

VLCAD**Very Long-Chain Acyl-CoA Dehydrogenase Deficiency****Action required**

Contact metabolic specialist today.
See infant today.

Issues to discuss with Metabolic Specialist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or metabolic clinic
 - § Plasma acylcarnitine analysis
 - 0.1 mL frozen plasma in sodium heparin green top tube
 - § Urine organic acids
 - 4.0 mL random urine, frozen
 - § Urine acylglycine
 - 5.0 mL random urine, frozen

False Positives

- Screening result can be impacted by carnitine supplementation and drug therapies containing valproic or benzoic acid

Review with family

Family has **not** been notified of result by MDH.

After discussion with metabolic specialist, contact family to coordinate clinic visit, lab work, and referral to metabolic clinic. Infant may present with symptoms when family is contacted and at clinic visit.

Prompt follow-up is critical.

NICU issues

Newborn screens cannot be accurately interpreted with carnitine supplementation, or drug therapies containing valproic or benzoic acid.

Clinical summary

VLCAD is an autosomal recessive disorder that results from the defective activity of very long-chain acyl-CoA dehydrogenase, an enzyme involved in long chain fatty acid oxidation. Newborns may present acutely in the neonatal period. If an infant is not screened and/or left untreated, symptoms begin to appear in infancy and can include hepatomegaly, cardiomyopathy, arrhythmia, lethargy, hypoketotic hypoglycemia, failure to thrive, and seizures.

Affected children require carnitine supplementation, life-long avoidance of fasting, and monitoring by both primary care and specialty providers.

Incidence of VLCAD: Rare. <1:75,000; affects all ethnic groups

Clinical expectations

If treated promptly, children with VLCAD can have satisfactory growth and development.

Affected children may need hospitalization during intercurrent illness - even if mildly ill - for treatment with IV dextrose.

Affected children should be monitored for dietary compliance and hypoglycemia.

Resources

GeneTests: www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening