeks of life and in time for the immediate treatment necessary to prevent the damaging effects of the disease. In cases of Maternal PKU, a woman with this metabolic disorder is pregnant and must maintain strict control of her diet to prevent toxic phenylalanine levels from arising in the intrauterine environment.



· How can a person control PKU through diet?

Protein is composed of amino acids strung together like beads on a string. One of these amino acids is phenylalanine. Therefore, to avoid a build-up of *phenylalanine*, a person with PKU must remove most forms of protein from his/her diet. Ordinarily, such a diet would not allow a person to grow and develop normally, since amino acids are

essential for life. To ensure proper nutrition, a person with PKU must consume a special formula that contains all the necessary amino acids except phenylalanine. Unfortunately, the formula has a strong odor and taste and is sometimes not considered palatable by children and adults. If strictly adhered to, a low-protein diet, which would include such foods as fruits, vegetables, sugars and low-protein products, combined with the formula, will result in the optimal health outcome for a person with PKU. The diet

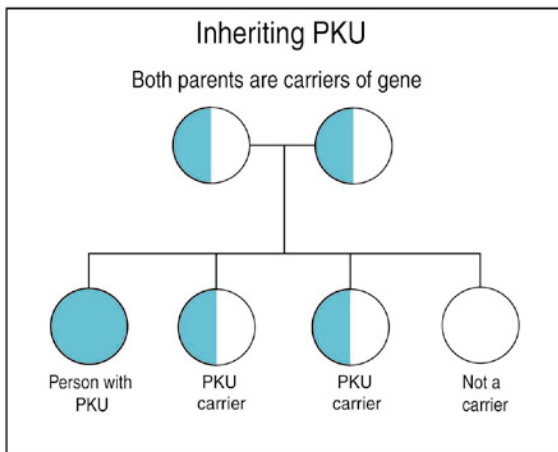
must be administered by the parent of a child with PKU in the early years, but as the child grows older, he/she must learn how to maintain the strict diet in everyday life. **Studies have suggested that the diet should be continued indefinitely, into adulthood, to avoid the harmful effects of elevated phenylalanine levels.** Pregnant women with PKU or women with the disease who are planning to conceive, must maintain a strict low-phenylalanine diet to protect the baby from toxic levels of phenylalanine.

· **Are all PKU cases the same?**


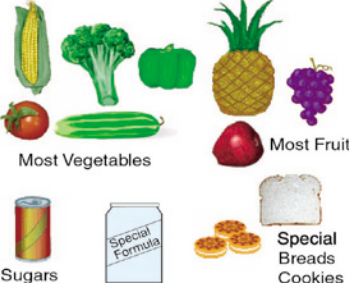
No, there exist different degrees of PKU in those who inherit the disorder. Some children and adults exhibit few or no symptoms while only loosely maintaining a diet while others experience serious symptoms unless they strictly adhere to the dietary treatment. Because of this variability, everyone diagnosed with the disorder must stay on a carefully monitored diet, while checking phenylalanine levels regularly.

· **How can PKU affect a person's performance in school and/or work?**

Children and adults with treated PKU are often faced with certain adverse effects of the disorder in work and school. Visual-motor deficits as well as spatial perception problems are common, leading to difficulty in arithmetic and other tasks requiring analytical skills. PKU often affects a person's fine motor speed, reaction time, and executive functioning skills



related to memory and problem solving. Attention deficits and hyperactivity, which can further aggravate learning disabilities, are also common among children with the disorder -- especially those whose metabolic control is less than optimal. A pregnant woman with PKU, in addition to these common emotional problems, often must face the additional stress and hormonal changes associated with a pregnancy.

High Phenylalanine Foods:	Low Phenylalanine Foods:
 <p>Fish</p> <p>Beans</p> <p>Diet Soda</p> <p>ASPARTAME</p> <p>Dairy</p> <p>Wheat</p> <p>Soy Products</p> <p>Meat</p> <p>Eggs</p> <p>Nuts & Legumes</p> <p>High-Protein Foods</p>	 <p>Most Vegetables</p> <p>Most Fruit</p> <p>Sugars</p> <p>Special Formula</p> <p>Special Breads Cookies Crackers</p> <p>Low-Protein Foods</p>

· **If strict dietary control is not maintained, what possible symptoms and/or medical complications can occur?**

Almost all individuals with PKU experience occasional lapses in dietary control, with few adverse effects. However, prolonged elevations in phenylalanine can cause emotional and psychological problems such as agoraphobia, anxiety, depression, attentional problems, headaches, sleep disorders and antisocial behavior. Many adults with PKU discontinue the diet either permanently or for a number of years because it is so difficult to maintain. In rare cases, seizures or other severe neurological symptoms can result from a terminated diet. In cases of Maternal PKU, the woman must maintain both her own health and the health of the fetus, therefore she must be even more persistent in resuming or maintaining the PKU diet.

· **Is the PKU treatment and diet expensive to maintain, and where can a person obtain extra formula if they exhaust their supply?**

The special formula can only be purchased through certain companies and is very expensive. Most people who purchase the formula use some form of insurance coverage or another form of financial support. The special low-protein products (pastas, cheeses, pizzas, breads, cookies, etc.) are also extremely expensive, but are critical to maintaining the diet.

MATERNAL PKU

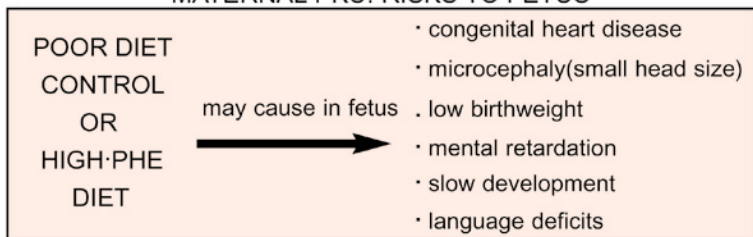
· Can women with PKU have children, and if so, will their children be born with PKU?

Yes, women with PKU may have healthy children if they maintain a strict diet prior to and during pregnancy. It is very important that a woman's phenylalanine levels be stabilized before conception. Even a strict diet may take weeks to reduce high phenylalanine levels, weeks during which a fetus can be seriously damaged. Therefore, women with PKU must plan their pregnancies very carefully. High phenylalanine levels in the intrauterine environment are toxic to the fetus and, if not closely monitored, can cause severe brain damage to the unborn child. In most cases of maternal PKU, the baby will not inherit the disorder (unless the father is also a carrier of the PKU gene). However, if the pregnancy is not treated, the baby will be born with one or a number of birth defects such as mental retardation, microcephaly (small head size), and congenital heart disease. These defects can be avoided if the mother brings her phenylalanine levels under control before becoming pregnant and maintains the low levels throughout pregnancy.

· How is coping with maternal PKU in everyday life challenging both socially and emotionally?

The strict PKU diet and large quantity of daily formula which must be consumed often disrupt daily activities and make it difficult for a woman with PKU to carry on with an uninhibited schedule. The diet and the formula can create awkwardness in social situations (particularly in restaurants), add considerable stress to the woman's life, and may cause depression and anxiety. Women who cope with maternal PKU and the demands of pregnancy, have the additional discomforts of nausea and vomiting. In addition, their stomachs may be easily upset by the formula or foods they normally include in their diet. PKU mothers also must resist high-phenylalanine food cravings common to pregnancy (peanut butter, ice cream, meat, etc.) The substantial amount of responsibility and stress involved in maintaining low phenylalanine levels may lead to frustration and lapse in dietary control.

MATERNAL PKU: RISKS TO FETUS



· **What happens if a woman with PKU does not maintain strict dietary control until after conception?**

The main risk if treatment is not started prior to pregnancy is that of a congenital heart defect in the fetus, since the heart forms within the first 30 gestational days. If diet is not brought under control until after the first trimester, permanent damage occurs: microcephaly, low birth weight, a reduction in IQ, and developmental delays.

· **How strict must the diet be?**

Women should keep their phenylalanine levels around 2-6ml/dl. In other words, the diet must be extremely strict. Moreover, the pregnant women with PKU must make sure they are gaining the proper amount of weight during pregnancy. To do this, they must eat foods high in fat while low in protein.

· **How can regular visits to the Metabolic Clinic help a young woman monitor her phenylalanine levels and her pregnancy?**

Regular visits to the clinic allow for frequent testing of blood-phenylalanine levels which need to be kept low during all stages of pregnancy. In addition to monitoring the woman physically, the clinic can help assess her psychological and emotional well being through tests and through discussions with physicians.

· **What can Community Health Care Providers do to ensure the health and well-being of young women with PKU?**

A primary care provider, school nurse, or other member of the woman's support network can greatly help her to cope with PKU during the difficult months of pregnancy. Many

studies have shown that psychosocial support is essential in helping a person with PKU maintain and stay committed to diet. It is important to give the woman with PKU positive emotional support and encouragement while reminding her of the importance of adhering to a strict diet. Staying in contact with the woman throughout her pregnancy will help provide a form of consistent and reliable support which she may or may not have at home. Congratulating the woman on successfully sticking to her diet and maintaining low phenylalanine levels throughout pregnancy will provide her with positive feedback and support. The woman may also need additional community resources which she may need help in finding or accessing. Therefore it may be helpful for someone in her support network to research and provide this information.

What is the Resource Mother's Program?

"The Resource Mothers" are mothers of children with PKU and are therefore very familiar with the low-phenylalanine diet, the special formula, and the frustration that often accompanies maintenance of a PKU diet. Each Resource Mother is paired with a woman with PKU who is pregnant or who is planning a pregnancy. The Resource Mother stays in close contact with the woman throughout her pregnancy, offering emotional support and encouraging her to comply with all aspects of treatment (diet, formula, blood tests.) The Resource Mother also helps the woman with PKU learn how to prepare special low-phenylalanine recipes which often require ingredient substitutes and special low-phenylalanine food products. This form of psychosocial support has been shown to be extremely helpful for some women who benefit from the knowledge and motivation of the Resource Mothers. To receive more information about the programs, call or email Susan Waisbren, PhD at 617-355-4686 (susan.waisbren@tch.harvard.edu) or write to:

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Important Points to Know about PKU and Maternal PKU

- I Individuals with PKU who maintain treatment generally experience **normal developmental progress and normal health**. However, some individuals experience learning disabilities and emotional disturbances, especially if phenylalanine levels are not well controlled.
- I Treatment consists of a **phenylalanine-restricted (low protein) diet** and a supplemental **formula** providing the essential elements of protein without the “offending” amino acid.
- I Individuals with PKU should drink the special **formula** daily in order to meet their protein needs. Starches, fruits and vegetables may be eaten in measured amounts. High protein foods (meats, eggs, dairy products) and Nutrasweet/Aspartame should be avoided.
- I Many individuals with PKU who are not on the low-phenylalanine diet may still **benefit significantly from the diet**. They should be seen regularly by a metabolic clinic
- I Women with PKU need to **prevent unplanned pregnancies, make a conscious reproductive decision, institute strict metabolic control prior to pregnancy, and maintain metabolic control** throughout pregnancy.
- I **Psychosocial factors** are important. **SAM**: social Support, positive Attitudes toward treatment, and resources to **Manage** the diet lead to greater success in maintaining metabolic control.
- I The National Institutes of Health Consensus Development Conference (October, 2000) recommended **dietary treatment for life**.

For more information contact the New England Consortium of Metabolic Programs
(www.childrenshospital.org/newenglandconsortium)