Phenylketonuria (PKU)

This sheet is about having phenylketonuria (PKU) in pregnancy or while breastfeeding. This information is based on research studies. It should not take the place of medical care and advice from your healthcare provider.

What is phenylketonuria (PKU)?

Phenylketonuria (PKU) is an inherited genetic condition. It is caused by a gene change that lowers the amount of an enzyme in the body needed to process the amnio acid phenylalanine (Phe). Phe is commonly found in food and can build up in the body of people with PKU. Build up can lead to problems with brain development and cause intellectual disability, trouble with attention, and conditions like anxiety or depression. PKU is a health condition that requires lifelong diet changes and / or treatment to avoid buildup of Phe in the body.

I have PKU and would like to get pregnant. What should I talk about with my healthcare providers?

It is very important to talk to your healthcare providers who are familiar with PKU, such as a geneticist, genetic counselor, and a metabolic dietician. The healthcare team can work with you to come up with the best way to treat your condition. Talk with your healthcare team before getting pregnant, when possible. 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 to stay on the diet and treatment even if you are not trying to get pregnant.

During pregnancy, Phe crosses the placenta (the temporary organ that develops during pregnancy and works as the blood connection between the woman who is pregnant and the fetus). Phe levels in the developing fetus can be higher than what is measured in the parent. High levels of Phe during a pregnancy can cause problems for the fetus. Getting Phe levels under control at least 3 months before getting pregnant and throughout pregnancy can reduce the chance of these problems.

The American College of Medical Genetics (ACMG) recommends that women with PKU have Phe levels ≤360 µmol/L before getting pregnant to prevent pregnancy complications and risks to the fetus. They also recommend that women with PAH deficiency who are pregnant maintain Phe levels ≤360 umol/L throughout pregnancy and in the postpartum time for the best maternal and infant outcomes. Talk with your healthcare team about your Phe levels and plans for treatment before, during, and after pregnancy.

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

It is not known if having PKU or high levels of Phe can make it harder to get pregnant.

Does having PKU increase the chance of miscarriage?

Miscarriage is common and can occur in any pregnancy for many different reasons. Some studies have found that having PKU with high levels of Phe can increase the chance of miscarriage.

Does having PKU increase the chance of birth defects?

Birth defects can happen in any pregnancy for different reasons. Out of all babies born each year, about 3 out of 100 (3%) will have a birth defect. We look at published research to try to understand if a certain exposure, like PKU, might increase the chance of birth defects or other problems in a pregnancy.

Women with PKU who are pregnant and who have high levels of Phe have a higher chance of having a baby with a very small head (microcephaly) and heart defects. Women who keep their Phe levels in an appropriate range before and during the pregnancy are not expected to have a higher chance of having a baby with a birth defect. Talk with your healthcare provider about how to get to and stay at your target levels of Phe.

Does having PKU increase the chance of other pregnancy-related problems?

Women who keep their Phe levels in an appropriate range before and during the pregnancy are not expected to have a higher chance of pregnancy-related problems such as preterm delivery (birth before week 37) or low birth weight (weighing less than 5 pounds, 8 ounces [2500 grams] at birth).

Women who have high levels of Phe have a higher chance of having a baby that is born smaller than expected. It is also important to not let the Phe levels get too low. Some studies show that very low Phe levels (below 100 micromoles/liter) can cause growth problems as well, especially during the second and third trimesters.

Does having PKU in pregnancy affect future behavior or learning for the child?

Babies born to women with PKU and who have high Phe levels have a higher chance of having intellectual disability, behavioral problems, and seizures. Chances of long-term problems are lower if the diet is started before getting pregnant or as soon as possible once pregnancy starts. Women with PKU who start their diet after the first trimester (after 12 weeks of pregnancy) had babies who did poorer on developmental tests. It is suggested to start the special diet as soon as possible.

What screenings or tests are available to see if my pregnancy has birth defects or other issues?

Prenatal ultrasounds can be used screen for some birth defects, such as small head size. Ultrasound can also be used to monitor the growth of the pregnancy. Talk with your healthcare provider about any prenatal screenings or testing that are available to you. There are no tests available during pregnancy that can tell how much effect there could be on future behavior or learning.

Breastfeeding with PKU:

A woman with PKU can breastfeed. Research suggests if a baby does not have PKU, the baby's body is expected to be able to breakdown the Phe in breast milk. It is important to remain on a special diet while breastfeeding 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 that have PKU can be breastfed. They need to be followed closely by a metabolic team (usually a dietitian and a geneticist/genetic counselor). Infant blood levels should be checked regularly to make sure that levels of Phe are not too high. Different approaches are possible. One example might be using a mix of breast milk and a special PKU formula (with low Phe levels). Be sure to talk to your healthcare providers about the best way to feed your baby, as well as all your breastfeeding questions.

If a man has PKU, can it affect his fertility or increase the chance of birth defects?

PKU might reduce a man's fertility (ability to get a woman pregnant). Small studies suggest no increased chance of birth defects when a man has PKU. In general, exposures that men have are unlikely to increase risk to a pregnancy. For more information, please see the MotherToBaby fact sheet Paternal Exposures at https://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/.

Please click here for references.