

Please remember:

**At one of your baby's first check-ups,
ask your baby's health care provider
about the newborn screening results.**

You can find out the results from your baby's health care provider or the hospital where your baby was born. Generally parents are notified only if retesting or further testing is needed.



Iowa Department of Public Health
Center for Congenital and Inherited Disorders
321 East 12th Street
Des Moines, IA 50319-0075

To order additional copies, contact the
Healthy Families Line at 1-800-369-2229.



What is newborn metabolic screening?

Your baby's health is important



And that's why your hospital and the Iowa Department of Public Health work together to make sure all babies born in Iowa are as healthy as they can be.

A metabolic disorder is a condition caused by the accumulation or lack of certain chemicals or hormones made naturally in the body. Your baby will have a blood test shortly after birth that looks for these rare, but serious disorders.

These disorders can cause serious health problems, but early treatment can help your baby.

Why is my baby tested?

Newborn screening finds problems early.



All parents want a healthy baby. Testing helps assure that your baby will be as healthy as possible. Babies can look very healthy at birth and still have one of these disorders.

Babies born with metabolic disorders often show no outward symptoms. If a metabolic condition is not found early, poor physical and mental development, and even death, may occur.

To make sure these disorders are found quickly, Iowa law requires that all babies be tested. If found early, babies with most of these conditions can be treated and live healthy lives.

How is my baby tested?

The test is safe and simple.



A few drops of blood are taken from your baby's heel. The blood is then sent to the state University Hygienic Laboratory in Ankeny, Iowa.

The test should be done before your baby leaves the hospital. If your baby is tested before 24 hours of age, a second test should be done before your baby is two weeks old. If your baby was born at home, then testing should be done before five days of age.

Can I refuse to have my baby tested?

You may refuse testing on behalf of your child. If after consulting with your health care provider, you decide to refuse the test, you must sign an *Iowa Neonatal Metabolic Screening Program Waiver for Newborn Screening Refusal* form and **accept the legal responsibility for the consequences of this decision.**

Unless the person or his/her legal authorized representative specifically prohibits such use in writing, the blood specimen and information obtained during the testing process becomes property of the state, and may be used for program evaluation or research by the Iowa Department of Public Health or department-approved scientific researchers to improve the health of mothers and children. Such studies are published without identifying the person or persons from whom these results were obtained.

I was told I need to repeat my baby's test. What does this mean?

Retesting does NOT mean there is anything wrong with your baby. It simply means that another blood sample must be taken. Retesting may be required for a number of reasons. Some of these reasons include:

Unsatisfactory specimen: There is not enough blood to complete all of the required screening tests, or the sample cannot be tested for other reasons, such as too much blood on the sample.

Early collection: The blood sample was drawn before your baby was 24 hours old or before your baby has eaten. A repeat test must be done as soon as possible to avoid missing a disorder.

Medications: Certain medications can cause an abnormal result.

Abnormal test result: An abnormal test result means your baby “might” have a disorder. Your doctor will work with you to determine if further testing is needed.

What will my baby be tested for?

Disorders identified by the Iowa Neonatal Metabolic Screening Program:

Galactosemia

Babies with this disorder cannot convert galactose, a sugar present in milk, into glucose, a sugar that the body uses as an energy source. Galactosemia can cause death in infancy, or blindness and mental retardation. The treatment for this condition is to eliminate milk and all other dairy products from your baby's diet.

*This disorder occurs
in about 1 in every
70,000 births.*



Congenital Adrenal Hyperplasia

Babies born with this disorder have adrenal glands that are unable to make enough of the hormone cortisol and, in some cases, are unable to make enough of the hormone aldosterone. This disorder may affect genital development and, without treatment, severe cases may result in death.



Treatment consists of replacing the deficient hormones by medication, or eliminating the source of excess hormones. *This disorder occurs in about 1 in every 12,000 births.*

Congenital Hypothyroidism

Babies with this disorder are born with a thyroid gland that does not make enough thyroid hormone. This can lead to poor growth and abnormal brain development. If it is detected in time, a baby can be treated with thyroid hormone pills and thus avoid the problems caused by low thyroid hormone. *This disorder occurs in about 1 in every 4,000 births.*

Biotinidase Deficiency

Babies born with biotinidase deficiency cannot reuse the vitamin biotin. Biotin helps maintain normal body functioning. Without treatment, this disorder can lead to seizures, developmental delay, eczema and hearing loss. Problems can be prevented with biotin treatment. *This disorder occurs in about 1 in every 60,000 births.*

Cystic Fibrosis (CF)

Cystic fibrosis is the most common inherited (genetic) disorder, affecting about 30,000 children and adults in the U.S. A defective gene causes lung infections and digestive problems with malnutrition. CF can be life-shortening. *Cystic fibrosis occurs in about 1 in every 3,200 Caucasian births (1 in every 3,900 births in the U.S.).*

Hemoglobinopathies

Hemoglobinopathies are inherited red blood cell disorders. Hemoglobin is the protein in blood that carries oxygen from the lungs to the body. The most common hemoglobin disorder is sickle cell disease. When sickle-shaped cells block small blood vessels, less blood can reach that part of the body. *Sickle cell anemia occurs in about 1 in every 375 African Americans.*

Expanded Screening Panel

Expanded screening is a group of up to 30 additional disorders. These disorders fall into three main types:

Amino Acid Disorders

Babies born with one of these disorders cannot process certain amino acids. These amino acids, along with other toxic substances build up in the body and cause serious effects on health, growth and learning. Treatment may include a special diet, close monitoring and/or vitamin and amino acid supplements. An example of an amino acid disorder is Phenylketonuria (PKU). Babies with PKU cannot process a substance called phenylalanine that is found in almost all food protein. Without treatment, phenylalanine builds up in the bloodstream and causes brain damage and mental retardation. If PKU is detected early, mental retardation can be prevented by feeding your baby a special diet. *This disorder occurs in about 1 in every 12,000 births.*

Fatty Acid Oxidation Disorders

Babies with fatty acid disorders are unable to breakdown stored fats for energy. One example of a fatty acid disorder is Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD). Babies born with MCAD cannot break down fat into energy because an enzyme is missing or does not work correctly. People with MCAD should not fast (go without food) for very long or they can experience low blood sugar, seizures, coma and even death.

This disorder occurs in about 1 in every 12,000 births.

Organic Acid Disorders

Babies born with organic acid disorders have a chemical imbalance in their body which can be toxic. Organic acids play an important role in the breakdown of fats, sugars and protein for the body's use and storage. Muscle wasting, seizures, developmental delays and even death can occur if untreated. Treatment may include a special diet, monitoring and medications.



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