

What are amino acid disorders, organic acid disorders, and fatty acid oxidation disorders?

Each of these disorders affects the body's ability to produce energy from food. These conditions cause the buildup of toxins 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 severe mental retardation and behavioral problems. Treatment places the baby on a special infant formula and life-long diet.

What is medium chain acyl-CoA dehydrogenase deficiency (MCADD)?

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



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.



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A TEST TO SAVE YOUR BABY'S LIFE



NEWBORN SCREENING

YOUR QUESTIONS ANSWERED

How will this test help protect your baby?

Most babies are born healthy and normal, but there are some health problems that are not always spotted at birth. Newborn screening is the best way to help identify and prevent serious health problems before your baby becomes sick. This is why North Carolina performs a simple blood test to check newborns for over 30 conditions. Many of these illnesses are life-threatening and 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 your 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 recognized leader in newborn screening.

What if your 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 requested **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 are all treatable and they include:

- Congenital primary hypothyroidism
- Galactosemia
- Congenital adrenal hyperplasia (CAH)
- Sickle cell disease
- Biotinidase deficiency
- Amino acid disorders (including phenylketonuria, PKU)
- Organic acid disorders
- Fatty acid oxidation disorders (including medium chain acyl-CoA dehydrogenase deficiency, MCADD)

What if we have no family history of these disorders?

Most of these health problems are inherited – passed down in families. However, a new baby can be the first person in the family to have the illness. 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 illnesses.

EXPLAINING THE DISORDERS

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

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 by hormone replacement.

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. The early symptoms may include vomiting, liver damage, or failure to thrive. Treatment involves changing to a galactose-free (soy-based) formula.

What is congenital adrenal hyperplasia (CAH)?

CAH is a group of disorders that affect the amount of hormones produced by the adrenal glands, which are located above the kidneys. These hormones are necessary for life and play an important role in sexual development. Symptoms of CAH can be life-threatening and may include weakness, dehydration, or even shock. If your baby has CAH, the baby's body cannot make these hormones. Treatment is by hormone replacement.

What is sickle cell disease?

Sickle cell disease is a disorder of red blood cells. Sickle cell disease affects the part of the red blood cell that carries oxygen throughout the body. People with sickle cell disease can have serious health problems that can lead to anemia (low red blood cell count), severe pain, life-threatening infections, strokes, and many hospitalizations. Penicillin helps prevent and treat 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. Biotinidase deficiency can cause seizures, mental retardation, skin rash, hair and hearing loss, and even death. Treatment consists of daily biotin.