

Minnesota Department of Health

Citrullinemia (CIT)**Action required**

Contact metabolic specialist today.
See infant immediately.

Issues to discuss with metabolic specialist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or metabolic clinic
 - § Plasma amino acids
 - 0.5 mL frozen plasma in a sodium heparin green top tube
 - § Urine organic acids
 - 4.0 mL random urine, frozen
 - § Plasma ammonia

False Positives

- Screening result can be impacted by administration of TPN

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

Liver disease and intervention in the NICU may impact interpretation of screening results.

Clinical summary

Citrullinemia (CIT) is an autosomal recessive urea cycle disorder that results from the defective activity of argininosuccinate synthetase, an enzyme in the urea cycle. 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 hyperammonemia, seizures, failure to thrive, lethargy, cerebral edema, and coma.

Affected children require arginine supplementation, life-long dietary restriction of protein, ammonia scavenging drugs, and monitoring by both primary care and specialty providers.

Incidence of Citrullinemia: Rare. ~1:57,000; affects all ethnic groups

Clinical expectations

If treated promptly, children with citrullinemia can be expected to have satisfactory growth and development, though long-term complications such as mental retardation may be difficult to prevent.

Episodes of hyperammonemia requiring hospital admission may still develop even with treatment.

Affected children should be monitored for dietary compliance, ammonia levels, and need for intervention.

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