Phenylketonuria (PKU)

Does this test have other names?

PKU screening, Guthrie assay, PKU test

What is this test?

This is a blood test to screen newborns for phenylketonuria (PKU). PKU is a condition that can cause brain damage and severe intellectual disability if it goes untreated. The problems often appear in the first year of life, causing babies to appear abnormally sleepy. They may have trouble feeding and develop a red, itchy rash similar to eczema. Such babies typically have lighter skin and hair than family members who don't have the condition.

PKU is an inherited (genetic) condition. People with PKU don't have the enzyme needed to process a substance called phenylalanine. This substance is an amino acid that is a part of proteins found in many foods. Without the enzyme to break it down, phenylalanine can build up to dangerous levels in the body. People with PKU also release a substance called phenylacetic acid in their urine and sweat. If PKU isn't treated, people with PKU may have a musty odor. Starting in infancy and all through their life, people with PKU must follow a diet that limits how much phenylalanine they can eat.

The link between PKU and intellectual disability has been known since the 1930s. In fact, PKU was the first condition that was screened for in newborns. All U.S. states screen newborn babies for PKU. This means that almost all cases are now found and treatment started at birth.

Why does my child need this test?

Your child may need this test because finding and starting treatment of PKU in a newborn can prevent intellectual disability and other developmental problems in your child. If your child has a controlled, low-protein diet that carefully limits phenylalanine in the first weeks of life and beyond, they are likely to live a healthy life.

Even though most babies with PKU are diagnosed soon after birth, screening for PKU should be considered for any child who has an intellectual disability or is developmentally delayed. Some babies adopted from other countries may also need to be screened for PKU and other inherited illnesses in the first year of life.

What other tests might my child have along with this test?

Newborns are also tested for other metabolic birth defects often before they leave the hospital.

What do my child's test results mean?

Test results may vary depending on your child's age, gender, health history, and other things. Your child's test results may be different depending on the lab used. They may not mean your child has a problem. Ask your healthcare provider what your child's test results mean for them.

The test screens for blood levels of phenylalanine. Normal levels of phenylalanine in the blood are less than 2 milligrams per deciliter (mg/dL). More than 4 mg/dL of phenylalanine in the blood is considered high and may mean your child has PKU. The test will be first done after your baby is 24 hours old, then may be repeated when your baby is 7 to 14 days old.

How is this test done?

Babies are often screened for PKU with a heel-prick test. This is done by getting a few drops of blood from the infant's heel.

A urine test is an alternative to the heel prick. The healthcare provider will collect a sample of your baby's urine.

Does this test pose any risks?

Having a heel-prick test carries some risks. These include bleeding, infection, or bruising. When the needle pricks your baby's heel, they may feel a slight sting or pain. Afterward, the site may be sore.

What might affect my child's test results?

The blood test may give a false-positive or false-negative result in certain cases:

- Your baby is premature or has liver problems. This could lead to a false-positive result because certain liver enzymes have not fully started working.
- Your baby has feeding problems, such as vomiting. This could give a false-negative result.

Medicines, such as aspirin or antibiotics, may affect the results of the urine test for PKU.

How do I get my child ready for this test?

The test should not be done before 24 hours after birth. If you are breastfeeding, be sure your healthcare provider knows about all medicines, herbs, vitamins, and supplements you are taking. This includes medicines that don't need a prescription and any illegal drugs you may use.