Newborn Screening ACT Sheet
[Elevated C8 with Lesser Elevations of C6 and C10 Acylcarnitine]
Medium-chain Acyl-CoA Dehydrogenase (MCAD) Deficiency

Differential Diagnosis: Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency.

Condition Description: MCAD deficiency is a fatty acid oxidation (FAO) disorder. Fatty acid oxidation occurs mainly during prolonged fasting and/or periods of increased energy demands (fever, stress), when energy production relies increasingly on fat metabolism. In an FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO enzymes.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly). If signs are present or infant is ill, transport infant to hospital for emergency treatment that would include IV glucose and any further treatment in consultation with the metabolic specialist.
- If infant is normal initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Educate family about need for infant to avoid fasting and the need for immediate medical attention if the infant even becomes mildly ill (poor feeding, vomiting, or lethargy).
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine analysis will show a characteristic pattern consistent with MCADD. Urine organic acid analysis may also show an abnormal profile. Diagnosis may be confirmed by mutation analysis of the MCAD gene.

Clinical Considerations: MCAD deficiency is usually asymptomatic in the newborn although it can present acutely in the neonate with hypoglycemia, metabolic acidosis, hyperammonemia, and hepatomegaly. MCAD deficiency is associated with high mortality unless treated promptly; milder variants exist. Hallmark features include vomiting, lethargy, and hypoketotic hypoglycemia. Untreated MCAD deficiency is a significant cause of sudden death.

Additional Information:

Emergency Treatment Protocol (New England Consortium of Metabolic Programs)
Genetics Home Reference

Referral (local, state, regional and national):

Testing
Clinical Services
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

New England Consortium of Metabolic Programs
http://newenglandconsortium.org/for-professionals/acute-illness-protocols/fatty-acid-oxidation-disorders/medium-chain-acyl-coa-dehydrogenase-deficiency-mcadd/

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=mediumchainacylcoenzymeadehydrogenasedeficiency

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/2189?db=genetests&country=United%20States

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/genetests/clinic?db=genetests

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®