KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: ELEVATED C8 WITH LESSER ELEVATIONS OF C6 AND C10 ACYLCARNITINE

CONDITION: MEDIUM-CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (MCADD)

DIFFERENTIAL DIAGNOSIS: Medium-chain acyl-CoA dehydrogenase deficiency (MCADD).

METABOLIC DESCRIPTION: MCAD deficiency is a fatty acid oxidation (FAO) disorder. FAO occurs 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.

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

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

CONFIRMATION OF DIAGNOSIS: Plasma acylcarnitine analysis will show elevated octanoylcarnitine (C8). Urine acylglycine will show elevated hexanoylglycine. Diagnosis is confirmed by mutation analysis of the MCAD gene.

CLINICAL EXPECTATIONS: 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. It is a significant cause of sudden death.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

Bryce Heese, MD
Biochemical Genetics
Children's Mercy Hospital- Kansas City, MO

Clinic phone: 816-234-3771
Hospital Operator: 816-234-3000
Office Fax: 816-302-9963