Maternal PKU and Pregnancy

The information below will help you determine if having PKU represents an increased fetal risk. With every pregnancy, all women have a 3 to 5 percent chance to have a baby with a birth defect.

Maternal PKU

What is PKU?

PKU stands for phenylketonuria, an inherited condition where the body is missing an enzyme that is needed to break down a protein called phenylalanine, or Phe for short. Since people with PKU cannot digest Phe appropriately, Phe and similar compounds build up in the body. This can lead to problems with brain development. However, treatment with a special diet can decrease the levels of Phe in the body so that this damage does not occur. Babies and children with PKU who are not on the special diet will have mental retardation.

Is there any reason to continue the diet until adulthood?

Currently, medical professionals recommend staying on the diet lifelong to ensure the healthiest development. Some people who stop the diet in early childhood have learning and behavior problems. It is particularly important for females with PKU to stay on the diet, since increased Phe levels during a pregnancy can cause problems for an unborn baby. This is referred to as Maternal PKU effects. Since half of all pregnancies are not planned, it is especially important for women with PKU to maintain the diet even if they are not actively trying to get pregnant.

What effects do high levels of Phe have on a developing baby?

Babies born to mothers with untreated PKU (women who are not on the special diet) may be born smaller, have mental retardation, a heart defect, behavior problems and characteristic facial features.

Is there anything I can do to prevent these effects?

The same diet you were on as a child can reduce your Phe levels, which in turn reduces the chance for your baby to have any of the problems related to Maternal PKU. The goal is to get your Phe levels below 6 mg/dl. Your doctor or health care professional can measure your Phe levels with a blood test. Ideally, dietary control should start before conception, because it may take some women longer than others to get their Phe levels down.

I am 11 weeks pregnant. Will the diet help if I go on it now?

Yes. Your baby continues to grow and the brain develops throughout the pregnancy. So, it is still a good idea to go on the diet and maintain low levels of Phe. However, the first 12 weeks of pregnancy are the critical period for the organs, including the heart, to form. Therefore, starting the diet after the first trimester does not lessen the risk for birth defects.

What does the diet consist of?

Foods containing high amounts of Phe, such as meats, dairy products and nuts need to be replaced with low protein foods such as certain grain products, fruits, and some vegetables. There is also a special low Phe formula to make sure that you will get the essential nutrients. A dietician or other health care professional can provide you with more specific information on the diet.
Is there any way to know if my baby will have problems related to Maternal PKU?

A detailed ultrasound after 18 weeks of pregnancy can look for a heart defect or growth problem. Changes in intelligence, behavior and facial features cannot, however, be determined before a baby is born.

Can I breast feed my baby if I have PKU?

Unless the baby also has PKU, breastfeeding is not a problem for the baby, but some doctors may recommend staying on the special diet while waiting for the baby to be tested for PKU. If you stay on the diet after you deliver, the baby should not be exposed to high levels of Phe. Your doctor can also measure the Phe levels in the baby to make sure they are not elevated after breastfeeding.

Will my baby need to be on the diet?

Your baby will only need to be on the diet if he or she also has PKU. Newborns in all states are tested for PKU before they leave the hospital by testing the levels of Phe in their blood.

What if the father of the baby has PKU instead of the mother?

There have been two small studies that suggest that there is no increased risk for birth defects when the father has PKU. In some men, PKU may reduce their fertility.

What is the chance that my baby will have PKU as I do?

A baby can only have PKU if both the mother and the father carry a specific gene for PKU. A gene contains the instructions for making the proteins that our bodies need to perform their daily functions. Since you have PKU, you will always pass on one non-working gene for PKU to your children. A person who has only one non-working gene for PKU is called a carrier for PKU. Carriers of PKU are healthy.

If the father of the baby does not have PKU and is not a carrier, none of your children will have PKU, but they will all be carriers. However, if you have children with someone who is a carrier of PKU, then there is a 50% chance for each child to have PKU. Finally, if you have children with someone who also has PKU, all of your children will have PKU. Testing to find out if a partner is a carrier of PKU is possible in some families, and if the specific genetic change is found, prenatal testing may also be available. A genetic counselor or other health care professional can provide more information.

May 2000
Copyright by OTIS. Reproduced by permission.

References:


