

Physician Action Sheet for Abnormal Newborn Screen - 3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC Deficiency)

Newborn Screen Findings: Elevated C5-OH on newborn screen, indicating possible 3-MCC 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.*
 - 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 3-MCC deficiency is confirmed.**

Clinical Summary: Autosomal recessive disorder caused by defective activity of the enzyme 3-methylcrotonyl-CoA carboxylase (3-MCC). Deficiency of 3-MCC enzyme results in abnormal leucine metabolism. Newborns are usually asymptomatic; however, with the stress of illness, fever or fasting, individuals of any age with 3-MCC deficiency may develop hypoglycemia, metabolic acidosis, vomiting, lethargy & sudden death.

Treatment: Individuals with 3-MCC deficiency will require avoidance of fasting. Treatment is life-long & will require follow-up with Metabolic Specialist. Intercurrent illnesses associated with poor intake, vomiting or fever may precipitate metabolic crises & require hospitalization. Early diagnosis, preventative care & prompt treatment during metabolic crises will help ensure the best outcome for individuals with 3-MCC deficiency.

Incidence: 1/33,000 in Indiana (1/50,000 nationally). 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://ghr.nlm.nih.gov/condition/3-methylcrotonyl-coa-carboxylase-deficiency>