

What is Spinal Muscular Atrophy (SMA)?

Spinal muscular atrophy, or SMA, is a condition that affects the nerve cells and nerves in the spinal cord. When these nerves don't work, muscles can't do their job and become very weak. Symptoms may include trouble breathing, problems with feeding or swallowing, difficulty in meeting motor milestones, or muscle weakness. If left untreated, these symptoms will get worse over time. Treatment should be started as soon as possible since babies with SMA tend to have better outcomes with early treatment.

What are Amino Acid Disorders, Organic Acid Disorders and Fatty Acid Oxidation Disorders?

Each of these conditions affects the body's ability to produce energy from food. These conditions cause the buildup of toxins or poisons in the body. Two of the most common conditions in this group are phenylketonuria (PKU) and medium chain acyl-CoA dehydrogenase deficiency (MCADD).

What is Phenylketonuria (PKU)?

PKU is a disorder that affects the body's ability to break down phenylalanine, which is part of the protein found in foods. The build up of phenylalanine in the blood can affect brain development and cause intellectual disability and behavioral problems. Treatment places the baby on a special infant formula. The child will follow a special diet lifelong.

What is Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)?

MCADD is a condition that affects the body's ability to make energy from stored fats. Babies born with this condition seem fine at birth, but they can suddenly have seizures and go into a coma if they have not eaten for a few hours. Without emergency treatment they may die or be left with developmental problems. Treatment includes avoiding long periods without eating, especially during times of illness.

What is X-Linked Adrenoleukodystrophy (X-ALD)?

X-ALD affects the brain and adrenal glands. People with X-ALD are unable to break down very long chain fatty acids (VLCFA), which can build up in the body and cause health problems. X-ALD is treatable if detected early. Without early detection and treatment, the most severe form of X-ALD can cause blindness and loss of muscle control.

What are Mucopolysaccharidosis (MPS) Types I and II?

MPS I and MPS II are rare inherited disorders that affect most parts of the body. Babies with MPS I or MPS II cannot break down a group of complex sugars. As a result, these sugars build up in cells and cause health problems. Starting treatment for MPS I or MPS II early has been shown to help prevent, stop, or delay many of the health problems. Babies with MPS I or MPS II can show signs in the first year of life. These symptoms can get worse quickly.

What is Pompe Disease?

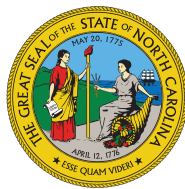
Pompe disease affects the baby's muscles. Babies with Pompe disease cannot break down a certain type of sugar, called glycogen. As a result, glycogen builds up in the body and causes health problems. The signs of Pompe disease may not be obvious at birth. In some cases, Pompe disease can become serious very quickly if it is not treated.



If you have any concerns about the results of the screening tests, please contact your baby's doctor.

Newborn screening is not a diagnostic test. Although a normal result is very reassuring, it does not guarantee that a baby does not have one of these disorders. If a baby develops symptoms of one of these disorders, the baby should be further examined, even if the newborn screen was normal.

Children with Special Health Care Needs Help Line 1-800-737-3028



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A TEST TO SAVE YOUR BABY'S LIFE

Newborn Screening

Your Questions Answered

How will this test help protect my baby?

Most babies are born healthy, but some health problems are not always found at birth. Newborn screening is the best way to help find and prevent serious health problems before your baby becomes sick. This is why North Carolina does a simple blood test to check newborns for over 30 conditions. Many of these conditions are life-threatening early in life and some can cause serious long-term problems if they are not treated early. The earlier a problem is found and treated, the better chance your baby will have for a healthy start in life!

How and where is my baby tested?

Before leaving the hospital, your baby's heel will be pricked and a few drops of blood will be collected. The blood will be sent to the State Laboratory of Public Health in Raleigh for testing. North Carolina is a nationally known leader in newborn screening.

What if my baby needs a retest?

After you get home from the hospital, your baby's doctor may ask you to bring your baby in for another newborn screen or other types of testing. If more testing is needed it is very important that you respond as soon as possible. Some of these conditions can cause life-threatening problems in just a few days.

What is included in the test?

The conditions your baby will be checked for will benefit from different kinds of medical care and treatment and they include:

- Congenital primary hypothyroidism
- Galactosemia
- Congenital adrenal hyperplasia (CAH)
- Sickle cell disease
- Biotinidase deficiency
- Cystic Fibrosis
- Severe Combined Immunodeficiency (SCID)
- Spinal Muscular Atrophy (SMA)
- Amino acid disorders (including phenylketonuria, PKU)
- Organic acid disorders
- Fatty acid oxidation disorders (including medium chain acyl-CoA dehydrogenase deficiency, MCADD)
- X-linked adrenoleukodystrophy (X-ALD)
- Mucopolysaccharidosis type 1 and II (MPS 1 and MPS II)
- Pompe disease

What if we have no family history of these disorders?

Most of these health conditions are inherited from other family members – passed down from grandparents or parents to their children. However, a baby can also be the first person in the family to have the condition. Parents who do not have a family history of these conditions, or have healthy children already, can still have a baby with one of these conditions. It is possible for both parents to be a carrier of a genetic condition even with healthy children. A carrier has a non-working gene that unknowingly can be passed from parent to child. Please be aware that newborn screening does NOT identify all newborns (or parents) that are carriers of an inherited genetic condition.



Explaining the Disorders

For each of these conditions, early diagnosis, medical care and treatment can prevent or reduce serious medical problems and may even save your baby's life. Some of these disorders may be life-threatening.

What is primary hypothyroidism?

The thyroid gland, which is located in the neck, makes a hormone that is important for normal growth, development, and learning. Primary hypothyroidism occurs when a baby's thyroid gland does not make enough thyroid hormone. Treatment is giving or replacing the hormone that is in short supply with a medicine the baby can take by mouth.

What is Galactosemia?

Galactosemia is a condition that does not allow the body to use a sugar called galactose that is found in milk. A baby with galactosemia can become very ill after just a few days of drinking breast milk or formula that contains this sugar. Some early signs and symptoms may include vomiting, yellowing of the skin or failure to grow or thrive. Treatment involves changing to a galactose-free (soy-based) formula which is easy to get from the store.

What is Congenital Adrenal Hyperplasia (CAH)?

CAH is a group of conditions that affect the amount of hormones produced by the adrenal glands, which are located above the kidneys. These hormones are needed for life and play an important role in sexual development. Symptoms of CAH can be life-threatening. Things to look for in your baby may include weakness, dehydration or even shock. If your baby has CAH, the baby's body cannot make these hormones. Treatment is by giving or replacing the hormone to the baby which is usually a medicine you can take by mouth.

What is Sickle Cell Disease?

Sickle cell disease is a problem with red blood cells. Sickle cell disease affects the part of the red blood cell that carries oxygen to all parts of the body. People with sickle cell disease can have serious health problems that can lead to anemia (low red blood cell count), severe pain, lifethreatening infections, strokes and many hospitalizations. Penicillin taken by mouth helps prevent and treat some symptoms.

What is Biotinidase Deficiency?

Biotinidase deficiency is a condition that affects the body's ability to recycle biotin, a common vitamin found in many foods and so there is not enough biotin in the body. Biotinidase deficiency can cause seizures, mental retardation, skin rash, hair and hearing loss and even death. Treatment consists of daily biotin which is a medicine taken by mouth.

What is Cystic Fibrosis?

Cystic Fibrosis (CF) is a condition that affects breathing and digestion or the body's use of food and liquids that the baby eats and drinks. People with CF have very thick mucus, which can clog the lungs and digestive system leading to a bad cough, life-threatening lung infections, and poor weight gain and growth. There are many kinds of treatments for CF that involve daily breathing therapy and medication. Early diagnosis and treatment can improve growth, lung function and add years to a child's life.

What is Severe Combined Immunodeficiency (SCID)?

SCID is an inherited condition in which the baby's immune system does not work well and the body is unable to fight off serious infections. If found and treated early, the baby can live and have a healthier life.