Newborn Screening Program -- Laboratory Tests that Make Sense for Your Baby

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Newborn Screening Program
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Colorado's Newborn Screening Program
Colorado screens infants for diseases and disorders that can harm newborn babies. Although a baby may look normal, many conditions and disorders cannot be seen. Through the Newborn Screening Program, hundreds of Colorado children have received early diagnosis and treatment for conditions that, if untreated, would lead to devastating illness, painful disability, severe mental retardation or death.

If test results indicate your baby has abnormalities, do not panic. First and second screenings may indicate there is a problem, but also may identify chemicals in the blood that occur as a result of childbirth. Contact your baby's doctor for more information. These test results must be verified by more tests before a diagnosis is made.

WHO . . . is tested?
All newborn babies.

HOW . . .is testing done?
At the hospital, a few drops of blood are drawn from the baby's heel, put on special paper, and sent to the laboratory.

WHEN . . . is testing done?
Babies are tested within 72 hours after birth. If the baby was born at home and the midwife or doctor did not do a test, take the baby to a hospital or your doctor's office as soon as you can. All babies are tested again at their first well-baby visit (usually by two weeks of age).

WHY . . . should my baby be tested?
Babies with these conditions appear normal at birth. If untreated, these conditions will affect the baby's brain or physical development or cause other medical problems. These conditions can begin to affect the baby in the first days or weeks of life. By testing all newborns, babies with these conditions can be found early. Early diagnosis and treatment can result in normal growth and

development, or prevent many of the medical problems associated with these conditions.

WHY . . . should my baby be tested a second time?
Most babies get their first newborn screening test before they are three days old. Sometimes these early test results are not accurate because the baby is too young. Some conditions may not be detected on the first screen. Therefore, all babies must be tested a second time, usually at the first well-baby visit (by two weeks of age).

HOW . . . will I be given the test results?
The Newborn Screening Laboratory will send a report of the results to your hospital or baby's doctor. You may ask for the results when you bring your baby in for a checkup. Your doctor will notify you if additional testing is required. Additional testing does not necessarily mean that your child has one of these conditions, but it does mean that more testing is needed to see if your baby has a problem. When this testing is completed, your doctor is notified promptly.

If you have questions or concerns about newborn screening, contact your baby's doctor. Colorado Newborn Screening Laboratory staff cannot give out test results to anyone but the baby's doctor.

WHAT . . . tests are done at the Colorado Newborn Screening Laboratory?
Colorado previously tested for seven disorders, and has added testing equipment to test for 23 more disorders. All these are listed below.

Hemoglobinopathies:
These are inherited diseases of the red blood cells, which carry oxygen from the lungs to every part of the body. Hemoglobin is the main protein inside red blood cells. In the diseases called sickling disorders, the protein is abnormal. It can cause some red blood cells to become stiff and be shaped differently. The very stiff blood cells can get stuck in tiny blood vessels, which causes pain.

Sometimes the stiff blood cells cause damage to organs. Young children with sickle cell anemia need to be protected from bacterial infections such as pneumonia and meningitis. The severest type of red blood disorders can cause stroke and sometimes death.

Treatment may include pain medications and blood transfusions. Parents should be sure their babies get all childhood shots.

Fatty Acid Oxidation Disorders:
The body uses sugar (glucose) to fuel activities. There are special chemicals, called enzymes, that convert fat into energy for the body to use when it runs out of sugar. However, in these disorders, the body blocks this process. Without

treatment, organs and the brain may be damaged. Sometimes the patient goes into a coma, or dies.

Early diagnosis and treatment is key for the patient to have a normal life. Treatment generally involves a special diet, nutritional supplements, and avoiding fasting. Colorado tests for the following FAO disorders:

- Medium Chain Acyl-CoA dehydrogenase deficiency
- Very Long Chain Acyl-CoA dehydrogenase deficiency
- Long-chain Acyl CoA dehydrogenase deficiency
- Trifunctional protein deficiency
- Carnitine Acyl-carnitine translocase deficiency
- Short Chain Acyl-CoA dehydrogenase deficiency
- Carnitine palmitoyltransferase II deficiency
- Glutaric acidemia Type 2

Amino Acid Disorders:
People with these disorders cannot digest protein into amino acids, or have problems getting rid of the nitrogen that makes up amino acid molecules. Harmful levels of amino acids or ammonia can build up in the body. Damage to the body may include liver or kidney damage, nerve damage, mental retardation, coma, eye damage, or death.

The main treatment for these diseases involves special diets. Patients may need vitamins or nutritional supplements, and medications to help with ammonia build-up. Patients should not fast. The following amino acid disorders are included in Colorado's newborn screening tests:

- Phenylketonuria
- Arginosuccinic academia
- Citrullinemia
- Tyrosinemia
- Hypermethionemia
- Maple Syrup urine disease
- Homocystinuria

Organic Acid Disorders:
In these disorders, an enzyme that breaks down chemicals in the body does not work. If these chemicals are not broken down, harmful acids build up in the body. It is important to catch these disorders very early. If the disorder is not diagnosed early, damage to the body can cause coma and death during the first month of life.
Treatment usually involves a special diet, dietary supplements, vitamins, and not fasting. Colorado tests newborns for the following organic acid disorders:

- Isovaleric academia
- Glutaric academia Type 1
- 3-hydroxy-3 methyl glutaric aciduria
- Multiple Carboxylase deficiency

- 3-methylcrotonyl-CoA carboxylase deficiency
- 3-methylglutaconic aciduria
- Methylmalonic acidemias
- Propionic academia
- beta-Ketothiolase deficiency

Others:
These diseases and disorders do not fit into a category. Symptoms range from mild to life-threatening. Early detection and treatment allows most of these patients to live a relatively normal life. Some patients may need life-long treatment.

Congenital Hypothyroidism:
If not diagnosed early, this thyroid hormone deficiency can retard growth and brain development. Treatment is very easy. The patient takes a thyroid supplement daily.

Galactosemia:
With this disorder, the body does not make a liver enzyme that converts milk sugar (galactose) into the simple sugar (glucose) that the body needs. The milk sugar accumulates in vital organs, causing damage to the organs, blindness, mental retardation, infection and death. Even if caught early and treated, there may be some developmental delays.

Treatment involves removing milk and dairy products from the baby's diet.

Cystic Fibrosis:
In this inherited disorder, an abnormal protein causes lung and digestive problems. Early diagnosis and treatment improves the growth of babies and children with Cystic Fibrosis. The average age of death is between 30 and 35 years.
Treatment includes diet supplements, vitamins, medications and respiratory therapy.

Biotinidase Deficiency:
With this disorder, the body does not have the biotinidase enzyme needed to recycle the vitamin biotin. This disorder may cause frequent infections, uncoordinated movement, hearing loss, seizures, mental retardation, coma and death.

The treatment consists of taking daily oral doses of biotin. This treatment prevents all the problems of the disorder.

Congenital Adrenal Hyperplasia:
In this disorder, the adrenal gland does not correctly make some

hormones needed by the body. Sometimes masculine genitals will appear in female infants. Severe forms of the disease can cause loss of salt from the body, which can be life threatening if not diagnosed.

Treatment for this disorder includes taking salt and hormone replacement.

Glossary

***Adrenal glands**:* A small pair of glands located above the kidneys that produce hormones that influence or regulate salt and water balance in the body, the body's response to stress, metabolism, the immune system, and sexual development and function. -(Pituitary Network Association - Glossary; Kids Health at the Nemours Foundation)

***Amino acids**:* Biological structures that combine to form proteins. When proteins are digested, amino acids are left over. The human body requires a number of amino acids to breakdown food.

Biotin: A water-soluble vitamin that is needed to form fatty acids and glucose, which are used as fuels by the body. Biotin also is important for processing amino acids and carbohydrates.

***Enzymes**:* Complex proteins that are produced by living cells and help with specific biochemical reactions at body temperatures.

Fasting: Not eating.

***First screening**:* Ideally, the baby should have an initial test in a period from 48 to 72 hours of age.

Gland: A group of cells that makes and gives off, or secretes, biological substances. A gland selects and removes materials from the blood, processes them, and secretes the finished biological product for use somewhere in the body. (Kids Health at the Nemours Foundation)

Second screening: The second specimen should be taken no less than 72 hours and no greater than 30 days of age. Optimally, the sample will be taken from eight to fifteen days after birth, usually at the well-baby visit.

Well-baby visit: About two weeks after leaving the hospital, the baby is taken to the doctor's office for a complete check-up, including the second screening.