

# MCAD

## Medium-Chain Acyl-CoA Dehydrogenase Deficiency

### Action required

Contact metabolic specialist today.  
See infant today.

### Issues to discuss with metabolic specialist

- Laboratory evaluation of infant
  - § Should testing be performed by primary care or metabolic clinic
  - § Plasma acylcarnitine analysis
    - 0.1 mL frozen plasma in sodium heparin green top tube
  - § Urine organic acids
    - 4.0 mL random urine, frozen
  - § Urine acylglycine
    - 5.0 mL random urine, frozen

### False Positives

- Common
- Screening result can be impacted by carnitine supplementation and drug therapies such as valproic or benzoic acid

### Review with family

Family has **not** been notified of result by MDH.

After discussion with metabolic specialist, contact family to coordinate clinic visit, lab work, and referral to metabolic clinic. Expect infant to be stable when family is contacted and at clinic visit - though some infants do present with acute symptoms.

Prompt follow-up is important.

### NICU issues

Newborn screens cannot be accurately interpreted after administration of TPN.

### Clinical summary

Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is an autosomal recessive disorder that results from the defective activity of MCAD, an enzyme involved in fatty acid oxidation. Newborns are usually asymptomatic. If an infant is not screened and or left untreated, symptoms begin to appear early in infancy and can include hypoketotic hypoglycemia, hepatomegaly, vomiting, lethargy, and sudden death, especially with fasting.

Affected children require a life-long low fat/high carb diet, avoidance of fasting, carnitine supplementation, and monitoring by both primary care and specialty providers.

**Incidence:** ~ 1/10,000; more prevalent in Caucasian individuals

### Clinical expectations

If treated promptly, children with MCAD can be expected to be clinically stable and develop normally.

Affected children may need hospitalization during intercurrent illness - even if mildly ill - for treatment with IV dextrose.

Affected children should be monitored for dietary compliance and hypoglycemia.

### Resources

**GeneTests:** [www.genetests.org](http://www.genetests.org)

**OMIM:** [www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM](http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM)

**ACT Sheets:** [www.acmg.net/resources/policies/ACT/condition-analyte-links.htm](http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm)

**MN Newborn Screening Program:**  
[www.health.state.mn.us/newbornscreening](http://www.health.state.mn.us/newbornscreening)