Indiana State Department of Health

Overview of Newborn Screening for Fatty Acid Oxidation 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 that includes **fatty acid oxidation disorders**. Babies who screen positive for a fatty acid oxidation disorder need follow-up tests done to confirm they have the condition. **Not all babies with a positive newborn screen will have a fatty acid oxidation disorder.**

What are fatty acid oxidation disorders?

Fatty acid oxidation disorders (also called FAO disorders) are conditions that occur when a person's body cannot use some fat from food. Normally, when we eat, our bodies use some food right away to make energy so that we can keep moving and working. However, some food is stored as fat for use later. When a person has not eaten for a period of time, the body starts to use this stored fat for energy. People with FAO disorders are not able to use this stored fat for energy.

Enzymes (proteins that help our bodies perform chemical reactions) usually help the body use stored fat. A person with a FAO disorder is missing at least one enzyme, or his/her enzymes don't work correctly.

What causes fatty acid oxidation disorders?

FAO disorders are **inherited** (passed from parent to child) conditions. Everyone inherits two copies of the genes that cause FAO 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 the gene from working correctly.

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

What FAO disorders are on Indiana's newborn screen?

Indiana's newborn screen tests for several fatty acid oxidation disorders, including:

- Acyl-CoA dehydrogenase deficiencies (MCAD, SCAD, VLCAD)
- Carnitine palmitoyltransferase deficiency types I and II (CPT I & CPT II)
- Glutaric acidemia type II
- Hydroxyacyl-CoA dehydrogenase deficiencies (SCHAD and VLCHAD)
- Primary carnitine deficiency

What are the symptoms of fatty acid oxidation disorders?

Every child with a fatty acid oxidation disorder is different. Most babies with FAO disorders will look normal at birth. Common symptoms of fatty acid oxidation disorders include vomiting, seizures, liver problems and possibly coma or death.

What is the treatment for fatty acid oxidation disorders?

There is no cure for fatty acid oxidation disorders. However, there are treatments that can help with the symptoms. People with FAO disorders should eat frequently and avoid fasting (long periods of time without food). They may also need a special diet or take special medicines. A person with an FAO disorder will need treatment for his/her entire life.

What happens next?

Good medical care makes a difference for children with fatty acid oxidation disorders. These children should see a metabolic geneticist (a doctor who specializes in FAO 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 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 fatty acid oxidation disorders?

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