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

### Amino Acid Disorders
- Arginase deficiency
- Argininosuccinic acidemia
- Citrullinemia
- Hypermethioninemia
- Maple syrup urine disease
- Phenylketonuria (PKU)
- Tyrosinemia

### Endocrine Disorders
- Congenital adrenal hyperplasia
- Congenital hypothyroidism

### Fatty Acid Oxidation Disorders
- Carnitine acylcarnitine translocase deficiency
- Carnitine palmitoyltransferase II deficiency
- 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 acidemia
- Multiple carboxylase deficiency
- Propionic acidemia

### Others
- Cystic Fibrosis (CF)
- Severe Combined Immunodeficiency Disease (SCID)
- Galactosemia
- Hearing (not a blood test)

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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!

Before your baby goes home from the hospital, check with hospital staff to make sure your baby got a newborn screening test.

When your baby is one to two weeks old, your baby should see a doctor for a “well-child check-up”.

Ask the doctor for the results of your baby’s first newborn screening test. In most cases, the results of your baby’s newborn screening test will be normal.

If your doctor tells you that your baby’s newborn screening test was not normal, your baby will need more testing. Follow your doctor’s instructions carefully and be sure to get the tests your baby needs right away!

All babies must get a second newborn screening test when they are one to two weeks old, even if the first test was normal. Be sure the doctor tells you how to get that second test, and do it right away.

A baby’s first newborn screening test is usually sent to the lab with the mother’s last name. If your baby might be known by more than one last name, include both names on the second test, so the lab will know that both tests are for the same baby.

Check back with the doctor for the results of your baby’s second newborn screening test.

Don’t rely on others to make sure your baby gets tested!
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.

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.