

## Physician Action Sheet for Abnormal Newborn Screen – Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD Deficiency)

**Newborn Screen Findings:** Abnormal levels on fatty acid screen, indicating possible MCAD deficiency.

The Indiana University Newborn Screening (NBS) Lab should have already contacted your office & the infant's birthing facility about the abnormal results & recommended confirmatory testing. The IU NBS Lab has also notified the Metabolic Specialist. *Please note that the IU NBS Lab does not contact the patient's family.*

### Required Actions

1. **Contact the Metabolic Specialist (317) 274-3966 (ask for Newborn Screening Nurse or Genetic Counselor) to discuss abnormal results, follow-up recommendations & confirmatory testing.**
2. **Contact the family as soon as possible.**
  - a. Inform the family of the abnormal NBS result & the need for additional confirmatory testing.
  - b. Determine the condition of the infant. *See the infant in your office immediately if there are concerns of poor feeding, inadequate intake in breast-fed infant, vomiting, hypotonia or lethargy. These symptoms require immediate evaluation & treatment with IV glucose (D10).*
  - c. Educate the family about the need for the infant to be fed at least every four hours (including night-time feedings) while waiting on test results.
  - d. Review the list of symptoms that require immediate contact to your office (see above).
3. **Arrange for confirmatory laboratory testing at the infant's birthing facility.**
4. **Plan for follow-up with the Metabolic Specialist if MCAD deficiency is confirmed.**

**Clinical Summary:** Autosomal recessive disorder of fatty acid oxidation associated with impaired energy production. Deficiency of the MCAD enzyme results in faulty formation of ketone bodies & accumulation of partially oxidized fatty acid metabolites during periods of fasting or stress. Newborns are usually asymptomatic; however, with stress or fasting, infants & individuals of any age with MCAD may develop hypoketotic hypoglycemia, metabolic acidosis, vomiting, lethargy & sudden death. Alternate names for this condition include MCADD deficiency & ACADM deficiency.

**Treatment:** Individuals with MCAD require avoidance of fasting, avoidance of medium-chain triglyceride (MCT) oil (found in preemie formulas), a high-carbohydrate/low-fat diet & supplemental L-carnitine. Treatment is life-long & will require follow-up with Metabolic Specialist. Intercurrent illnesses associated with poor intake, vomiting or increased energy demands may require hospitalization & treatment with IV dextrose. Early diagnosis, preventative care & prompt treatment during metabolic crises will help ensure the best outcome for individuals with MCAD.

**Incidence:** 1/18,000 in Indiana (1/10,000 - 1/20,000 nationally). MCAD is more prevalent in Caucasian population. Genetic testing available.

### **Contact Information:**

- Regular business hours: (317) 274 – 3966; ask for the Newborn Screening Nurse or Genetic Counselor.
- Emergency night/weekend contact: (317) 944 – 5000; have the on-call Metabolism physician paged.

### **Resources:**

- Newborn Screening ACT sheets – [www.acmg.net](http://www.acmg.net) (select “Resources,” click on “ACT Sheets”)
- Information for clinicians – <http://www.fodsupport.org/mcad.htm>