

FACT SHEET
Healthcare Provider

Long -Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency (LCHADD)

Description:

Long-Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency is an autosomal recessive genetic condition caused by enzyme defects in the mitochondrial beta-oxidation cycle. Individuals with this disorder are unable to break down fatty acids into an energy source. Metabolic crisis can occur during periods of fasting and can cause episodes of hypoglycemia, vomiting, coma, and even death.

Incidence in General Population:

1:75,000 live births

Symptoms:

Metabolic crisis may be precipitated by intercurrent illnesses and may present with symptoms that include hypoglycemia, lethargy, failure to thrive, hypotonia, seizures, developmental delay, peripheral neuropathy, cardiomyopathy, coma, or sudden death. Severe, untreated cases may present as SIDS. Affected infants and children usually present by 2 years of age.

Diagnosis:

Newborn screening—Tandem mass spectrometry identifies elevations in long-chain acylcarnitines (C14-C18).

The disorders cannot be differentiated by tandem mass spectrometry methods. 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:

Metabolic decompensation can be triggered by the catabolic processes that occur in the course of infections, after an immunization, increased physical activity, or with a prolonged period of fasting. Typical clinical features are lethargy, vomiting, hypoglycemia, metabolic acidosis, and cardiac decompensation.

Monitoring:

Clinical observation is the most important tool for monitoring patients with LCHADD. They should be observed and assessed for hepatic function, neurological status, recurrent vomiting, refusal to eat, increased lethargy, apnea, or seizures. In these situations, immediate evaluation in the emergency room is necessary. In situations of metabolic decompensation, hypoglycemia can develop but normal blood glucose does not rule out metabolic instability and should never be a reason to delay therapy. It is also important for the primary care provider and the Metabolic Treatment Center to develop an on-going collaborative relationship in caring for these patients.

Treatment:

- Avoid fasting.
- Feed at regular intervals during the day and limit overnight fasting.
- 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 long-chain fatty acids to 10% of total energy
- Carnitine supplements—monitor and add carnitine only if necessary; there are concerns that long-chain acylcarnitines may induce arrhythmias in some patients.
- 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.
- If the child is vomiting or refuses to eat, the child needs to be taken to an emergency room for IV administration of at least 10% dextrose. These patients may go on to develop metabolic acidosis or hyperammonemia, in addition to severe hypoglycemia.
- Cardiac and ophthalmologic status should be reviewed on a regular basis.
- Infants and children with LCHADD should have regularly scheduled visits at the Metabolic Treatment Center.
- LCHADD chronic management is complicated, as many children take a significant amount of time (days to weeks) to improve clinically even after their biochemical parameters have normalized. Particular problems include gradual improvement in mental status, hypotonia, hepatomegaly, and cardiomyopathy. It is important to be aware that, despite therapy, children with LCHADD have died or been left with chronic neurological, cardiac, and hepatic problems.
- The parents should have an emergency protocol with them at all times. This protocol can be provided by the Metabolic Treatment Center, and it should contain basic information about the disorder, necessary diagnostic investigations, and guidelines for treatment.

Illness:

- Any illness can potentially lead to metabolic decompensation.
- Prevention and/or early intervention are of particular importance.
- Consult with the Metabolic Treatment Center within 24 hours of the onset of the illness or at the time of hospitalization.
- Provide high-carbohydrate feedings including cornstarch.
- Avoid dehydration.
- Closely monitor blood glucose and intake; even if blood glucose is normal, metabolic decompensation can occur.

Immunization:

- Immunizations must be kept current.
- All children with LCHADD should have a yearly vaccine for influenza.
- There is no contraindication to any immunization because of LCHADD.
- Parents and physicians should be alerted to the need for immediate evaluation if high fever, lethargy, or vomiting occurs in the first 24 hours.
- After an immunization without 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 LCHADD 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 anticipated, 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.

Note: A pregnant woman carrying a fetus with LCHADD is at risk for HELLP syndrome (hemolysis, elevated liver enzymes, and low platelet counts) or acute fatty liver of pregnancy.



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