



# NEWBORN SCREENING ACT SHEET

**SCREEN FOR:** ELEVATED C14:1 +/- OTHER LONG-CHAIN ACYLCARNITINES

**CONDITION:** VERY LONG CHAIN ACYL-COA DEHYDROGENASE DEFICIENCY (VLCADD)

**DIFFERENTIAL DIAGNOSIS:** Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD).

**METABOLIC DESCRIPTION:** VLCAD deficiency is a fatty acid oxidation (FAO) disorder. Fatty acid oxidation occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In a 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, 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.
- ◆ 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 profile may show increased C14:1 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 EXPECTATIONS:** 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 in infancy include hepatomegaly, cardiomyopathy and arrhythmias, lethargy, hypoketotic hypoglycemia and failure to thrive. Treatment is available.

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

## SPECIALISTS:

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**DISCLAIMER:** These standards and guidelines were adapted from the American College of Medical Genetics ACT sheets. They are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonable directed to obtaining the same results. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines