

**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)
  - Severe Combined Immunodeficiency Disease (SCID)
  - Galactosemia
  - Hearing (not a blood test)



**As parents of a newborn baby, what can we do?**

As parents of a newborn baby, there are many things you can do to make sure your baby gets this important testing!



**Be sure your baby gets a newborn screening test by the time your baby is two days (48 hours) old.** Some of the diseases screened for could cause severe illness by as early as one week of age if they are not found and treated right away.

**Check with your midwife for the results of your baby's first newborn screen.**

**When your baby is 1 to 2 weeks old, your baby should get a second newborn screening test, even if the results of the first test are normal.** Some of the conditions screened for could be missed on the first screen because it is collected so early, and if those conditions are not found and treated early in life, they can negatively affect a child's mental or physical health for a lifetime.

There is not an additional charge for the second newborn screening test; the cost of both screens is included in the newborn screening fee.

**Check with your midwife for the results of your baby's second newborn screening test.**

If your baby needs further testing—get that testing *right away!* Screening results that are not normal can be a sign that your baby is very sick.

**Don't rely on others to make sure your baby gets tested!**



**I have questions about newborn screening. Where can I get more information?**

Your midwife and your baby's doctor can answer questions you have about newborn screening. *Ask!*

Visit these websites:

[www.babysfirsttest.org](http://www.babysfirsttest.org)

[www.marchofdimes.com](http://www.marchofdimes.com) (English and Spanish)

[www.newbornscreening.info](http://www.newbornscreening.info) (English and Spanish)

Colorado Department of Public Health and Environment, Laboratory Services Division  
 Email: [cdphe.lab@state.co.us](mailto:cdphe.lab@state.co.us)  
 Phone: 303-692-3670



**Colorado's Newborn Screening Program**



**First Steps to a Healthy Start for Your Baby**

**For Midwife-Attended Home Births**



**COLORADO**  
 Department of Public Health & Environment





The Colorado Department of Public Health and Environment wants to help your baby get a healthy start. Shortly after your baby is born, your baby's blood will be tested to detect rare but serious health disorders. If these disorders are not detected and treated early in your baby's life, they can negatively affect a child's mental or physical health for a lifetime.



### What is a newborn screening test?

A newborn screening test is a simple and safe blood test which looks for disorders that are **rare**, but **very serious**.

### Why does my baby need the test?



Newborn babies who have these disorders look healthy, but babies with some of these disorders will get very sick or die just a few days or weeks after they are

born. The newborn screening test helps find babies with these disorders early, so they can get medical treatment before they get sick. With early treatment, most babies live a healthy life.

### Who is tested?

State law requires that all babies born in Colorado have a newborn screening test.

Parents have the right to refuse screening. But remember, newborn screening is safe and simple and is one of the *most important* things you can do to protect your child's health.

### Who pays for testing?

Private insurance, Medicaid or direct payment to your midwife are the options to pay for the test.

### How is the test done?

A few drops of blood are taken from your baby's heel and put on a special paper. The state public health laboratory does the testing.

### When will my baby be tested?

Your baby will be tested **two** times.

Your newborn baby **must** get a newborn screening test by the time your baby is two days (48 hours) old.



All babies get a **second** newborn screening test when they are one to two weeks old.

### Why does my baby need to be tested twice?



Some disorders might be missed because the first newborn screening test is done so soon after birth.

The second newborn screening test is for your baby's safety. It can find a disorder that was missed on the first newborn screen.



### What will my baby be tested for?

A complete list of disorders that your baby will be tested for is included in this brochure.

### How do I get my baby's test results?

The state public health laboratory will give the results to your midwife or your baby's doctor who will tell you the results of your baby's newborn screening test. If the results are not normal, your midwife will be notified by phone *immediately*.

### What does it mean if my baby's test is not normal?

If your baby's newborn screening test result is **not normal**, it means **your baby needs more testing**. Many **healthy** babies have newborn screening results that are **not normal**. Your midwife or your baby's doctor will tell you if your baby needs more testing. Your baby might need to see a specialist. Get any additional testing your baby needs *right away!*



### If my baby has one of these disorders, is there a cure?

There are no cures for these disorders, but if these disorders are found and these babies get treatment early, the serious problems caused by these disorders can be prevented or reduced.

If babies with these disorders get early and continuous treatment, most can develop normally and live healthy lives.



### What is a "carrier"?

Once in a while newborn screening will find babies who are "carriers" for a disease but do not have the disease themselves. (Newborn screening does not find all carriers.) If a baby is a carrier, it is advisable for the baby's parents to have what is called "carrier testing" before a future pregnancy. If you have questions about what it means for a baby to be a carrier, talk to your healthcare provider.

**Amino Acid Disorders** Babies with these disorders cannot process certain amino acids (building blocks of protein) because of a missing enzyme. These amino acids, along with harmful substances, build up in the body and can cause health problems.

Treatment often includes a special diet and sometimes medication.

#### Endocrine Disorders

##### Congenital Hypothyroidism

Babies with this disorder cannot make enough thyroid hormone. Thyroid hormone is necessary for normal brain development.

##### Congenital Adrenal Hyperplasia

The adrenal glands do not produce normal amounts of hormones. Without early treatment, babies can have a life-threatening illness and can become disabled and some may die.

**Organic Acid Disorders** Babies with organic acid disorders are missing an enzyme in their body and cannot break down protein properly. This causes harmful substances to build up in the body and can cause serious health problems even within the first few days of life.

Some babies require a special diet, while others just need medication.

**Cystic Fibrosis** Cystic fibrosis is a condition that causes mucus to build up in the lungs and other organs leading to chronic poor growth and lung infections.

Early treatment can improve growth and minimize or delay complications.

**Fatty Acid Disorders** Babies with fatty acid oxidation disorders cannot break down fats for energy. Fats are an important energy source for the brain and muscles when glucose levels are low.

Some babies will need a special diet, whereas other babies will just need to avoid fasting.

**Galactosemia** Babies with this disorder cannot break down a sugar found in milk.

Babies need soy formula to avoid health problems.

**Hemoglobinopathies** These are inherited diseases that cause the red blood cells to become stiff and distorted in shape.

If not properly managed, the child experiences pain, increased infections and has an increased risk of stroke or death.

#### Severe Combined Immunodeficiency (SCID)

Babies with this disorder cannot fight off viruses and infections and usually die within the first year or two of life.

Treatment is a bone marrow transplant.