NORTH CAROLINA NEWBORN SCREENING PROGRAM

Acylcarnitines: C8 and C8/C10 elevations

Possible conditions

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

Fact Sheet for Providers

What do C8 and C8/C10 elevations lead to?

C8 and C8/C10 elevations result from a deficiency of either medium-chain acyl-coenzyme A dehydrogenase (MCAD) enzyme or medium-chain ketoacyl-CoA thiolase (MCKAT) enzyme. MCAD and MCKAT deficiencies are fatty acid oxidation disorders that suppress the body's ability to convert medium-chain fatty acids into energy, particularly during periods of fasting or illness. MCAD deficiency is the most likely diagnosis associated with these elevations.

MCAD deficiency is life-threatening and requires immediate attention. A diagnosis of MCAD deficiency can result in hypoglycemia, lethargy, seizures, or even sudden death if untreated.

- **Until diagnostic evaluation is complete, the infant should be fed every 2-3 hours.**
- **AVOIDANCE OF FASTING IS CRITICAL.**
- **If the mother is breastfeeding, then the family should supplement after every feed with either pumped breastmilk or formula.**

With early diagnosis and treatment, individuals with MCAD deficiency will live a healthy life. MCKAT deficiency can also be associated with this screening result but is rare.

What are the signs and symptoms of MCAD deficiency?

Signs and symptoms of MCAD or MCKAT deficiency are not often present at birth but typically appear during periods of fasting or illness and include:

- Hypoglycemia
- Lethargy
- Vomiting
- Seizure
- Coma
- Hepatomegaly
- Sudden death (especially during the newborn period)

How is MCAD deficiency diagnosed?

Infants with an abnormal newborn screening result for MCAD deficiency require additional testing to confirm the diagnosis. This will include biochemical testing and may also include genetic testing of the *ACADM* gene.

How is MCAD deficiency treated?

Because individuals with MCAD deficiency cannot convert medium-chain fats for energy, especially during periods of fasting or illness, treatment focuses on preventing low blood sugar levels and metabolic stress. Individuals with MCAD deficiency should avoid fasting. Infants should be fed every 2-3 hours, prior to bedtime, and feedings overnight. **If the mother is breastfeeding, then the family should supplement after every feed with either pumped breastmilk or formula.** Newborns identified with MCAD deficiency should be referred to a tertiary care team, including a dietitian, familiar with the diagnosis of management of inborn errors of metabolism. This team will advise about emergency care protocols for management of intercurrent illness and/or prolonged fasting.

Strategies for managing MCAD deficiency include:

- Avoiding prolonged fasting (especially during illness)
- Reaching out to the metabolic team during illness to determine if IV dextrose therapy is needed

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How do I handle abnormal screening for MCAD deficiency?

This result is considered an urgent result. Immediately reach out to the family to confirm that the infant is eating every 2-3 hours. If the mother is breastfeeding, the infant should be offered supplemental feeding, either pumped breastmilk or formula, after every feed. If there is any concern regarding lethargy or inadequate intake (i.e. mother's milk supply is not yet in), the infant should be immediately assessed either at the office or local emergency department for possible hypoglycemia. Please contact UNC Genetics and Metabolism by calling the UNC Hospital's paging operator (984-974-1000, request the pediatric geneticist on call).

Where do I go for more information?

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

MCAD ACT sheet

GeneReviews

Mayo Clinic: MCAD deficiency

Where do I send parents for 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

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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