

Overview of Newborn Screening for MCAD Deficiency (MCADD) – 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. One of these conditions is **medium chain acyl-CoA dehydrogenase deficiency** (also called **MCAD deficiency** or **MCADD**). Babies who screen positive for MCADD need follow-up tests done to confirm they have MCADD. **Not all babies with a positive newborn screen will have MCADD.**

What is MCAD deficiency, or MCADD?

MCADD is one of a group of conditions called **fatty acid oxidation disorders** (also called **FAO disorders**). 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 MCADD has low levels of an enzyme called **medium chain acyl-CoA dehydrogenase** (also called **MCAD**). Without MCAD, a person's body cannot use stored fat to make energy. This causes dangerous substances to build up in the blood of a person with MCADD.

What causes MCADD?

MCADD is an **inherited** (passed from parent to child) condition. Everyone inherits two copies of the MCAD gene (one from our fathers and one 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 MCADD, he or she must have two MCAD gene changes. People with one MCAD gene change do not have MCADD.

What are the symptoms of MCADD?

Every child with MCADD is different. Most babies with MCADD will look normal at birth. Symptoms of MCADD most often appear around three months of age, but they can also show up shortly after birth or later in childhood. Without treatment, people with MCADD can have a **metabolic crisis** (period of illness). Symptoms of a metabolic crisis include poor appetite, sleepiness, low blood sugar, and vomiting. If a person with MCADD does not receive treatment, he/she can also have breathing problems, seizures, liver problems, or possible coma or death.

What is the treatment for MCADD?

There is no cure for MCADD. The symptoms of MCADD often appear when a person has not had any food for several hours or when he/she has an illness where he/she is not eating. People with MCADD should eat frequently and avoid fasting (long periods of time without food). People who have MCADD will need to follow a special low-fat diet and may be given a special medication called **L-carnitine**. A person with MCADD will need treatment for his/her entire life.

What happens next?

Although there is no cure for MCADD, good medical care makes a difference. Children with MCADD should see a metabolic geneticist (a doctor who specializes in MCADD and other related conditions) as well as their pediatrician. Your child's doctor will work with the metabolic geneticist to coordinate 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 MCADD?

- **STAR-G** - <http://www.newbornscreening.info/Parents/fattyacid disorders/MCADD.html>
- **Region 4 Genetics Collaborative** – <https://www.region4genetics.org/education/families/>