Pediatric Associates
OF FAIRFIELD

Newborn Screening Tests

What are newborn screening tests?

Newborn screening tests are blood tests given to infants within a few days after birth. These tests check for disorders that could cause serious harm and can only be found by blood tests. The tests are provided by your state's department of health. Each state has rules about which tests are done.

Newborn screening tests are usually accurate, but they are not perfect. Sometimes they show that a child has a disease that the child does not have. All children who test positive for a disease should be tested again. It is rare for a child to test negative when he does have a disease.

What diseases are tested for?

All states in the U.S. test for these kinds of diseases:☐

Hemoglobin disorders such as sickle cell anemia, hemoglobin C, and otherblood disorders. A severe shortage of normal red blood cells may cause weakness, shortness of breath, or even heart failure.

Endocrine disorders. Certain disorders that affect the adrenal glands and thyroid glands can seriously affect a baby and are able to be treated with medicines.

Cystic fibrosis. This disease affects the lungs and other organs and is helped by early treatment.

Phenylketonuria (PKU). This can cause intellectual disability, which used to be called mental retardation, if not treated early. A child with an intellectual disability may be slow to learn or may never learn basic skills such as reading, problem solving, math, telling time, social skills, and self-care.

Galactosemia. This condition can cause blindness, growth problems, and intellectual disability if not treated.

Biotinidase deficiency. This condition may lead to seizures,

hearing loss, intellectual disability, and problems with the immune system.

Most states also screen for a variety of fatty acid, organic acid and amino acid disorders.

If these rare diseases are diagnosed early, some of them can be effectively treated.

How are the tests done?

Your child's healthcare 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 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 gets copies of the newborn screening test results.

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 or personal beliefs. Parents who refuse to have the testing done must sign forms stating they refuse the tests.

Hearing tests are also part of newborn screening in most states. The newborn needs to be quiet or asleep for this test. The test measures brain waves that result when a sound is made. If hearing loss is not treated early, speech, language and learning can be affected.

You can get more information on newborn screening from your healthcare provider or from the state health department.