infection due to malfunction of their immune system. Although they 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 those with less severe forms can be treated with special medicines.

Other rare genetic disorders identified are Spinal Muscular Atrophy (SMA) type 1 and Krabbe disease, also known as globoid cell leukodystrophy.

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

Metabolic disorders

Phenylketonuria (PKU)

This disorder makes baby's body unable 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 intellectual disability. Treatment includes a special formula and a diet low in phenylalanine.

Less common amino acid disorders are:

- Homocystinuria (HCY)
- Maple syrup urine disease (MSUD)
- Citrullinemia (CIT)
- Argininosuccinic aciduria (ASA)
- Tyrosinemia (TYR I)

These disorders can cause seizures and severe brain damage. Treatment includes a carefully planned diet.

Galactosemia

This carbohydrate disorder means baby's body cannot use galactose, a sugar found in cow milk-based formula and breast milk. Babies who are not treated can get life threatening infections and experience severe intellectual disability. Treatment includes feeding baby a soy-based formula.

Less common carbohydrate (lysosomal storage) disorders include: Pompe and MPSI

Organic acid disorders

Babies with these disorders cannot remove certain waste products from their blood. They may even go into a coma if left untreated.

Treatment may include a special diet and medicine.

Some of these time critical disorders are:

- Propionic acidemia (PA)
- Methylmalonic acidemia (MMA)
- Isovaleric academia (IVA)
- 3-Hydroxy-3- methyl-glutaric aciduria (HMG)
- Beta ketothiolase deficiency (BKT)
- Holocarboxylase synthase deficiency (MCD)
- Glutaric aciduria I (GA I)

Fatty acid disorders

Medium chain acyl co-A dehydrogenase deficiency (MCAD)

This fatty acid disorder makes baby's body unable to use certain kinds of fat to make energy.

Babies with MCAD may get very sick if they are ill and cannot eat well. They can have trouble breathing and have seizures. Their hearts may even stop beating.

Treatment includes feeding every few hours and receiving fast medical care when sick.

Less common fatty acid disorders:

- Very Long chain acyl CoA dehydrogenase deficiency (VLCAD)
- Long chain L-3- hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP)

Treatment may include a special diet and medicine.

Note: There are other metabolic and fatty acid 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 for any purpose other than newborn screening. If you have questions about how your baby's blood sample is handled, call the SC Public Health Laboratory at (803) 896-0800 or the Newborn Screening Program at (803) 898-0767

What else can I do to care for my baby?

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

For more information contact:

SC DHEC
Newborn Screening Program
(803) 898-0767;

your local health department;

or

Scan the QR code below

DHEC ML-000032 04/23

Newborn Screening

For Your Baby's Health

dhec

Newborn Screening

For Your Baby's Health

Many parents worry about the health of their new baby. Usually, a baby who looks healthy is healthy. But sometimes, this may not be true. Babies can have problems that can't be seen, and, if left untreated, these problems could lead to intellectual disability, abnormal growth, dangerous infections, or even death.

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

Use this resource to find out what newborn screening is, what health disorders your baby is tested for and what to do if any problems are found. We also encourage you to ask questions; your baby's doctor, nurse, or DHEC can help.

What is "newborn screening?"

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

Soon after birth, all babies have a small sample of their blood tested for several rare and common disorders. This blood is taken from baby's heel and sent to DHEC's Public Health Lab 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 healthy? Are these tests really needed?

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

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

Though rare, these disorders are serious. Testing every baby at birth is the fastest way to find which ones have these disorders so they can be treated right away.

How accurate is newborn screening?

Newborn screening lab tests are highly accurate, but no test is perfect. In very rare cases, a baby with a "normal" newborn screening result may be diagnosed with a disorder later in life.

How will I get my baby's test results?

Your baby's doctor will share the results at their first checkup. This is why it is so important that you choose a doctor for your baby **before** he or she is born and that the hospital has the name of that doctor. That way, they can ensure the doctor is listed on the newborn screening form.

If abnormal results are found, the doctor will be notified right away and contact you. A follow-up test or appointment may be needed.

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

Your baby may need a second test if the first blood sample:

- Could not be used for testing
- Was taken before baby was 24 hours old
- Gave false positive test results
- Showed there is a small chance that your baby has a disorder

If you are asked to have your baby retested, please **do so quickly**. A second test can be scary, but it's important.

If the second test is positive, your baby may have a disorder. In rare cases, doctors may begin treatment after the first test. Early treatment can give your baby the best chance for a healthy life.

Can my baby be cured if they have one of these disorders?

No. But, all of these disorders can be treated. With early treatment, serious effects may be lessened and often prevented.

What disorders are tested for in South Carolina?

The test panel looks for:

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

These disorders are explained below.

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 intellectual disability. Treatment includes 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 properly. Some babies may even die. CAH can be treated with medicine.

Biotinidase deficiency

In this disorder, baby's body cannot use biotin, a B vitamin found in food. Without biotin, babies cannot grow and develop the right way. Treatment includes taking 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.

Genetic Disorders

Hemoglobin Diseases and Hemoglobin Traits

Hemoglobin diseases are blood disorders. They can cause anemia and many other health problems.

Sickle Cell Disease (SCD)

SCD is a hemoglobin disorder. It causes sickle-shaped red blood cells, anemia, severe pain, and a high risk for infections. Medicine is used to treat this disease. Babies with a hemoglobin disease need to see a medical specialist called a hematologist.

Sickle Cell Trait (SCT)

Babies with SCT have one gene that makes sickle-shaped red blood cells and one gene that makes normal red blood cells. They are usually not sick but can have kidney problems as they get older. They should get genetic counseling about sickle cell trait. Seeing a doctor for checkups every year is important too.

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. Treatment includes 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 cannot fight