Newborn Screening Tests

What are newborn screening tests?

Newborn screening tests are blood tests given to infants just after they are born. These tests check for certain disorders before they cause serious damage. Newborns can then be treated to prevent problems later in life. State departments of health provide the tests. Different states have different rules about which tests are done. Newborn screening tests are not perfect. Sometimes they show that a child has a disease that the child does not actually have. All children who test positively for a disease should be tested again. Rarely, the tests do not identify children who actually do have the disease.

What diseases are tested for?

All states in the U.S. test for 4 diseases:

- Hypothyroidism. If babies do not have enough thyroid hormone, they may not develop normally.
- Phenylketonuria (PKU). This can cause mental retardation if not treated early.
- Galactosemia. This condition can cause blindness, mental retardation, and growth problems if not treated.
- Hemoglobinopathy, including sickle-cell disease. Blood diseases may cause newborns to be at risk for dangerous infections.

Many but not all states test for:

- Homocystinuria. This disorder can cause mental retardation, blood clotting problems, and skeletal problems.
- Congenital adrenal hyperplasia. This disorder may cause death if not treated.
- Biotinidase deficiency. This disorder may lead to seizures, hearing loss, mental retardation, and problems with the immune system.

If these rare diseases are diagnosed early, they can be treated. Some can be completely cured.

Some states test for amino acid, organic acid, and fatty acid oxidation defects. In other states these tests are only available through private testing labs. Early diagnosis and treatment of these disorders may help to prevent

serious problems like mental retardation. Ask your health care provider about these screening tests.

How are the tests done?

Your child's health care provider makes a tiny cut in the baby's heel to get a small amount of blood to test. Well infants are usually tested just before they go home from the hospital, but not later than 72 hours after birth. Sick or premature infants are tested at 1 week of age, or earlier if a screenable disease is suspected.
If a test suggests your child has a disease, the health department will contact you and your baby's doctor. If the tests do not show any diseases, you will generally not be contacted. Your baby's doctor usually has copies of the newborn screening test results.

If your baby needs a blood transfusion, blood tests should be done before the transfusion.

Some states provide a second set of newborn screening tests between 1 and 2 weeks of age. This is important if the newborn leaves the hospital less than 24 hours after birth.

Parents may refuse to have their newborn screened because of religious beliefs or personal beliefs. Parents who refuse to have the testing done must sign waiver forms.

Additional information on newborn screening is available from your health care provider or from the state health department.

*NOTE: This information is provided as a public educational service. The information does not replace any of the instructions your physician gives you. If you have a medical emergency please call 911 or call the Hospital at (208) 529-6111. If you have questions about your child's care, please call Idaho Falls Pediatrics at (208) 522-4600.