

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

Health Care Professional Fact Sheet

A newborn screening test is a **screen** and not diagnostic testing. An “abnormal” or “critical” result on a newborn screen indicates the baby may be at a higher risk of having a disorder; however, it does not diagnose the baby with the condition. Follow-up testing is **vital** to determine if the baby has the disorder indicated. In the event the condition is diagnosed, timely follow-up testing will result in earlier treatment and better outcomes.

Disorder Indicated: Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) is a fatty acid oxidation disorder in which the body cannot break down certain fats to use for energy due to an inherited enzyme deficiency. This results in an accumulation of fatty acids in the body. If left untreated, MCAD could cause brain damage, coma, or death. However, if the condition is detected early and treatment is begun, individuals with MCAD can have healthy growth and development.

Incidence	Estimated 1 in every 15,000 newborns.
Analyte Measured	Primary Marker: C8 (<i>octanoylcarnitine</i>) Secondary Markers*: C6 (<i>hexanoylcarnitine</i>), C10 (<i>decanoylcarnitine</i>), or C8/10 <i>*an abnormal secondary marker paired with an abnormal primary marker will result in critical group and require referral and diagnostic testing</i>
Normal Test Results	C8 <0.50 µmol/L
Abnormal Test Results	C8 ≥ 0.50 µmol/L to < 1.0 µmol/L
Critical Test Results	C8 ≥ 1.0 µmol/L (Critical results require immediate evaluation and follow-up)
Signs and Symptoms <i>Please note: these findings may not be present in young infants or in milder forms of the disease</i>	When a child has MCAD, you may see symptoms including: <ul style="list-style-type: none"> • Poor appetite • Tiredness/Sleeping longer or more often • Vomiting/Diarrhea • Hypoglycemia (low blood sugar) • Behavior changes/irritability • Breathing difficulty • Seizures <p><i>Symptoms can be triggered or exacerbated by periods of fasting, illness, or infections.</i></p>
Next Steps may include:	Discuss the next steps of evaluation and possible treatment with the regional Geneticist Provide parental education (see accompanying sheet) Clinical Assessment Assay: Plasma Acylcarnitines, Urine Organic Acids
Treatment (if indicated)	Restricted diet/Supplements (Discuss with the regional Geneticist)
Additional Resources	VDH Newborn Screening http://vdhlivewell.com/newbornscreening Baby’s First Test www.babysfirsttest.org American College of Medical Genetics (ACMG) ACT Sheets www.ACMG.net Genetics Home Reference https://ghr.nlm.nih.gov/ Fatty Oxidation Disorder Family Support Group http://www.fodsupport.org/

Educational content adapted from www.babysfirsttest.org

