Newborn Screening

All states in the U.S. have some type of newborn screening program. Tests are done to detect certain serious diseases as soon as possible after the baby is born. For many of these diseases, early treatment can avoid serious health consequences for your baby. Screening requires only a simple blood test. In most states, screening is required by law. Call our office if you have questions about these important screening tests.

What is newborn screening?

There are many types of congenital diseases (present from birth), a large number of which are genetic (inherited). Genes contain information that determines our physical and mental characteristics. Genetic diseases may result when a baby inherits abnormal genes from one or both parents. Many families don't know about the abnormal genes until they have an infant diagnosed with a genetic disease. Many congenital and genetic diseases have serious complications (such as mental retardation) that can only be prevented if they are recognized and treated as soon as possible after birth.

For this reason, all states recommend certain screening tests for all newborn infants. The list of diseases tested for varies among states. All states test for phenylketonuria (PKU, a type of metabolic disease), and congenital hypothyroidism (a disease leading to low thyroid hormone levels). Both of these diseases can cause mental retardation, which can be prevented through proper treatment.

Most states screen for other diseases as well. The tests are done using a single blood sample obtained from a small puncture in the baby's heel. In most states, newborn screening is required by law.

What diseases will my baby be screened for?

The exact list of diseases varies by state. In addition to the required tests, your doctor may recommend other tests, depending on your family history, the baby's condition at birth, and other factors.

The number of screening tests varies widely. Some states screen for only a few diseases, whereas others screen for dozens. New automated tests have been developed in recent years, making it possible to screen for many diseases at the same time.

All states screen for:

- *Phenylketonuria* (PKU). Babies with PKU don't have a certain enzyme needed to metabolize (use) an amino acid called phenylalanine. They need a special diet to prevent mental retardation.
- *Congenital hypothyroidism*. In babies with this disease, the thyroid gland doesn't produce normal levels of thyroid hormones. These infants need treatment with thyroid hormones to prevent problems with growth, development, and mental ability.

Most states screen for:

- *Galactosemia*. Babies with galactosemia don't have a certain enzyme needed to metabolize a sugar called galactose, which is found in milk or regular infant formula. Treatment may be needed to prevent mental retardation and other complications. (Some forms of galactosemia don't cause these complications.)
- *Sickle cell disease*. An inherited blood disease that causes anemia (low levels of hemoglobin, which carries oxygen in the blood) and other medical problems. It is most common in African Americans but can occur in other racial/ethnic groups as well. Your baby may also be screened for sickle cell trait, a gene abnormality that is very common in African Americans. Although it's important to know whether or not your family has sickle cell trait, it is *not* a disease and causes no medical problems. If a child inherits this trait from both parents, he or she will have *sickle cell anemia*.
- *Hearing loss*. Most states require a hearing test for all infants. Regardless of the cause, children with hearing loss need early intervention to promote normal development.

Some states screen for other genetic and congenital diseases, such as adrenal hyperplasia, biotidinase deficiency, and cystic fibrosis.

How common are these diseases?

Fortunately, many of these diseases are rare. Of the 4 million infants who are screened each year in the United States, only about 3000 are found to have a congenital disease, excluding hearing loss. That means that only about 1 in 1300 newborns screened has any abnormal results. If your baby does have a congenital disease, then early detection and treatment provide the best chance to avoid future problems with health and development.

Does my baby have to have these tests?

In most states, newborn screening is required by law. In some states, parents can choose not to have their baby tested if it goes against their religious beliefs.

Some parents have other reasons for not wanting their baby to be tested. However, even if screening is not required by law, most pediatricians would recommend having the tests done. The screening tests are simple, are very unlikely to do any harm to the baby, and have a low but real

chance of detecting a serious disease that needs immediate treatment.

Many families have concerns about privacy related to congenital and genetic diseases. Most states have laws protecting the parents' and children's confidentiality.

Are there any risks of testing?

Routine newborn screening has little or no risk for your infant. Most of the time, all tests can be performed using a single blood sample; your baby will feel a little pain from a pinprick on the heel. A simple hearing test may be performed as well.

The biggest risk is not testing! Without newborn screening, there is a small but real chance of complications occurring from unrecognized diseases.

What if there is an abnormal result?

- Every state has a system for informing the hospital, doctor, or parent about any abnormal newborn screening results. If any of the results are abnormal, further tests are performed to make sure the original result was correct.
- Depending on the disease identified, we will probably recommend a visit to a specialist (for example, a medical geneticist) for full diagnostic testing. If any genetic (inherited) diseases are suspected, the parents and possibly other family members may be tested as well.
- If a genetic disease is detected, genetic counseling will be arranged for your family. This involves education about your family's gene abnormality and the resulting disease, including the risk of disease for other children or family members. Genetic testing may be recommended for parents and other family members.
- Treatment depends on the specific disease. For example:
  - Children with PKU need a special type of formula immediately and need to follow a special diet for the rest of their lives.
  - Children with congenital hypothyroidism or congenital adrenal hyperplasia need treatment to replace the hormones that their bodies are missing.
  - Children with sickle cell disease need lifelong medical follow-up examinations. Sickle cell trait does not cause any illness, but it does mean there is a genetic abnormality that could be passed on to future children. Genetic counseling is generally recommended.
- The one thing that all of these diseases have in common is that early recognition and treatment are needed to prevent complications.

When should I call your office?

Call our office if:

- You have any questions or concerns about newborn screening.
- You have any questions about the findings of your child's screening tests, especially if there are any abnormal results.

Please type your custom instructions and/or office contact information here.