Maternal 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 psychiatric disorders 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. Experts recommend keeping Phe levels in the body between 2 and 6 milligrams/deciliter (120-360 micromoles/liter).

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. Your healthcare team can work with you to come up with the best diet and treatment plan. High Phe levels during a pregnancy can cause problems for the developing baby and getting Phe levels under 6 milligrams/deciliter at least 3 months before getting pregnant can help prevent these problems. 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. If your pregnancy is unplanned, contact your healthcare providers as soon as you learn you are pregnant. Once pregnant, you should be closely followed by your geneticist and dietician/nutritionist as well as a high risk pregnancy doctor (maternal fetal medicine specialists). They can measure the levels of Phe in your body often during pregnancy.

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

Based on the data available, it is not known if having PKU or high levels of Phe can make it harder to get pregnant.

I just found out I am pregnant. Should I stop medications I take for PKU?

Talk with your healthcare providers before making any changes to how you take your medication(s). You can also contact MotherToBaby with questions on your specific medication.

Does having PKU increase the chance for miscarriage?

Miscarriage can occur in any pregnancy. 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/getting 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. 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 are not expected to have a higher chance of having a child 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 with intellectual disability, a very small head (microcephaly), and heart defects.

Babies born to people who start the diet before getting pregnant have similar chance for heart defects and brain development as babies born to people without PKU. 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.
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. 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. However, it is also important to not let the Phe levels get too low either because 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/getting PKU in pregnancy cause long-term problems?
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 children 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 diet is started before getting pregnant or as soon as possible once pregnancy starts.

Is there any way to know if my baby will have problems related to maternal PKU?
A detailed ultrasound as well as an echocardiogram (special ultrasound of the baby’s heart) 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.

I have PKU. Will my baby have PKU and need to be on the diet?
Most babies of people with PKU are not born with PKU. Whether the baby gets PKU is not related to Phe levels during the pregnancy. PKU is an inherited genetic disorder and the only way a baby would get PKU is if both parents carry genes for PKY and the baby inherits 2 disease causing gene variants (one from each parent). Your healthcare provider may refer you to a genetic counselor to discuss the chances that your baby will get PKU and available testing for your partner and/or the baby during pregnancy.

In the United States, newborns are tested for PKU through newborn bloodspot 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. Your baby will only need to be on the special low Phe diet if they also have PKU.

Can I breastfeed while I have PKU?
If a baby does not have PKU, their body is able to breakdown the Phe in breast milk. 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 that have 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.

I have PKU. Can it make it harder for me to get my 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 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/.

Please click here for references.