Indiana State
Department of Health

Overview of Newborn Screening for 3-MCC Deficiency – 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 **3-methylcrotonyl CoA carboxylase deficiency** (also called **3-MCC deficiency**). Babies who screen positive for 3-MCC deficiency need follow-up tests done to confirm they have 3-MCC deficiency. **Not all babies with a positive newborn screen will have 3-MCC deficiency.**

What is 3-MCC deficiency?

3-MCC deficiency is one of a group of conditions called **organic acidemias**. People with organic acidemias are unable to use protein from food 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** (proteins that help our bodies perform chemical reactions) usually help our bodies break down food and create energy.

A person with **3-MCC deficiency** has low levels of an enzyme called **3-methylcrotonyl CoA carboxylase** (also called 3-MCC). Without 3-MCC, a person cannot break down an amino acid called **leucine**. Leucine is found in any food that has protein. When a person with 3-MCC deficiency eats food containing leucine, his/her body cannot break down the leucine, which then builds up in the blood.

What causes 3-MCC deficiency?

3-MCC deficiency is an **inherited** (passed from parent to child) condition. Everyone inherits two copies of the 3-MCC 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 3-MCC deficiency, he or she must have two 3-MCC gene changes. People with one 3-MCC gene change do not have 3-MCC deficiency.

What are the symptoms of 3-MCC deficiency?

Every child with 3-MCC deficiency is different. Most babies with 3-MCC deficiency will look normal at birth. Symptoms of 3-MCC deficiency can appear around 3 months of age, or they may show up later in childhood. Some people with 3-MCC deficiency will never have symptoms.

Without treatment, people with 3-MCC deficiency can have a **metabolic crisis** (period of illness). Symptoms of a metabolic crisis include poor appetite, sleepiness, behavior changes, muscle weakness, nausea and vomiting. If a metabolic crisis is not treated, a person with 3-MCC deficiency can also have breathing problems, seizures, or other problems. Most people with 3-MCC deficiency are healthy in between metabolic crises.

What is the treatment for 3-MCC deficiency?

There is no cure for 3-MCC deficiency. However, there are treatments that can help prevent the symptoms of 3-MCC deficiency. Children who have 3-MCC deficiency will need to follow a special diet low in leucine and take a medication called **L-carnitine**. Some people with 3-MCC deficiency will never need treatment; other people will need to be on treatment permanently.

What happens next?

Although there is no cure for 3-MCC deficiency, good medical care makes a difference. Children with 3-MCC deficiency should see a Metabolic Geneticist (a doctor who specializes in 3-MCC deficiency 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 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.

Where can I get more information about 3-MCC deficiency?

- **STAR-G** - http://www.newbornscreening.info/Parents/organicaciddisorders/3MCC.html
- **Region 4 Genetics Collaborative** - http://region4genetics.org/family_resources/gc_organicaciddisorders.aspx