Maternal PKU

This sheet talks about PKU during pregnancy and while breastfeeding. This information should not take the place of medical care and advice from your healthcare 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, it can build up in the body. This can lead to problems with brain development and cause intellectual disability and psychiatric disorders. The treatment for PKU is a special diet that can lower the levels of Phe in the body, so that it does not lead to brain damage or additional medical problems.

Should the diet be continued in adulthood and pregnancy?

Yes. Healthcare providers recommend a person stay on the diet for their entire life in order to have the best chance of avoiding the effects of high Phe levels. Women with PKU who are pregnant need to stay on the diet throughout pregnancy. The amount of Phe a baby is exposed to during pregnancy is 1.5 – 2 times higher than the amount in a pregnant woman’s blood. High Phe levels during a pregnancy can cause problems for the developing baby. This is especially true during the first trimester of pregnancy, which is an important time in the baby’s development. Since half of all pregnancies are unplanned, and because many women find out they are pregnant between 5-8 weeks of pregnancy, it is important for women with PKU to stay on the diet even if they are not actively trying to get pregnant.

I have PKU. Can it make it harder for me to become pregnant?

There is no data to suggest PKU or high levels of Phe make it harder for a woman to become pregnant.

Does having PKU increase the chance for miscarriage?

Miscarriage can occur in any pregnancy. There is no data to suggest PKU or high levels of Phe increase the chance for miscarriage.

Does having PKU increase the chance of birth defects?

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. Women with PKU who follow their diet and take their medical food appropriately are most likely to have normal Phe levels that are not thought to increase the chance of having a child with a birth defect. However, babies born to mothers with PKU who have high levels of Phe (women who are not on the special diet or do not take their medical food appropriately) are at a higher chance of being born smaller, having microcephaly (baby’s head is much smaller than expected), intellectual disability, behavior problems, heart defects, and seizures.

Does having PKU in pregnancy cause long-term problems for the baby?

In general, women with PKU who follow their diet and take their medical food properly have normal Phe levels during pregnancy and do not have children with long-term problems. A baby born to a mother with high Phe levels have a higher chance of having long-term health, behavioral, and/or intellectual problems.

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 dietitian and geneticist before and during their pregnancy, as they will provide specific information on the diet throughout pregnancy. Proper nutrition, especially vitamins from the B group like folic acid and vitamin B12, is also important for your baby’s development.

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 some birth defects and growth issues related to maternal PKU. 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 the United States, newborns are tested for PKU through newborn screening before they leave the hospital. If your baby is not born in a hospital, or is born outside of the United States, talk to your healthcare provider about screening for PKU.

Can I breastfeed my baby if I have PKU?

If a baby does not have PKU, their body is able to breakdown the Phe in breastmilk. It is still important to remain on diet while breastfeeding your baby to minimize exposure to high levels of Phe. Your healthcare 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. They need to be followed closely by a dietitian and a geneticist. Additionally, their Phe blood levels should be checked regularly to make sure their levels of Phe are not too high. Different approaches are possible, depending on the experience of the medical team taking care of you and your baby. For example, your medical team may encourage you to use a mixture of breast milk and a special PKU formula (with low Phe levels). Be sure to talk to your healthcare team about all your breastfeeding questions.

If a man has PKU, could it affect his fertility (ability to get partner pregnant) or increase the chance of birth defects?

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.

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 at https://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/pdf/.

Please click here for references.

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