



MotherToBaby

Medications & More During Pregnancy & Breastfeeding  
Ask The Experts

Fact Sheet

by the **Organization of Teratology Information Specialists (OTIS)**

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## Maternal PKU

In every pregnancy, a woman starts out with a 3-5% chance of having a baby with a birth defect. This is called her background risk. This sheet talks about whether having PKU might increase the risk for birth defects over that background risk. This information should not take the place of medical care and advice from your health care provider.

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

PKU stands for phenylketonuria (fen-ul-ke-toe-NU-re-uh), an inherited condition where the body can not break down (digest) an amino acid called phenylalanine, or Phe for short. Since people with PKU cannot properly digest Phe, Phe can build up in the body. This can lead to problems with brain development and cause intellectual disability and psychiatric disorders. However, treatment with a special diet can lower the levels of Phe in the body so that this brain damage does not occur.

### ***Should the diet be continued in adulthood and pregnancy?***

Yes. Medical professionals recommend staying on the diet lifelong to have the healthiest development. Women with PKU and who are pregnant need to stay on the diet throughout pregnancy. High Phe levels during a pregnancy can cause problems for the developing baby, called Maternal PKU effects. Since half of all pregnancies are not planned, it is important for women with PKU to stay on the diet even if they are not actively trying to get pregnant.

### ***What effects can high levels of Phe have on a developing baby?***

Babies born to mothers with untreated PKU (women who are not on the special diet) are commonly born smaller, have microcephaly (an abnormally small head), intellectual disability, behavior problems, and a higher chance for heart defects and seizures.

The amount of Phe a baby is exposed to during pregnancy is 1.5 – 2 times higher than the amount in mom's blood. This is another reason why staying on a low Phe diet is important before becoming pregnant. Many women find out that they are pregnant between 5 and 8 weeks, which is a very critical period of development. Having a high Phe level at this critical period can increase the chance for the features seen in Maternal PKU.

### ***Is there anything I can do to prevent these effects?***

Yes. Staying on your diet will lower your Phe levels. Lower Phe levels will lower the chance for your baby to have any of the problems related to Maternal PKU. It is recommended that Phe levels be less than 6 mg/dL for at least 3 months before becoming pregnant. Once pregnant, the goal is to keep your Phe levels between 2-6 mg/dL (120 and 360 micromoles/liter).

One study looked at over 550 pregnancies in women with PKU, some of which were on a restricted diet before conception and others who began the diet once the pregnancy was recognized. Babies born to mothers on the special diet before conception or before 8 to 10 weeks of pregnancy had similar brain development as babies born to women without PKU. Women who did not start the diet until after the first trimester (after 12 weeks of pregnancy) had babies who did poorer on developmental tests. Therefore, the special diet should be started as soon as possible in order to increase your chances of having a healthy baby.

Women should work with a nutritionist and geneticist before and during their pregnancy, as they will provide you with more specific information on the diet throughout your pregnancy. Getting enough protein (other than Phe) and vitamins, especially vitamins from the B group like folic acid and vitamin B12, is also important for your baby's development.

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

Yes. Your baby continues to grow and the brain develops throughout the entire pregnancy. Untreated PKU has a direct effect on growth and brain development. 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 lower the chance for birth defects. Consult a dietician and a geneticist as soon as possible when you find out you are pregnant.

***Is there any way to know if my baby will have problems related to Maternal PKU?***

A detailed ultrasound around 18-20 weeks of pregnancy can screen for a heart defect or a growth problem, including microcephaly. However, changes in learning and behavior cannot be seen before a baby is born.

***Will my baby need to be on the diet?***

Your baby will only need to be on the special low phenylalanine diet if he or she also has PKU. In all states and provinces in North America, newborns are tested for PKU through newborn screening before they leave the hospital.

***Can I breastfeed my baby if I have PKU?***

If the baby does not have PKU, breastfeeding is not a problem. If you stay on the diet after you deliver, the baby should not be exposed to high levels of Phe. Your health care provider can also measure the Phe levels in the baby to make sure they are not too high after breastfeeding.

Babies with PKU can also be breastfed, but they need to be followed strictly by a dietician and a geneticist, and their Phe blood level checked to make sure they receive the correct amount of phenylalanine. Alternating breastfeeding with a special PKU formula (with low Phe levels) is usually done. Different approaches are possible, depending on the experience of the medical team taking care of you and your baby. Be sure to talk to your health care team about your breastfeeding questions.

***What if the father of the baby has PKU?***

There have been two small studies that suggest that there is no increased chance for birth defects when the father has PKU. In some men, PKU may reduce their fertility (make it harder for them to get a partner pregnant).

In general, exposures that fathers have are unlikely to increase risks to a pregnancy. For general information, please see the MotherToBaby fact sheet Paternal Exposures and Pregnancy at <https://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/pdf/>.

**References Available By Request**

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