Newborn Screening

Newborn Screening is a series of blood tests run on a sample of your baby's blood obtained from his heel. It is usually done within 24 to 48 hours after a baby is born. The American College of Medical Genetics (ACMG) recommends that 29 disorders be screened for at birth. Colorado's newborn screening program runs the infant's blood sample through a series of tests and then notifies your midwife of the results. More than four million infants are born and undergo newborn screening in the U.S. each year. While metabolic disorders are rare, nearly every child in the United States is tested. A recent study suggests that for every true positive test result from newborn screening, there are 12 false positive ones.

Newborn Screening in Colorado: Colorado is one of a handful of states that requires 2 newborn screening tests. The first test is usually obtained at the hospital within 24 hours of birth. A second test is recommended sometime between 10 and 14 days old. The rational for two tests is 2-fold. The majority of the disorders screened for are genetic and can be caught in the first screen. It is easy and convenient for the test to be administered at the hospital before the baby goes home. There are a few disorders however, that cannot be caught until the baby has started digesting protein which doesn't occur until mom's milk comes in, typically on the 3rd day after mom and baby have gone home. Therefore, a 2nd test is called for to screen for the disorders that may have been missed. The cost of the screening is $92.00, which covers the first and second screen. The fee is subject to change and is typically covered by insurance and Medicaid.

Storage and Use of Dried Blood Spots: After the newborn screening is completed, there will remain some dried blood on the screening test cards. These are called "residual dried blood spots." The residual dried blood spots are stored at the lab for 6 months for the purpose of retesting the newborn if necessary. Genetic information linked to the individual is considered the property of the individual and it will never be released without the patient or the patient's guardian's consent. During storage, information from the newborn screening process may be given to a research facility as long as the researcher does not get any information that identifies who the information belongs to. This means that a researcher will only have access to anonymous samples. Release of any identifiable information will require written consent from the baby's parent or guardian Your newborn's identifying information will never be used for anything other than therapeutic purposes and will never be used to deny group disability or long-term care insurance or any other nontherapeutic purpose. However, the genetic information obtained during newborn screening may be released to be used for a criminal investigation or prosecution. Following the six-month storage period, the blood spots are separated from the infant's identifying information and the spots become unusable through a process called autoclaving and the information section is shredded.

During the six months that the infant's blood spot card is stored, parents may request their infant's newborn screening card to be released to them by submitting a letter to the laboratory requesting it. Parents may then pick up their infant's newborn screening card from the lab by showing valid identification at the time of the release. If parents want to store their child's residual dried blood spot for a longer period of time for any reason, they may request to have it returned and then pay for it to be stored at a private laboratory.

Complete Listing of Disorders Tested for in the Colorado Newborn Screening Program

Amino Acid Disorders

- Arginase deficiency
- Argininosuccinic acidemia
- Citrullinemia
- Homocystinuria
- Hypermethioninemia
- Maple syrup urine disease
- Phenylketonuria (PKU)
- Tyrosinemias

Endocrine Disorders

- Congenital adrenal hyperplasia
- Congenital hypothyroidism

Fatty Acid Oxidation Disorders

- Carnitine acylcarnitine translocase deficiency
- Carnitine palmitoyltransferase II deficiency
- Carnitine palmitoyltransferase deficiency 1a
- Carnitine uptake defect
- Long-chain L-3-hydroxyacyl-CoA dehydrogenase deficiency
- Medium-chain acyl-CoA dehydrogenase deficiency
- Short-chain acyl-CoA dehydrogenase deficiency
- Trifunctional protein deficiency
- Very long-chain acyl-CoA dehydrogenase deficiency

Hemoglobinopathies

- Beta-thalassemia
- Sickle cell anemia
- Hemoglobin SC disease

Organic Acid Disorders

- 3-Hydroxy-3-Methylglutaryl-CoA Lyase deficiency
- 3-Methylcrotonyl-CoA carboxylase deficiency
- 3-Methylglutaconic aciduria (3-MGA)
- Beta-ketothiolase deficiency
- Biotinidase deficiency
- Glutaric acidemia type I
- Glutaric acidemia type II
- Isovaleric acidemia
- Malonic acidemia
- Methylmalonic acidemias
- Multiple carboxylase deficiency
- Propionic acidemia

Others

- Cystic fibrosis (CF)
- Galactosemia

- Hearing (not a metabolic/bloodspot screen)

Benefits: The newborn screen is valuable in that it can catch metabolic diseases that otherwise won't show up for months or even years, and may have already begun to do permanent damage by the time symptoms appear.

Risks: While there are very few risks of taking a small blood sample from a healthy infant, the baby does feel momentary pain when his heel is poked to make it bleed. Most states require only 1 test within 72 hours of birth (so there is enough time for mom's milk to come in). None of the metabolic diseases that are screened for cause damage if not caught within a few days after birth. One test at 10 to 14 days of age is just as sufficient as 2 tests.

Sources:

-http://www.cdphe.state.co.us/ps/hcp/nbms/metabolicscreening.html

-www.**babysfirsttest**.org