

Physician ACT SHEET for POSITIVE Newborn Screening Result for MCAD (Medium Chain Acyl-CoA Dehydrogenase Deficiency)

Disease Category: Inborn error of fatty acid oxidation metabolism

POTENTIAL NEONATAL EMERGENCY YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- **Contact family to inform them of the newborn screening result, provide feeding instructions (at least every 4 hours) and schedule an immediate visit. If infant is lethargic or not feeding well emergency care is warranted.**
- **Emergency treatment includes avoiding fasting, determining blood glucose level and provide intravenous glucose if hypoglycemic or symptomatic**
- **Immediately consult a metabolic specialist.**
- **Undertake definitive investigations in consultation with metabolic specialist.**
- **Report findings back to state newborn screening program.**

Pediatric metabolic specialists are available through the metabolic centers at Children's Hospital and UNMC/Nebraska Medical Center. The pediatric metabolic specialist on service/call today is: _____ MD and can be paged at: _____

Meaning of the Screening result: Substantially elevated C8 acylcarnitine (octanoylcarnitine) likely indicates MCAD. Elevations of C8 slightly above, or above normal may indicate MCAD, MADD or a variant.

Metabolic Description: Fatty Acid Oxidation Disorders (FAOD's) impair utilization of fatty acids as an energy source. MCAD is due to a defect in the mitochondrial enzyme medium chain acyl-CoA dehydrogenase, which is responsible for a step in fatty acid oxidation. Hallmark features can include critical hypoketotic hypoglycemia, especially during times of fasting, catabolism, or illness.

Confirmation of Diagnosis: If concentration of C8 is reported as "substantially" above normal, **immediately arrange to collect a urine specimen for organic acid analysis, and order plasma acylcarnitines.**

Clinical Expectations: MCAD has variable presentation. The newborn may be asymptomatic however; the clinical phenotype can rapidly progress to hypoketotic hypoglycemia causing lethargy, vomiting and sudden death.

Additional Information:

- New England Metabolic Consortium – Emergency Protocols - www.childrenshospital.org/newenglandconsortium
- Gene Tests/Gene Clinics - www.genetests.org
- U.S. National Newborn Screening & Genetics Resource Center - www.genes-r-us.uthscsa.edu