

3-methylcrotonyl-CoA carboxylase deficiency 3-MCC

(3-methyl-crow-ton-eel co-A car-box-il-ace de-fish-in-sea)

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests 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. There are many other conditions that can cause a similar positive result on newborn screening.

A positive newborn screen does not mean your baby has 3-MCC or any of the other conditions above, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like 3-MCC. Sometimes, a baby has a positive newborn screen because the mother has a hidden form of 3-MCC. Mothers of babies with positive newborn screens also need testing.

What is 3-MCC?

3-MCC affects a special 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 with 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 disorder that is passed on, or inherited, from a child's mother and father. Because 3-MCC is a genetic disease, family members are at risk of having 3-MCC too, even if no one in the family has had it before.

What problems can 3-MCC cause?

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

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

- Serious illness (metabolic crisis)
- Muscle weakness
- Sleepiness
- Seizures
- Poor growth
- Mental retardation
- 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 don't even know they have it. This is why mothers of babies with 3-MCC need testing.

What is the treatment for 3-MCC?

3-MCC can be treated. The treatment is life-long. Treatment for children with 3-MCC can include:

- Medications to help the body get rid of harmful toxins
- Low-protein diet - a dietician will help you set up the best diet for your child

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

Children with 3-MCC can benefit from prompt and careful treatment.

Resources for parents

Genetics Home Reference
<http://ghr.nlm.nih.gov>

Organic Acidemia Foundation
www.oaanews.org

MN Children & Youth with Special Health Needs
(651) 201-3650 or (800) 728-5420

MN Newborn Screening Program
www.health.state.mn.us/newbornscreening