



Genetic Disorders

Hemoglobinopathies & hemoglobinopathy traits

Hemoglobinopathies are blood disorders. They can cause misshaped red blood cells, anemia, severe pain and high risk for infections. Sometimes medicines are used to treat these disorders. If your child has a **hemoglobinopathy trait**, they are not sick but can later cause their own babies to have a hemoglobinopathy.

Sickle Cell Anemia

This disorder is a hemoglobinopathy. It causes sickle-shaped red blood cells, anemia, and other health problems. Medicine is used to treat this anemia.

Sickle Cell Trait

A baby with Sickle Cell Trait has one gene that makes sickle-shaped red blood cells and one gene that makes normal red blood cells. These babies are not sick. Treatment is not needed.

Cystic fibrosis

This disorder causes severe lung and digestive problems due to thick and sticky body fluids. Babies can have serious infections in their lungs and cannot digest their food well. To treat this disorder, babies take medicine to help fight infections and enzymes to help digest food.

Severe Combined Immunodeficiency

Severe Combined Immunodeficiency (SCID) and related disorders involve the immune system. Babies with SCID are not able to fight infection due to malfunction of their immune system. Although babies with SCID can appear healthy at birth, they can become sick very quickly from common illnesses like a cold or stomach virus. Some babies with SCID need a bone marrow transplant while less severe forms of these disorders can be treated with special medicines.

Note: There are other genetic disorders that cannot be found through newborn screening.

What happens to my baby's blood sample after the lab tests it?

DHEC will destroy your baby's blood sample once it is no longer needed for testing. It will not be used in research or for any purpose other than newborn screening. If you have questions about how your baby's blood sample is handled, call the Division of Children's Health, Newborn Screening Follow-up Program, at (803) 898-0767 or the Bureau of Laboratories at (803) 896-0800.

What else can I do to take care of my baby?

Make sure your baby gets well baby checkups. Along with newborn screening, checkups help you make sure that your baby is healthy and that problems are found before they become serious.

For more information, contact

- Your county public health department
- DHEC Newborn Screening Follow-up Program
Box 101106
Columbia, SC, 29211
Phone (803) 898-0767

Glossary

Adrenal gland

Organ in the body that makes hormones that help the body maintain a proper energy supply and blood sugar level, maintain normal levels of "salt minerals" like sodium and potassium, and maintain proper growth

Amino acid

Small compounds that are the building blocks of proteins

Anemia

A blood condition that can cause weakness and many other problems

Disorder

In this brochure, a health problem your baby is born with

Enzymes

Proteins in the body that make systems work properly

Genes

Regulate the way certain cells in the body are made

Genetic

Related to genes

Hormones

The chemical messengers of the body

Metabolism

The chemical and physical processes going on in the body's cells, including the changing of food into living tissue and the changing of living tissue into energy and waste products

Thyroid gland

Organ in the body that makes hormones that help the body grow and mature properly



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Newborn Screening

For Your Baby's Health



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Parents sometimes worry about the health of their new baby. Usually a baby who looks healthy is healthy. But sometimes, this may not be true. A baby may have problems that can't be seen. If untreated, these problems could lead to mental retardation, abnormal growth, dangerous infections or even death.

Finding the causes of these problems is the goal of the South Carolina Department of Health and Environmental Control's (DHEC) Newborn Screening Program.

This brochure will explain what newborn screening is, what health problems (disorders) your baby is tested for, and what to do if any of these problems are found. A glossary at the end of this brochure can help you understand some of the words used in this brochure. We also encourage you to ask questions—your baby's doctor, nurse, or DHEC can help you.

What is "newborn screening"?

Newborn screening is a blood test that checks for hidden health disorders in newborn babies.

All babies are tested soon after birth for several genetic and chemical problems. Tests are done on a small sample of baby's blood. This blood is taken by pricking baby's heel. The blood is sent to a DHEC laboratory for testing. If a problem is discovered, early treatment can give baby the best chance for a healthy life.

South Carolina law requires newborn testing.

What if my new baby seems very healthy? Are these tests really needed?

Yes. Most babies who have one of these problems seem healthy at birth. Most are born into families who have no history of these problems. Blood tests are the only way these disorders can be found early, before serious illness or death occurs.

The chance of having one of these disorders is small. Why is there a state law about screening?

These disorders are not common, but they ARE very serious. Testing every baby at birth is the fastest way to find babies who have these problems. Then they can be treated right away.

How accurate is newborn screening?

The results of newborn screening tests are correct almost all the time, but no test is perfect. In very rare cases, a baby with a "normal" newborn screening result may be diagnosed with one of the newborn screening disorders later in life.

How will I get my baby's test results?

Your doctor will tell you the results at baby's first check up. That is why it is very important that you choose a doctor for your baby before he or she is born. Give the hospital the name of the doctor who will be taking care of your baby so that they can make sure your baby's doctor is listed on the newborn screening form. All test results will be mailed to that doctor. Results are also mailed to the place where your baby was born. The results are mailed within 14 days after the blood sample is received at the laboratory.

If any positive results are found, the doctor is notified right away. The doctor will then contact you. A second test may be needed.

If testing shows that your baby has sickle cell trait or other similar blood trait, a specially trained counselor from a regional sickle cell community organization will also contact you.

What does it mean if I'm told my baby needs a second test?

There are four reasons why your baby might need a second test:

1. The first blood sample could not be used
2. The first blood sample was taken before baby was 24 hours old
3. The first blood sample test gave false positive results
4. The first blood sample showed that there is a small chance that your baby has a disorder

If you are asked to have your baby retested, please do so quickly! Taking your baby for a second test can be scary, but it's important. If your baby's second test is positive, your baby may have a disorder. In rare cases, doctors may begin treatment before they have the results of the second test. Early treatment can give your baby the best chance for a healthy life.

Can my baby be cured if he or she has one of these disorders?

No. However, all of these disorders can be treated. With treatment, serious effects can be lessened—and often prevented—if started early.



What disorders are tested for in South Carolina?

The test panel in South Carolina looks for:

- Metabolism disorders (amino acid, carbohydrate, organic acid, fatty acid)
- Hormone disorders
- Enzyme disorders
- Genetic disorders

These disorders are explained below.

Metabolism disorders

Phenylketonuria (PKU)

This amino acid disorder keeps baby's body from being able to use certain amino acids found in breast milk and formula. An amino acid called phenylalanine builds up in the baby's system and can hurt growing brain cells, causing mental retardation. To treat this disorder, doctors give babies a special formula and a diet low in phenylalanine.

Less common amino acid disorders

- Homocystinuria
- Maple syrup urine disease
- Citrullinemia
- Argininosuccinic aciduria
- Tyrosinemia

These disorders can cause seizures and severe brain damage. To treat these disorders, doctors put babies on a carefully planned diet.

Galactosemia

This carbohydrate disorder means baby's body cannot use a sugar, called galactose, found in cow milk-based formula and breast milk. Babies who are not treated can get life threatening infections and become mentally retarded. To treat this disorder, baby must be fed soy-based formula.

Organic acid disorders

Babies with these disorders cannot remove certain waste products from their blood. They may even go into a coma if they are not treated. Babies with these disorders may be treated with a special diet and medicine. Some of these disorders are:

- Propionic acidemia,
- Methylmalonic acidemia
- Malonic acidemia
- Isobutyryl coA dehydrogenase deficiency
- Isovaleric acidemia
- 2-methylbutyryl coA dehydrogenase deficiency
- 3-methylcrotonyl coA carboxylase deficiency
- Beta ketothiolase deficiency
- 3-methyl-3-OH-glutaryl coA lyase deficiency
- 3-methylglutaconyl coA hydratase deficiency
- Multiple carboxylase deficiency
- Glutaric aciduria I

Medium chain acyl co-A dehydrogenase deficiency (MCADD)

This fatty acid disorder causes the baby's body to be unable to use certain kinds of fat to make energy. Babies with MCADD may get very sick if they are ill and cannot eat like usual. They can have trouble breathing and have seizures. Their hearts may even stop beating. To treat this disorder, baby must eat every few hours and receive fast medical care when sick.



Less common fatty acid disorders

- Medium chain ketoacyl coA thiolase deficiency
- Short chain acyl coA dehydrogenase deficiency
- Medium and short chain 3-OH acyl coA dehydrogenase deficiency
- Dioneoyl coA reductase deficiency
- Long chain 3-OH acyl coA dehydrogenase deficiency
- Trifunctional protein deficiency
- Very long chain acyl coA dehydrogenase deficiency
- Glutaric aciduria II
- Carnitine uptake/transport deficiency
- Carnitine palmitoyltransferase I deficiency
- Carnitine palmitoyltransferase II deficiency
- Carnitine/acylcarnitine translocase deficiency

To treat these disorders, baby may receive a special diet and medicine.

Note: There are other metabolism disorders that cannot be found through newborn screening.

Hormone and Enzyme Disorders

Congenital hypothyroidism

In this disorder, the thyroid gland does not work properly. Baby does not grow or function normally and may develop severe mental retardation. To treat this disorder, babies take a special medicine.

Congenital adrenal hyperplasia (CAH)

In this disorder, the body's adrenal glands do not work normally. Babies with CAH will not grow or mature the right way. Some of these babies may even die. A baby with CAH can be treated with medicine.

Biotinidase deficiency

In this disorder, baby's body cannot use biotin, a vitamin in food. Without biotin, babies cannot grow and develop the right way. To treat this disorder, baby must take a special form of biotin in a capsule or tablet.

Note: There are other hormone and enzyme disorders that cannot be found through newborn screening.