MOUNT AUBURN HOSPITAL

Newborn Screening for your Baby's Health

Why is my baby tested?

To help ensure that your baby will be as healthy as possible. A simple, quick, and economical blood test provides important information about your baby's health that you or even your doctor might not know otherwise. The Newborn Screening Program identifies the few infants who may have one of the several rare birth defects and alerts the doctor to this possibility. With early diagnosis and medical treatment, serious illness can usually be prevented.

For how many disorders is my baby tested?

Five. They are listed at the end of this sheet. Although these disorders are rare, they are usually serious. Some may be life threatening while others may slow down physical development or cause mental retardation or other problems.

The disorders can affect a child very early in life, some in the first few days or weeks of life, and others in the first few years. For this reason, prompt testing and diagnosis are important.

My baby seems very healthy. Are the tests necessary?

Yes, they are. Most infants with birth defects identified by the Newborn Screening Program show no obvious signs of disease immediately after birth. In each of these disorders, there is an "invisible" problem in one of the many chemicals which are produced naturally in the baby's body.

With special laboratory tests, the Newborn Screening Program can identify the infant who may have one the disorders and can alert the doctor to the need for special care of the infant. Usually, this can be done before the problem has time to cause damaging effects.

We don't have any birth defects in our family.

Parents who have already had healthy children do not expect any problem with birth defects, and they are almost always right. These disorders are quite rare and the chances are excellent that your child will not have one of the disorders. However, the few children who are born with these problems are generally from healthy families. By testing every baby at birth, we can be sure that each infant who has a disorder will be identified and started on early treatment.

How is my baby tested?

All of the tests are performed on one tiny sample of blood obtained from the baby's heel. The sample is usually obtained on the day of discharge from the hospital. The blood is allowed to dry on a piece of absorbent paper, which is then sent for testing to a special laboratory.

Form 950c-D, Rev. 1/8/2019, MAH

Will I be told if the tests show no birth defects?

Your doctor or health clinic will be informed when the tests are completed. Generally, parents are notified only if there is a problem, but you can ask about the results when you bring the baby to the doctor or clinic for a regular checkup.

Although, "no news is good news" it is important that these tests provide information only about five rare chemical disorders (listed at the end of this sheet). Even if an infant is free of disorders, there may be other medical problems for which we cannot test by these methods.

So, it is very important for your baby to have regular checkups and good general medical care.

Does a "Retest" mean my baby may have a birth defect?

Not necessarily. Retesting may be required for a number of reasons. The most frequent reason is that the first sample contained too little blood for all five tests. This does not mean that there is anything wrong with your baby. It simply means that another sample must be obtained so that the complete set of tests can be performed.

On the rare occasions when the first tests indicate a possible problem, the results are not considered final. Instead, a new blood sample is obtained and the tests are repeated. As a general rule, only when a child's test is unusual for the second time will the doctor discuss the need for further evaluation. Only on very rare occasions, because of the potential severity of a particular disorder, will the doctor insist on treating child immediately while waiting for the results of the second series of tests.

If you are asked to have your child retested, please act quickly so the repeat test can be completed and final results obtained while the baby is still very young.

What if my baby has one of these disorders? Can it be cured?

Because most of these disorders are "inborn" chemical problems, they cannot be "cured," just as eye color or height cannot be permanently changed. However, the serious effects of the disorder can be lessened and often completely prevented if a special diet or other medical treatment is started early.

If this child has a disorder, will my future children have it also?

This question is a very individual one. It can be answered only by a trained professional who has more information about your family's health history. Many families seek genetic counseling to better understand why they happened to have a child with a birth defect. They may also wish to discuss possible risks to their future children. If you wish more information about genetic counseling, your doctor will help obtain it.

How can I make it easier for the doctor to help my baby?

If your doctor asks you to bring in the baby for retesting, do it as soon as you can. If your child does have a disorder, your prompt action in following the doctor's instructions can be very important.

If you do not have a telephone, it would be helpful to leave the telephone number of a friend, relative, or neighbor with the doctor. You can also help by notifying your doctor or clinic immediately if you move soon after the baby is born. Inform them of your new address and telephone number. Then, if your child needs to be retested for any reason, your doctor will know where you can be reached.

Remember, time is of great importance. As a parent, you can help assure the health of the new generation by your cooperation with the Newborn Screening Program.

Birth defects identified by the newborn screening program:

PHENYLKETONDRIA – also called PKU

A component of food protein, phenylalanine cannot be broken down by the body due to lack of a chemical (enzyme). Brain damage, which would normally result, can be prevented by a special diet low in phenylalanine. Occurs in about one of every 15,000 newborns.

BRANCH-CHAIN KETONURIA –

This is also called maple syrup urine disease (MSUD) because the urine has a maple syrup odor resulting from the baby's inability to break down some components of food protein.

Again, this is due to the lack of an enzyme. Treatment with a special diet can prevent life-threatening complications. Occurs in about one of every 210,000 newborns.

GALACTOMSEMIA –

This is a milk sugar, galactose cannot be broken down by the body due to the lack of an enzyme. A diet low in galactose can prevent life-threatening complications. Occurs in about one of every 75,000 newborns.

HOMOCYSTINURIA –

This is a lack of an enzyme in the liver. Can produce mental retardation and body changes. Treated with a special diet. Occurs in about one of every 225,000 newborns.

HYPOTHYROIDISM –

This is inadequate production of a hormone, thyroxine. Can lead to mental and growth retardation. Treated by giving thyroxine tablets. Occurs in about one of every 4,000 newborns.

SICKLE CELL ANEMIA –

This is a disorder resulting from inheritance of the trait for sickle cell hemoglobin from both parents. Children with sickle cell anemia often have a very low blood count and may have attacks of pain, jaundice, infections, and other problems throughout their lives.