NORTH CAROLINA NEWBORN SCREENING PROGRAM

Medium-chain acyl-coenzyme A dehydrogenase (MCAD) Deficiency or Medium-chain ketoacyl-CoA thiolase (MCKAT) Deficiency

(Newborn screening result: C8 and C8/C10 elevations)

Fact Sheet for Parents

All babies born in North Carolina are screened at birth to look for health problems that can be treated if caught early. The newborn screening result showed that your baby might have medium-chain acyl-coenzyme A dehydrogenase (MCAD) deficiency or Medium-chain ketoacyl-CoA thiolase (MCKAT) deficiency. **Most commonly the diagnosis with C8 and C8/C10 screening result is MCAD deficiency.** Your baby will be referred to a specialist for more testing to know for sure.

There are usually no signs of MCAD or MCKAT deficiency at birth.

What is MCAD deficiency?

MCAD deficiency is a rare genetic condition that prevents the body from breaking down certain fatty acids and using them for energy. This can cause low blood sugar (hypoglycemia) when a baby has not eaten for a while (fasting) leading to serious health problems and possibly sudden infant death. With early diagnosis and treatment, individuals with MCAD deficiency will live a healthy life.

What are the signs and symptoms of MCAD deficiency?

Symptoms of MCAD deficiency may not be visible at birth, and are not the same for everyone, signs and symptoms can include:

- Low blood sugar
- Vomiting
- Low energy
- Seizures
- Breathing difficulties
- Coma
- Death

What happens next?

Your baby's pediatrician will refer your baby to a specialist who treats babies and children with MCAD deficiency. It is important for your baby to be seen by the specialist as soon as possible. Until then, feed your baby every 2-3 hours. Your child's doctor may request additional testing to confirm your baby has MCAD deficiency.

How is MCAD deficiency treated?

A baby with MCAD deficiency must eat frequently, every 2-3 hours.

- If you are breastfeeding, offer either pumped breastmilk or formula after every breastfeeding session.
- Call your pediatrician if you are concerned about your infant's feeding.
- **If your baby is vomiting or has a fever, or you are unable to wake your baby to feed, call 911 or take your baby to the emergency department.**

As your baby grows, frequent eating is important, but the time between feedings will get longer. School-age children and adults with MCAD deficiency do not need to wake during the night to eat. Your baby's healthcare provider will provide guidance on management as your baby grows. MCAD deficiency is a lifelong condition, but with the proper diet and lifestyle support, children with MCAD deficiency develop normally and lead healthy, active lives.

Where do I go for more information?

Use your phone's camera to scan the QR codes below.

Newborn Screening Information Center

Genetic and Rare Disease Information Center

FOD patient advocacy group

Mayo clinic

MAY 20, 1775
APRIL 12, 1776

NC DEPARTMENT OF
HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services Division of Public Health

www.ncdhhs.gov

https://slph.dph.ncdhhs.gov/newborn/default.asp

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