

Carnitine Uptake/Transport Deficiency (CUD/CTD)

Action required

Contact metabolic specialist today.

Issues to discuss with metabolic specialist

- Laboratory evaluation of infant and mother
 - § Should testing be performed by primary care or metabolic clinic
 - § Plasma acylcarnitine analysis
 - 0.1 mL frozen plasma in sodium heparin green top tube
 - § Plasma carnitines (Free, Total, Esterified)
 - 0.4 mL frozen plasma in sodium heparin green top tube

False Positives

- Screening result can be impacted by carnitine supplementation
- Clinically unrecognized women with CUD/CTD can have an infant with a positive screen
- Poor maternal nutrition and/or vegetarian diet can impact screen

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. Expect infant to be stable when family is contacted and at clinic visit.

Prompt follow-up is important.

NICU issues

Newborn screens cannot be accurately interpreted after drug therapies such as valproic or benzoic acid and carnitine.

Clinical summary

CUD/CTD is an autosomal recessive disorder that results from the defective activity of carnitine transporters, enzymes involved in carrying carnitine into cells and maintaining carnitine levels in the body. Newborns are usually asymptomatic. If an infant is not screened and/or left untreated, symptoms may begin to appear in early infancy or childhood and can include lethargy, hypotonia, hepatomegaly, hypoglycemia, and cardiomyopathy.

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

Incidence of CUD/CTD: Rare. ~1:100,000; more prevalent in individuals of Japanese descent (1:40,000). Many affected individuals are unrecognized.

Clinical expectations

If treated promptly, children with CUD/CTD usually live healthy lives with satisfactory growth and development.

Episodes of recurrent metabolic crises can occur even with treatment. Learning disabilities or mental retardation may still occur.

Affected children should be monitored for carnitine levels.

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