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

Newborn Screening ACT Sheet

Elevated C14:1 +/- Other Long-chain Acylcarnitines

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)

Differential Diagnosis

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency.

Condition Description

VLCAD 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.

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

Medical Emergency: Take the Following IMMEDIATE Actions

- **Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, and lethargy);**
- **Consult with pediatric metabolic specialist (See attached list.);**
- **Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly, arrhythmia, evidence of cardiac decompensation);**
- **If signs are present or infant is ill, initiate emergency treatment with IV glucose and oxygen. Transport to hospital for further treatment in consultation with metabolic specialist;**
- **If infant is normal, initiate timely confirmatory/diagnostic testing, as recommended by specialist;**
- ***Initial testing: plasma acylcarnitine profile and plasma (free and total) carnitine levels;***
- **Repeat newborn screen if 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; and**
- **Report findings to newborn screening program.**

Diagnostic Evaluation

Plasma acylcarnitine profile may show increased acylcarnitine (and lesser elevations of other long-chain acylcarnitines). Diagnosis is confirmed in consultation with the metabolic specialist by mutation analysis of the VLCAD gene and additional biochemical genetic tests.

Clinical Considerations

VLCAD deficiency may present acutely in the neonate and is associated with high mortality unless treated promptly; milder variants exist. Features of severe VLCAD deficiency include hepatomegaly, cardiomyopathy and arrhythmias, lethargy, hypoketotic hypoglycemia, and failure to thrive. 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. Treatment is available.

Additional Information

American College of Medical Genetics and Genomics – C14 ACT Sheet
STAR G FELSI – VLCADD
Baby's First Test – Very Long-Chain Acyl-CoA Dehydrogenase Deficiency

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