



Newborn Screening ACT Sheet

Elevated C8 with Lesser Elevations of C6 and C10 Acylcarnitine Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

Differential Diagnosis: Medium-chain acyl-CoA dehydrogenase deficiency (MCAD).

Condition Description: MCAD deficiency is a fatty acid oxidation (FAO) disorder. FAO occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In an FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO enzymes.

Medical Emergency: Take the Following IMMEDIATE Actions

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist. (See attached list.)
- Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly).
- If signs are present or infant is ill, initiate emergency treatment with IV glucose. Transport to hospital for further treatment in consultation with metabolic specialist.
- If infant is normal, initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Initial testing: plasma acylcarnitine profile; urine organic acids (including acylglycines) and plasma carnitine levels.
- Repeat newborn screen if the second screen has not been done.
- Educate family about need for infant to avoid fasting. Even if mildly ill, immediate treatment with IV glucose is needed.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine analysis will show elevated octanoylcarnitine (C8). Urine acylglycines will show elevated hexanoylglycine. Diagnosis is confirmed by mutation analysis of the MCAD gene.

Clinical Considerations: MCAD deficiency is usually asymptomatic in the newborn, although it can present acutely in the neonate with hypoglycemia, metabolic acidosis, hyperammonemia, and hepatomegaly. MCAD deficiency is associated with high mortality unless treated promptly; milder variants exist. Hallmark features include vomiting, lethargy, and hypoketotic hypoglycemia. It is a significant cause of sudden death. Acylcarnitines may normalize on the second screen on affected babies, therefore an infant with an out of range first newborn screen and normal second newborn screen will still need a metabolic evaluation.

Additional Information:

Emergency Treatment Protocol

<http://www.childrenshospital.org/newenglandconsortium/NBS/MCADD.html>

Gene Tests

<http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=NexQvDbtfnPSK&gry=&fcn=y&fw=091z&filename=/profiles/mcad/index.html>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=mediumchainacylcoenzymeadehydrogenasedeficiency>

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<http://www.newbornscreening.info/Pro/fattyacid disorders/MCADD.html>

<http://www.newbornscreening.info/Parents/fattyacid disorders/MCADD.html>



Newborn Screening FACT Sheet

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

What is MCAD?

MCAD is a type of fatty acid oxidation disorder. People with MCAD have problems breaking down fat into energy for the body.

What Causes MCAD?

Enzymes help start chemical reactions in the body. MCAD happens when an enzyme called “medium chain acyl-CoA dehydrogenase” is either missing or not working. This enzyme breaks down certain fats in the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with MCAD?

[Symptoms are something out of the ordinary that a parent notices.]

MCAD can cause bouts of illness called Metabolic Crises. Children with MCAD often show symptoms for the first time between 3 months and 2 years of age. Some of the first signs of a Metabolic Crisis are:

- too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- poor appetite

If a Metabolic Crisis is not treated, a child with MCAD can develop:

- breathing problems
- seizures
- mental retardation
- cerebral palsy
- coma, sometimes leading to death

What is the Treatment for MCAD?

The following treatments are often used for children with MCAD:

1. Do not go a long time without food – Babies and young children with MCAD need to eat often to avoid low blood sugar or a Metabolic Crisis. They should not go without food for more than 4 to 6 hours. Some babies need to eat even more often. It is important that

babies be fed during the night. They need to be woken to eat if they do not wake up on their own. Young children with MCAD may need to have a starchy snack (such as bread, cereal, rice) before bed and another during the night. They may need another snack first thing in the morning. Your dietitian can give you ideas for good night-time snacks. Dietitians know what are the correct foods to eat. Most teens and adults with MCAD can go without food for up to 12 hours without problems when they are well. They need to continue the other treatments for life.

2. Diet – Sometimes a low-fat, high carbohydrate diet (such as vegetables, fruits, grains) is advised. Your dietitian can create a food plan with the right type and amount of fat your child needs. Ask your doctor whether or not your child needs to have any changes in his or her diet.

3. L-carnitine – L-carnitine (Carnitor) may be prescribed for some children. This is safe and natural and helps body cells make energy. It also helps the body get rid of harmful wastes.

Things to Remember

Always call your doctor when your child has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever

People with MCAD need to eat extra starchy foods and drink more fluids during any illness – even if they don’t feel hungry – or they could develop low blood sugar or a Metabolic Crisis. Children who are sick often don’t want to eat. If they won’t or can’t eat, they may need to be treated in the hospital to prevent problems.