

Phenylketonuria (PKU) Screening

What is a PKU screening test?

A PKU screening test is a blood test given to newborns 24–72 hours after birth. PKU stands for **phenylketonuria**, a rare disorder that prevents the body from properly breaking down a substance called phenylalanine (Phe). Phe is part of **proteins** that are found in many foods and in an artificial sweetener called aspartame.

If you have PKU and eat these foods, Phe will build up in the blood. High levels of Phe can permanently damage the **nervous system** and brain, causing a variety of health problems. These include **seizures**, psychiatric problems, and severe intellectual disability.

PKU is caused by a **genetic mutation**, a change in the normal function of a gene. Genes are the basic units of heredity passed down from your mother and father. For a child to get the disorder, both the mother and father must pass down a mutated PKU gene.

Although PKU is rare, all newborns in the United States are required to get a PKU test.

- The test is easy, with virtually no health risk. But it can save a baby from lifelong brain damage and/or other serious health problems.
- If PKU is found early, following a special, low-protein/low-Phe diet can prevent complications.
- There are specially-made formulas for infants with PKU.
- People with PKU need to stay on the protein/low-Phe diet for the rest of their lives.

Other names: PKU newborn screening, PKU test

What is it used for?

A PKU test is used to see if a newborn has high levels of Phe in the blood. This may mean the baby has PKU, and more tests will be ordered to confirm or rule out a diagnosis.

Why does my baby need a PKU screening test?

Newborns in the United States are required to get a PKU test. A PKU test is usually part of a series of tests called a **newborn screening**. Some older infants and children may need testing if they were **adopted** from another country, and/or if they have any symptoms of PKU, which include:

- Delayed development
- Intellectual difficulties
- A musty odor in the breath, skin, and/or urine
- Abnormally small head (microcephaly)

What happens during a PKU screening test?

A health care provider will clean your baby's heel with alcohol and poke the heel with a small needle. The provider will collect a few drops of blood and put a bandage on the site.

The test should be done no sooner than 24 hours after birth, to ensure the baby has taken in some protein, either from breast milk or formula. This will help ensure the results are accurate. But the test should be done between 24–72 hours after birth to prevent possible PKU complications. If your baby wasn't born in the hospital or if you left the hospital early, be sure to talk to your child's health care provider to schedule a PKU test as soon as possible.

Will I need to do anything to prepare my baby for the test?

There are no special preparations needed for a PKU test.

Are there any risks to the test?

There is very little risk to your baby with a needle stick test. Your baby may feel a little pinch when the heel is poked, and a small bruise may form at the site. This should go away quickly.

What do the results mean?

If your baby's results were not normal, your health care provider will order more tests to confirm or rule out PKU. These tests may include more blood tests and/or [urine tests](#). You and your baby may also get [genetic tests](#), since PKU is an inherited condition.

If the results were normal, but the test was done sooner than 24 hours after birth, your baby may need to be tested again at 1 to 2 weeks of age.

Learn more about [laboratory tests](#), [references ranges](#), and [understanding results](#).

Is there anything else I need to know about a PKU screening test?

If your baby was diagnosed with PKU, he or she can drink formula that does not contain Phe. If you would like to breastfeed, talk your health care provider. Breast milk does contain Phe, but your baby may be able to have a limited amount, supplemented by the Phe-free formula. Regardless, your child will need to stay on a special low-protein diet for life. A PKU diet usually means avoiding high-protein foods such as meat, fish, eggs, dairy, nuts, and beans. Instead, the diet will probably include cereals, starches, fruits, a milk substitute, and other items with low or no Phe.

Your child's health care provider may recommend one or more specialists and other resources to help you manage your baby's diet and keep your child healthy. There are also a variety of resources available to teens and adults with PKU. If you have PKU, talk to your health care provider about the best ways to take care of your dietary and health needs.