

FACT SHEET
Healthcare Provider

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

Description:

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency is an autosomal recessive disorder of fatty acid oxidation resulting in an inability to break down long-chain fatty acids because an enzyme is either missing or not functioning correctly. Reduced carbohydrate intake as a result of a prolonged fasting state or increased energy needs from a catabolic state (infection, stress, etc.) may cause metabolic decompensation if energy needs are not sufficiently provided for by increased caloric intake.

Incidence in General Population:

1:75,000 live births

Symptoms:

Because of the inability to utilize very long-chain fatty acids for energy production, prolonged fasting (more than 4 to 6 hours), intercurrent illness, and excessive activity make the child vulnerable to acute episodes of metabolic decompensation, hypoglycemia, coma, liver dysfunction, congestive heart failure, and hepatic encephalopathy. The initial presentation may occur in the neonatal period but more often when the infant is being weaned from nighttime feeds. The usual picture is vomiting and/or lethargy after a period of fasting. This can progress to hypoglycemic seizures or coma within 1-2 hours of onset of symptoms. There may be a history of a recent viral infection associated with diminished oral intake or of a similar episode in the past. Fatty acid oxidation disorders (FAODs) are responsible for a small but significant proportion of sudden infant death syndrome, which may be preventable with prompt recognition and treatment.

Diagnosis:

Newborn screening—Tandem mass spectrometry identifies elevations in plasma long-chain acylcarnitines (C14:1, C16, C18:1).

A second dried-blood-spot filter paper card may be requested by the Newborn Screening Laboratory if the initial screening result is above the normal range. Infants with presumptive positive screening (critical) results require prompt follow up. If this occurred, the clinician would be contacted by the Metabolic Treatment Center. When notified of these results, the clinician should immediately check on the clinical status of the baby and facilitate referral to the Metabolic Treatment Center. The Metabolic Treatment Center will provide consultation and assistance with diagnostic testing.

Situations That Risk Metabolic Decompensation:

Children with VLCADD may be clinically symptomatic. Metabolic decompensation can be triggered by the catabolic processes that occur in the course of an infection, post-immunization, or with a prolonged period of fasting. Lethargy, vomiting, tachypnea, or apnea, with or without hypoglycemia are typical clinical features.

Monitoring:

Clinical observation is the most important tool for monitoring patients with known VLCADD. They should be observed for lethargy, recurrent vomiting, refusal to eat, Tachypnea, or apnea. In these situations, immediate evaluation in the emergency room is necessary. In situations of metabolic decompensation, hypoglycemia with small or no urinary ketones can develop, but a normal blood glucose level does not rule out metabolic instability and should never be a reason to delay therapy.

Cardiac Evaluation:

Any child diagnosed with VLCADD should receive an immediate referral to a pediatric cardiologist for the presence of cardiomyopathy. Regular evaluations should occur thereafter for assessment of clinical symptoms suggestive of cardiomyopathy such as tachypnea, hepatomegaly, tachycardia, feeding problems, and exercise intolerance. However, it is important to know that cardiomyopathy can be present without any clinical symptoms.

Treatment:

- Avoid fasting and prolonged exercise.
- Feed at regular intervals during the day and limit overnight fasting.
- Children should not go without food intake longer than 4 hours for the first 4 months of life, 6 hours for ages 4-8 months, and no longer than 8 hours thereafter.
- The Metabolic Treatment Center will determine the patient's diet prescription that establishes the optimum percentage of fat, carbohydrate, and protein.
- Restrict very long-chain fatty acids to 10% of total energy.
- Carnitine supplements—monitor blood levels and add carnitine supplementation only if necessary; there are concerns that long-chain acylcarnitines may induce arrhythmias.
- Supplementation with Medium Chain Triglycerides Oil (MCT) provides 10-20% of total energy.
- Increased carbohydrate intake and cornstarch therapy is necessary during acute illness. The use of cornstarch therapy is an ongoing treatment and is even more necessary during acute illness.

Illness and Immunizations:

- Any illness can potentially lead to metabolic decompensation.
- Prevention and/or early intervention is of particular importance.
- Provide high-carbohydrate feedings including cornstarch.
- Avoid dehydration.
- Closely monitor blood glucose and intake; even if blood glucose is normal, metabolic decompensation can occur.
- Immunizations must be kept current.
- All children with VLCADD should be annually vaccinated for influenza.
- There are no immunization contraindications because of VLCADD.
- Parents and physicians should be alert to the need for immediate evaluation if high fever, lethargy, or vomiting occurs within the first 24 hours.
- After an immunization with any other clinical symptoms, administration of acetaminophen or ibuprofen is warranted.

Surgical/Surgical Procedures:

- Discuss any plans for surgical and dental procedures with the Metabolic Treatment Center.
- Infants and children with VLCADD can undergo necessary anesthetic/surgical procedures.
- Any surgical procedure constitutes a potentially catabolic situation.
- Any surgery should include hospitalization preoperatively and postoperatively.
- Preoperative fasting should be avoided with 10% dextrose being started preoperatively and continuing postoperatively until the child is eating and drinking well.
- If complicated surgery or a postoperative period as an inpatient is required, the procedure should be done at a hospital with a metabolic service.

Growth and Development:

- It is crucial to closely monitor all growth parameters on a regular basis.
- In cases with neurological deficits, the child should be referred to an early intervention program and developmental progress should be closely monitored by both the metabolic team and the primary care provider.



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