

Phenylketonuria (PKU)

(Sounds like FEEN-IL-KE-TON-U-RE-AH)



What is newborn blood spot screening?

This is a blood test, from a heel poke, that is done a day or 2 after babies are born to test for treatable conditions including phenylketonuria (PKU) that you, your doctor, or midwife can't see by just looking at your baby. This screening test is different from those done during pregnancy.



What does it mean if my baby needs more tests for PKU?

A baby needs to have more testing for PKU when their screening test result is abnormal or positive. This doesn't mean your baby has PKU. It means your baby has a higher chance of having the condition and will need more tests to find out if they have PKU or not.

It's normal to feel worried if your baby needs more testing. Your specialists and other healthcare providers are there to support you. They'll explain what will happen and answer your questions.

What is PKU?

PKU is a metabolic condition that affects how your body uses protein from the food you eat to make energy. This includes breastmilk and regular formula. With PKU, your body can't break down 1 of the building blocks of protein called phenylalanine (PHE). PHE then builds up in your body and can cause serious health problems, such as brain damage.

What causes PKU?

PKU is genetic. This means babies with PKU are born with a gene from both parents, which has a change in it. This change stops the gene from working properly. PKU isn't caused by anything that happened during pregnancy.

There may be no signs of PKU at birth. It's important to remember that this condition can't be seen by looking at your baby.



What can I expect with more testing?

Everything that will happen with more testing will be explained to you. Your baby may need to have their blood and urine tested to check for PKU.

This testing may be done at a clinic in Edmonton or Calgary that specializes in PKU.

What if more testing shows my baby has PKU?

If testing shows that your baby has PKU a specialized healthcare team will work with you to make a care plan for your baby. You and your baby will get the best care and support possible.

How is PKU treated?

Babies with PKU usually go on a low-protein diet and drink a special formula. The specialized healthcare team will give you detailed instructions about how to manage your baby's health.



