

Overview of Newborn Screening for Amino Acid Disorders – For Parents

What is newborn screening?

Before babies go home from the nursery, they have a small amount of blood taken from their heel to test for a group of conditions, including **amino acid disorders**. Babies who screen positive for an amino acid disorder need follow-up tests done to confirm they have the condition. **Not all babies with a positive newborn screen will have an amino acid disorder.**

What are amino acid disorders?

Amino acid disorders are conditions that occur when a person's body is not able to break down protein. Normally, when we eat, our bodies digest (or break down) food into protein. Protein is then broken down into **amino acids**. Our bodies use amino acids to make energy. **Enzymes** (special proteins that help our bodies perform chemical reactions) usually help our bodies break down food and make energy.

A person with an amino acid disorder is missing at least one enzyme, or his/her enzymes do not work correctly. When these enzymes are missing or do not work, food cannot be broken down and made into energy. If food cannot be broken down, dangerous substances build up in the body. This build-up can happen shortly after birth.

What causes amino acid disorders?

Amino acid disorders are **inherited** (passed from parent to child) conditions. Everyone inherits two copies of the genes that cause amino acid disorders. We receive one copy of each gene from our fathers, and one copy from our mothers. Sometimes these genes have changes (also called mutations) that prevent them from working correctly.

In order for a person to have an amino acid disorder, he or she must have two changed copies of the gene that causes a particular amino acid disorder. People with one amino acid gene change do not have an amino acid disorder.

What amino acid disorders are on Indiana's newborn screen?

Indiana's newborn screen tests for the following amino acid disorders:

- Citrin deficiency
- Homocystinuria
- Maple syrup urine disease (also called MSUD)
- Phenylketonuria (also called PKU)
- Tyrosinemia
- Urea cycle disorders

What are the symptoms of amino acid disorders?

Every child with an amino acid disorder is different. Most babies with amino acid disorders will look normal at birth. Symptoms of amino acid disorders can appear shortly after birth, or they may show up later in childhood. Common symptoms of amino acid disorders include vomiting, poor feeding, damage to the body's organs (such as the liver), mental retardation, developmental delay, or other health problems.

What is the treatment for amino acid disorders?

There is no cure for amino acid disorders. However, there are special diets and supplements that can help with the symptoms. Children who have amino acid disorders will need to be on these treatments for the rest of their lives.

What happens next?

Good medical care makes a difference for children with amino acid disorders. These children should see a Metabolic Geneticist (a doctor who specializes in amino acid disorders and other related conditions) as well as their pediatrician. Your child's doctor will work with the Metabolic Geneticist to set up any treatment, tests, or appointments that your child needs.

Call your child's doctor or the Metabolic Genetics clinic if your baby has poor feeding, extreme sleepiness or fussiness, or seizures. Be sure your baby is fed every 4 hours (including at night).

Where is Indiana's Metabolic Genetics Clinic? Indiana's Metabolic Genetics Clinic is located at Riley Hospital for Children in Indianapolis. You can reach the Metabolic Genetics Clinic by calling (317) 274 – 3966.

For more information

- **MedLine Plus Medical Encyclopedia** - <http://www.nlm.nih.gov/medlineplus/>
- **Region 4 Genetics Collaborative** - http://region4genetics.org/family_resources/genetic_conditions.aspx