

Phenylketonuria (PKU)

This sheet is about having phenylketonuria (PKU) in a pregnancy or while breastfeeding. This information should not take the place of medical care and advice from your healthcare providers.

What is PKU?

PKU stands for phenylketonuria, an inherited condition where the body cannot break down an amino acid called phenylalanine, or Phe for short. Phe is commonly found in food. Since people with PKU cannot properly digest Phe, it can build up in the body. This build up can lead to problems with brain development and cause intellectual disability, difficulties with attention, and mental health conditions like anxiety or depression.

PKU is a health condition that requires lifelong treatment. PKU is treated with a special diet and use of medical foods that can lower the level of Phe in the body to avoid buildup of Phe. Along with the special diet, PKU has also been treated with certain medications such as sapropterin (Kuvan®), pegvaliase-pqpz (Palynziq®), and large neutral amino acids (LNAA). Healthcare providers recommend a person stay on the diet and or treatment for their entire life in order to have the best chance of avoiding the problems caused by high Phe levels.

I have PKU and would like to become pregnant. What should I discuss with my healthcare providers?

It is very important to talk to your healthcare providers who are familiar with PKU, such as a geneticist and a metabolic dietician. The healthcare team can work with you to come up with the best diet and treatment plan. Start this conversation before getting pregnant, if possible. Since half of all pregnancies are unplanned, and because many people 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 a pregnancy, Phe crosses the placenta. (The placenta is a temporary organ that develops during pregnancy and works as the blood connection between you and your baby.) Phe levels in the developing baby can be higher than what is measured in the parent. High Phe levels during a pregnancy can cause problems for the developing baby. Getting Phe levels under control at least 3 months before getting pregnant and throughout pregnancy can help prevent these problems.

I have PKU. Can it make it harder for me to become 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 for 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 increases the chance of miscarriage; however, other studies did not show a higher chance.

Does having PKU increase the chance of birth defects?

Every pregnancy starts out with a 3-5% chance of having a birth defect. This is called the background risk. Based on the studies reviewed, people who are pregnant with PKU and who follow their diet / treatment plan to keep Phe levels in the body low before and during the pregnancy do not have a higher chance of having a baby with a birth defect or intellectual disability. However, people who are pregnant with PKU and who have high levels of Phe have a higher chance of having a baby a very small head (microcephaly) and heart defects.

Does having PKU cause pregnancy complications?

People who are pregnant with PKU and who follow their diet and treatment plan to keep Phe levels in the body low before and during the pregnancy are not expected to have a higher chance of having pregnancy complications such as preterm delivery (birth before week 37) or low birth weight (weighing less than 5 pounds, 8 ounces (2500 grams) at birth). However, people who are pregnant with PKU and who have high levels of Phe have a higher chance of having a baby that is born smaller than expected. However, it is also important to not let the Phe levels get too low, as 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?

People who are pregnant and have PKU who follow their diet and treatment plan to keep Phe levels low before and during pregnancy are not expected to have a child with long-term problems. Babies born to people who are pregnant with PKU and have high Phe levels have a higher chance of having intellectual disability, behavioral problems, and seizures. Chances for long-term problems are lower if the diet is started before getting pregnant or as soon as possible once pregnancy starts. People with PKU who start their diet 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.

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 a pregnancy that can tell if there has been any effect on behavior or ability to learn.

I have PKU. Will my baby have PKU and need to be on the diet?

PKU is an inherited genetic disorder. For a baby to inherit PKU, both parents would need to carry gene variants that cause PKU and the baby would need to inherit 2 disease causing gene variants (one from each parent). The healthcare provider may refer people with PKU to a genetic counselor to discuss the chances that the baby will get PKU and available screening and or testing for your partner and/or the baby during pregnancy. Most babies born to people with PKU do not have PKU. High or low Phe levels during the pregnancy do not cause PKU in a baby.

In the United States, newborns are tested for PKU through newborn bloodspot screening before they leave the hospital. If the baby is not born in a hospital, or is born outside of the United States, talk to the healthcare provider about screening for PKU.

Breastfeeding while I have PKU:

A person with PKU can breastfeed. PKU is not transmitted in breastmilk.

If a baby does not have PKU, the available data suggests that the baby's body is able to breakdown the Phe in breast milk. It is important to remain on a special diet while breastfeeding the 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 that have PKU can be breastfed. They need to be followed closely by a metabolic team (usually a dietitian and a geneticist). Additionally, the Phe blood levels should be checked regularly to make sure that 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 male has PKU, can it make it harder to get a partner pregnant or increase the chance of birth defects?

There have been two small studies that suggest no increased chance for birth defects when the father has PKU. In some men, PKU might reduce their fertility (make it harder to get a partner pregnant). In general, exposures that fathers or sperm donors have are unlikely to increase the risk to a pregnancy. For more information, please see the MotherToBaby fact sheet Paternal Exposures at <https://mothertobaby.org/fact-sheets/paternal-exposures-pregnancy/>.

Selected references.

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