

## 3-Methylcrotonyl CoA carboxylase (3-MCC) deficiency

### What is the Wisconsin Newborn Screen?

All babies in Wisconsin have a test called the Newborn Screen (NBS) in their first 24 hours of life. A blood sample is taken from baby's heel for this test. This test looks for rare medical conditions that may not be caught or show up in your baby right away.

Newborn screening gets babies the help they need by:

- Finding out they have a condition before they have any medical problems
- Allowing treatment to be started right away
- Preventing medical emergencies

### What was my baby's NBS result?

Your baby's NBS has screened positive with a high level of a marker called "C5-OH acylcarnitine." This means your baby may have called 3-methylcrotonyl CoA carboxylase deficiency (3-MCC) or other conditions that are less common.

This test is a screening test. We will need more information to find out if he or she has this diagnosis.

### What is 3-MCC and why is it important?

3-MCC affects how protein is used in the body to make energy. Most people with 3-MCC are healthy and do not have any signs of the condition. The other disorders that can cause high C5-OH acylcarnitine can cause problems in newborns, including difficulty eating, staying awake, or responding to you.

### What are the next steps?

**Feed your baby every three hours, even overnight.**

The most important thing you can do while waiting for test results is to make sure your baby eats every three hours, even overnight, to help their body make energy. Do this until your doctor tells you it is safe to stop.

### Tests and Doctor's Visits

This will help us know if your baby has 3-MCC or not. Your baby's health care provider will order:

- An urgent visit to the primary care clinic and/or emergency department
- Blood and urine (pee) tests
- Referral to the Genetics Clinic is possible but not always needed

### What if I have questions or concerns?

**Contact your primary care provider right away:** If your baby is not eating well, gets sick, or you have questions or concerns about how he or she is doing.

**Go to the emergency room right away:** If your baby is not eating, throwing up, very sleepy and not waking up, has a seizure, or has other symptoms you are worried about.

**Call the Children's Wisconsin Genetics Center:** If you have questions about Newborn Screen, 3-MCC, or other disorders, or your baby's test results.

- Call 414-266-3347, option 3
- Leave a message and a nurse or genetic counselor from the Newborn Screen program will call back.
- Office is open Monday-Friday, 8 AM-4PM

Scan the QR codes below for links to more information

Medline Plus  
Genetics:



NewbornScreening.Info:

