Newborn Screen Blood Test

State law requires that all babies born in Ohio have their blood drawn and tested. This is called a Newborn Screen Blood Test. This used to be called a PKU test. The Newborn Screen Blood Test checks to see if your baby has certain disorders (see below).

Your baby may look healthy, even if he or she has one of these disorders. If your baby has one of these disorders and it is not found early, the baby can develop serious health problems. This blood test will tell if your baby has a high chance of having developing these disorders. Newborn Screen Blood Test cannot tell for sure whether the baby has the disorder, so it is important for babies who test positive to have further follow-up tests. Many times, the follow-up tests will show that the baby actually does not have the disorder.

How the test is done

All infants are screened before they are released from the hospital where they were born.

If your baby goes home from the hospital before he or she is 24 hours old, the test will have to be repeated. **It is important that the blood sample be collected after the child is 24 hours old and before the 5th day of life.**

- The test is done with 5 drops of blood collected from the heel of the baby and dripped on a special paper (Picture 1). All the newborn screens are sent to the Ohio Department of Health Laboratory.
- When a premature or sick baby is transferred from another hospital, the doctors and nurses at Nationwide Children’s Hospital make sure the needed Newborn Screen Blood Test or follow-up tests are done.
• You will not be notified of the results unless further testing is needed.

• The baby’s primary care doctor is responsible for keeping the test results in your baby’s medical record.

The Ohio Newborn Screen Blood Test checks for more than 36 different disorders. These are a few examples:

**Amino acid disorders** – These disorders are caused by a problem with the body’s ability to properly use certain amino acids found in food. Poisonous chemicals build up in the body starting soon after birth and can damage the brain and other organs. Brain and organ damage and other health problems may occur. Special diets or supplements may help treat these problems. The Ohio newborn screen tests for 11 amino acid disorders.

**Fatty acid disorders** – These disorders interfere with the body’s ability to turn fat into energy. Babies with fatty acid disorders can have seizures, extreme weakness, and heart and breathing problems. Special diets, eating frequently, and medicine may help prevent symptoms. The Ohio newborn screen tests for 9 fatty acid disorders.

**Organic acid disorders** – These disorders prevent the body from removing certain waste products from the blood. This can lead to vomiting, low blood sugar, coma or death. Treatment may include a special diet and medicine. The Ohio newborn screen tests for 12 organic acid disorders.

**Biotinidase deficiency** – This disorder occurs when babies cannot properly use the vitamin biotin. Problems with skin rashes, seizures, hearing loss, or mental retardation may be prevented by adding extra biotin to the diet.

**Congenital adrenal hyperplasia (CAH)** – This disorder occurs when the body does not make certain hormones produced by the adrenal glands. Babies with this condition can develop life threatening episodes of dehydration and coma. Baby girls can have abnormal-looking genitalia. Medicine may help prevent life-threatening complications of this condition.

**Congenital hypothyroidism** – This disorder occurs when the body does not make enough thyroid hormone to keep the baby growing and developing. Medicine may help prevent growth problems and mental retardation.

**Cystic fibrosis** – This disorder is an inherited disease of the lungs and digestive system that can cause repeated chest infections and malnourishment. Early detection may improve growth and decrease risk of infections.

**Galactosemia** – This disorder occurs when the body cannot break down part of milk sugar (galactose). A special diet without milk sugar may prevent brain and liver damage.
Sickle cell disease and other hemoglobinopathies – These are disorders that affect red blood cells. Some of these disorders can cause severe pain, frequent infections, other serious health problems or even death. Taking antibiotics daily greatly lowers the chance of infection and other problems. Babies diagnosed with sickle cell disease will be referred to the Sickle Cell Treatment Center at Nationwide Children’s Hospital.

Severe combined immunodeficiency – This includes a group of rare but serious immune disorders. The body is unable to make certain cells that protect the body from infection. Untreated babies develop life-threatening infections due to bacteria, viruses and fungi. Treatment can reduce the threat of infections.

Lysosomal storage disorders – Lysosomal storage disorders are conditions that occur when the body does not make enzymes needed to break down large molecules into smaller usable substances. The large molecules build up in the cells and may damage the brain, heart, liver and other organs. Treatments may include enzyme replacement or bone marrow transplant.


If you have any questions, be sure to ask your child’s doctor or nurse.

For directions to the nearest Laboratory Service Center, please call Laboratory Services at (800) 934-6575 or visit NationwideChildrens.org/Lab.