TEXAS Health and Human Services | Texas Department of State Health Services

Newborn Screening ACT Sheet

Elevated C8 with Lesser Elevations of C6 and C10 Acylcarnitine

Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

Differential Diagnosis

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD). Medium-Chain Ketoacyl-CoA Thiolase Deficiency (MCAT) (MCKAT).

Condition 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. MCKAT multiple analytes may be elevated.

Conditions associated with this analyte have been identified by the Society of Inherited Metabolic Disorders (SIMD) as critical and require immediate action.

Take the Following IMMEDIATE Actions

- **Contact family to inform them of the newborn screening result and *ascertain clinical status (poor feeding, vomiting, and lethargy);***
- **Immediate telephone consultation with pediatric metabolic specialist (See attached list);**
- **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 asymptomatic initiate timely confirmatory/diagnostic testing, as recommended by specialist;**
- ***Initial testing: plasma acylcarnitine profile, plasma carnitine levels, urine acylglycines, and urine organic acids;***
- **Repeat newborn screen if the second screen has not been done;**
- **Educate family about need for infant to avoid fasting. Even if mildly ill, immediate treatment with IV glucose is needed. FEED INFANT EVERY 2-3 HOURS AROUND THE CLOCK – SET ALARM; and**
- **Report findings to newborn screening program.**

Diagnostic Evaluation

Plasma acylcarnitine analysis will show characteristic pattern consistent with MCAD deficiency. Urine organic acid analysis may also show an abnormal profile. Diagnosis may be confirmed by mutation analysis of the MCAD gene. MCKAT is extremely rare.

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 is a significant cause of sudden death. Acylcarnitines may normalize on the second screen on affected babies, therefore an infant with an out of range first newborn screen and normal second newborn screen will still need a metabolic evaluation.

Additional Information

American College of Medical Genetics and Geonomics – C8-C6-C10 ACT Sheet
U.S. National Library of Medicine, Medline Plus – MCAD
STAR G FELSI – MCADD
STAR G FELSI – Medium-chain acyl-CoA dehydrogenase deficiency

Revised: 5/23/2024