

# **Transition to Adult Health Care**

## **Maternal PKU**



**Children's Hospital Boston  
Transition to Adult Care Education Initiative**



**Children's Hospital Boston**

## Maternal PKU

Maternal PKU is when a woman with PKU is pregnant. In this case it is necessary that she maintains strict metabolic control both before and throughout her pregnancy to protect her baby.

Bringing a baby in the world is a wonderful experience. There is no reason for a woman with PKU not to enjoy this unique experience of her life. The difference is that she needs to plan her pregnancy and be more careful and strict with her diet.

Let's take a closer look to maternal PKU and answer some important questions.

### Can a woman with PKU have children, and will her children be born with PKU?

Yes, women with PKU may have healthy children if they maintain strict diet and take the formula prior to and during pregnancy.

### Why is it important to maintain the diet before, as well as during pregnancy?

High phenylalanine levels in the intrauterine environment are toxic and can be bad for the fetus. If not closely monitored they can harm the unborn child and cause severe brain damage. When a person is not on diet, even a strict diet may take weeks to reduce high phenylalanine levels. During those weeks, however, the fetus can be seriously damaged. That's why women with PKU must plan their pregnancies very carefully and maintain strict diet even before pregnancy. However, in case of an unplanned pregnancy, a woman with PKU should still return to and maintain the strict diet as soon as possible. If the pregnancy is not treated, the baby will be born with one or several of birth defects, such as mental retardation, small head size and heart related problem that is present since birth (congenital heart disease). These **adverse implications though can be avoided** and it is up to the mother to bring her phenylalanine levels under control before becoming pregnant if possible, and maintaining low levels throughout pregnancy.

### Is it certain that since I have PKU my child will also develop PKU?

In most cases of maternal PKU, the baby will not inherit the metabolic disorder. Only if the father is also a carrier of the PKU gene, the child will also develop PKU.

Specifically, every person has 2 genes that can cause PKU, one gene inherited from the father and one from the mother. Each of the 2 genes may carry PKU or be normal. The following may occur:

- If both parents pass along to their child a normal PKU gene, then the child will not have PKU.
- If one parent passes along a PKU gene, but the other parent passes along a normal gene, then the child will not have PKU.
- The only way that the child will develop PKU, is if both parents have the PKU gene and pass it to the child.

### How strict must the diet be?

Every woman can handle a different amount of phenylalanine. However, women should keep their blood phenylalanine levels around 2-6 mg/dl. In other words, the diet must be extremely strict. Also, the pregnant woman with PKU must make sure she is gaining the proper amount of weight during pregnancy.

### How can a pregnant woman with PKU gain the proper amount of weight that she needs?

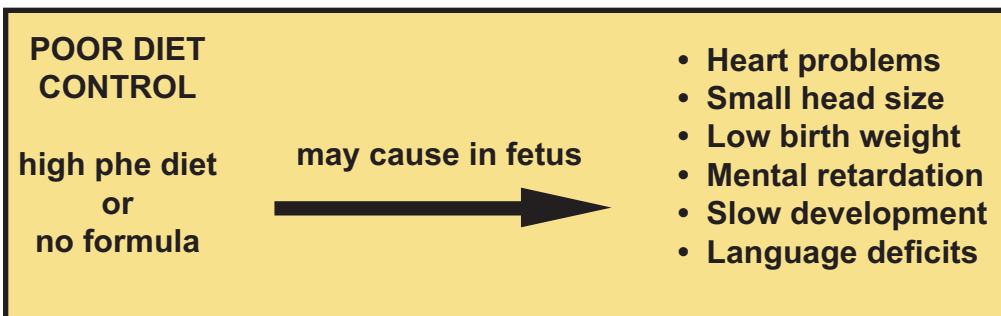
A woman with PKU that is pregnant must eat for two, gaining weight like any other pregnant mother. To gain the proper amount of weight during pregnancy, she must eat foods high in fat while low in protein. Because of the diet limitations, weight gain is often promoted with sugars and fats. Pregnant women with PKU can buy regular foods low in protein avoiding low-fat, low-cal options. Some things to try are: fried foods, butter, mayonnaise, Cool Whip, non-dairy creamers, canned frosting, salad dressing and olive oil. Special low protein products, such as cookies, pizzas, and baked goods can also give you a lot of calories.

## Is taking the formula important?

Yes! The special PKU diet requires taking the formula throughout life. Specifically, for maternal PKU taking the formula is extremely important for having a healthy baby. Because a woman with PKU cannot metabolize the amino acid phenylalanine, she must limit her intake of proteins. Proteins are made of amino acids that are important for the body to grow, and therefore, the body needs to take them in another form. This is where the formula comes in. The formula is made of all the important amino acids except phenylalanine. It provides the necessary proteins needed for a health baby. Without the formula, simply restricting phe intake will not protect the unborn child.

## What may happen to the unborn child if a woman does not maintain strict dietary control (phe restriction and formula)?

If treatment does not start before pregnancy, the main risk is congenital heart defect in the fetus, because the heart forms within the first 30 days the embryo develops. If the pregnant woman with PKU does not manage to maintain strict metabolic control until after the first three months, permanent damage occurs.



## What are some difficulties a pregnant woman with maternal PKU should expect?

- The strict PKU diet and the daily intake of formula may interrupt daily activities and can be frustrating on an every day basis.
- It may be hard to follow the diet and take the formula in social situations and particular in restaurants.
- Women with PKU that are pregnant have the additional discomforts of nausea and vomiting. In addition, their stomachs may be easily upset by the formula or foods they need to include in their diet.
- PKU mothers should also resist high phenylalanine food cravings that are common to pregnancy (peanut butter, ice cream, meat, etc.)

Yes, it can be frustrating. But, it is also a wonderful experience and these challenges are small compared to the risks that not following the diet may cause to your unborn child. Read advice and ideas to cope with these challenges in "make PKU formula part of your life" and "WHAT CAN I EAT?"

Don't let stress and responsibility for maintaining low phe levels take away the joy of bringing a new life in the world. Maintaining strict diet and taking the formula is necessary for the child's health. But you can manage it with planning, determination and support.