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.**
- **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.**
- **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:
National Center for Biotechnology Information
http://www.ncbi.nlm.nih.gov/books/NBK55827/bin/C14.pdf
STAR G FELSI
http://www.newbornscreening.info/Parents/fattyaciddisorders/VLCADD.html
Baby's First Test
http://www.babysfirsttest.org/newborn-screening/conditions/very-long-chain-acyl-coa-dehydrogenase-deficiency

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Newborn Screening FACT Sheet

Very Long Chain Acyl-CoA Dehydrogenase Deficiency
(VLCAD)

What is VLCAD?
VLCAD is a type of fatty acid oxidation disorder. People with VLCAD can't break down certain types of fat into energy for the body.

What Causes VLCAD?
Enzymes help start chemical reactions in the body. VLCAD happens when an enzyme called "very long chain acyl-CoA dehydrogenase" is missing or not working. This enzyme breaks down certain fats from the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with VLCAD?

[Symptoms are something out of the ordinary that a parent notices.]

There are three forms of VLCAD — infant, childhood, and adult. Symptoms can be mild or serious. Infant and childhood types of VLCAD may cause periods of illness called Metabolic Crises, or low blood sugar. Some of the first signs of a Metabolic Crisis are:

- Too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- poor appetite

If a Metabolic Crisis is not treated, a child with VLCAD can develop:

- breathing problems and seizures
- coma, sometimes leading to death

Other problems include enlarged liver, enlarged heart, and muscle problems.

What is the Treatment for VLCAD?
The following treatments are often used for children with VLCAD:

1. Do not go a long time without food – Babies and young children with VLCAD should eat often to avoid low blood sugar or a Metabolic Crisis. They shouldn't go without food for more than 4 to 6 hours. Some babies may need to eat even more often. Children with VLCAD should have a starchy snack (such as bread, cereal, rice) before bed and another during the night. They need another snack first thing in the morning. Raw cornstarch mixed with water, milk, or other drink is a good source of long-lasting energy. Your dietitian can give you ideas for good night-time snacks.

2. Diet – Sometimes your child will need to eat a diet low in fat (lean meat and low-fat dairy foods) and high in carbohydrates (such as bread, noodles, fruits, vegetables). Your dietitian will make any needed diet changes. Dietitians know right food your child should eat.

3. MCT oil and L-carnitine and other supplements – Your doctor may prescribe MCT oil. This special oil has medium chain fatty acids that can be used in small amounts for energy. Sometimes the doctor will prescribe L-carnitine. This is safe and natural and helps the body make energy.

Things to Remember
Always call your doctor when your child has any of the following:

- poor appetite
- too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- continued muscle pain or weakness
- reddish-brown color to the urine

Children with VLCAD need to eat extra starchy food (such as bread, cereal, rice) and drink more fluids during any illness. When they become sick, they often need to be treated in the hospital to prevent serious health problems.