

Physician Action Sheet for Abnormal Newborn Screen – Fatty Acid Oxidation Disorders

Newborn Screen Findings: Abnormal fatty acids on newborn screen, indicating possible fatty acid oxidation (FAO) disorder. Abnormal metabolites will vary depending on possible disorder.

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 an FAO disorder is confirmed.**

Clinical Summary: Autosomal recessive disorders of fat metabolism associated with impaired energy production. Deficiencies of specific enzymes in the fatty acid metabolic pathway result in faulty formation of ketone bodies & accumulation of partially oxidized fatty acid metabolites during periods of fasting or stress. *Indiana's newborn screening panel includes acyl-CoA & hydroxacyl-CoA dehydrogenase deficiencies (MCAD, SCAD, VLCAD; SCHAD & VLCHAD), carnitine palmitoyltransferase deficiency types I and II (CPT I & CPT II), glutaric acidemia type II, primary carnitine deficiency & others.* Newborns usually asymptomatic; however, individuals of any age with an FAO disorder may develop hypoketotic hypoglycemia, metabolic acidosis, vomiting, lethargy & sudden death with stress/fasting. Some FAO disorders associated with cardiomyopathy, hepatic dysfunction & muscle involvement.

Treatment: Individuals with FAO disorders require avoidance of fasting, other diet modifications and/or medications specific for the disorder. Treatment is life-long & requires follow-up with Metabolic Specialist. Intercurrent illnesses associated with poor intake, vomiting, fever 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 FAO disorders.

Incidence: 1/11,000 in Indiana (total incidence of all FAO disorders on Indiana's newborn screening panel). 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 (select "Resources," click on "ACT Sheets")
- Information for clinicians - http://www.fodsupport.org/medical_info.htm