

Minnesota Department of Health

LCHAD**Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency**MEDICAL FACT SHEET
POSITIVE NEWBORN SCREEN**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 certain 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

LCHAD is an autosomal recessive disorder that results from the defective activity of long chain 3-hydroxyacyl-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 early in infancy and can include hepatomegaly, cardiomyopathy, lethargy, hypoketotic hypoglycemia, lactic acidosis, hypotonia, and failure to thrive.

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

Incidence of LCHAD: Rare. ~1:100,000; more prevalent in individuals of Finnish descent (1:240 carrier rate)

Clinical expectations

If treated promptly, children with LCHAD can be expected to have satisfactory growth and development. Typically, neurologic problems occur only in children with LCHAD who have hypoglycemic crises prior to treatment.

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