Indiana State Department of Health

Overview of Newborn Screening for Organic Acidemias – 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 **organic acidemias**. Babies who screen positive for an organic acidemia need follow-up tests done to confirm they have the condition. **Not all babies with a positive newborn screen will have an organic acidemia.**

What are organic acidemias?

Organic acidemias are conditions that occur when a person's body is not able to use protein to make energy. Normally, when we eat, our bodies digest (or break down) food into certain proteins. Those proteins are used by our bodies to make energy. **Enzymes** (special proteins that help our bodies perform chemical reactions) usually help our bodies break down food and create energy.
A person with an organic acidemia is missing at least one enzyme, or his/her enzymes do not work correctly. When these enzymes are missing or do not work correctly, 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 organic acidemias?

Organic acidemias are **inherited** (passed from parent to child) conditions. Everyone inherits two copies of the genes that cause organic acidemias. We receive one copy of each gene from our fathers and one copy of each gene from our mothers. Sometimes these genes have changes (also called mutations) that prevent the gene from working correctly.
In order for a person to have an organic acidemia, he or she must have two changed copies of the gene that causes a particular organic acidemia. People with one organic acidemia gene change do not have an organic acidemia.

What organic acidemias are on Indiana's newborn screen?

Indiana's newborn screen tests for several organic acidemias. Some of the organic acidemias on Indiana's newborn screen are:

- 3-Methylcrotonyl-CoA carboxylase deficiency (also called 3-MCC deficiency)
- Glutaric acidemia, type I
- Isovaleric acidemia
- Methylmalonic acidemia
- Multiple-CoA carboxylase deficiency
- Propionic acidemia

What are the symptoms of organic acidemias?

Every child with an organic acidemia is different. Most babies with organic acidemias will look normal at birth. Symptoms of organic acidemias can appear shortly after birth, or they may show up later in infancy or childhood. Common symptoms of organic acidemias include weakness, vomiting, low blood sugar, hypotonia (weak muscles), spasticity (muscle stiffness) or other health problems.

What is the treatment for organic acidemias?

There is no cure for organic acidemias. However, there are special diets and supplements that can help with the symptoms. A person with an organic acidemia will need treatment for his/her entire life.

What happens next?

Good medical care makes a difference for children with organic acidemias. These children should see a metabolic geneticist (a doctor who specializes in organic acidemias 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 four hours (including at night).

Where are Indiana's metabolic genetics clinics?

Indiana's metabolic genetics clinics are located at Riley Hospital for Children at IU Health, Indianapolis, IN, (317) 274-3966 and The Community Health Clinic, Topeka, IN, (260) 593-0108.

Where can I get more information about organic acidemias?

- **STAR-G -** http://www.newbornscreening.info/Parents/facts.html
- **Region 4 Genetics Collaborative –** https://www.region4genetics.org/education/families/