3-METHYLCROTONYL-CoA CARBOXYLASE DEFICIENCY (3-MCC)

FAMILY FACT SHEET

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called 3-MCC.

A positive newborn screen does not mean your baby has 3-MCC, but it does mean your baby needs more testing to know for sure.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing. Mothers of babies with positive newborn screens also need testing.

What is 3-MCC?

3-MCC affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. In 3-MCC, the enzymes used to break down proteins are missing or not working properly.

A person who has 3-MCC doesn't have enough enzyme to break down protein containing leucine, which can cause harmful toxins to build up in the body.

3-MCC is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause 3-MCC. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What problems can 3-MCC cause?

3-MCC is different for each child. Some children with 3-MCC have few health problems, while others may have serious complications.

If 3-MCC is not treated, a child might develop:

- Serious illness (Metabolic crisis)
- Muscle weakness
- Sleepiness
- Seizures
- Poor growth
- Brain damage
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

Many people with 3-MCC have no obvious problems, so they do not even know they have it. This is why mothers of babies with 3-MCC need testing. Sometimes, a baby has a positive newborn screen because the mother has a hidden form of 3-MCC.

What is the treatment for 3-MCC?

3-MCC can be treated. Treatment is life-long and can include:

- A low-protein diet – a dietitian will help you with the best diet for your child.
- Medications to help the body get rid of harmful toxins.

Children with 3-MCC should see their regular doctor, a doctor who specializes in 3-MCC, and a dietitian.

Prompt and careful treatment helps children with 3-MCC live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Children's Special Health Care Services
Toll-free: 1-800-359-3722

Michigan Metabolic Coordinating Center
C.S. Mott Children's Hospital, Michigan Medicine:
1-734-764-0579

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Phone 1-866-673-9939
www.michigan.gov/newbornscreening