

SCAD

Short-Chain Acyl-CoA Dehydrogenase Deficiency

Action required

Contact metabolic specialist today.
Check infant status today.
Arrange for laboratory evaluation.

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
 - § Urine C4 acylcarnitine
 - 3.0 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 possible 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 with carnitine supplementation or with drug therapies containing valproic or benzoic acid.

Clinical summary

SCAD is an autosomal recessive disorder that results from the defective activity of short chain acyl-CoA dehydrogenase, an enzyme involved in short chain fatty acid oxidation. Newborns are usually asymptomatic. If an infant is not screened and/or left untreated, symptoms may appear early in infancy and can include vomiting, lethargy, seizures, hypoketotic hypoglycemia, seizures, and failure to thrive.

Affected children require avoidance of fasting and monitoring by both primary care and specialty providers. Some affected children may require carnitine supplementation.

Incidence of SCAD: Rare. ~1:85,000; affects all ethnic groups

Clinical expectations

Not all children with SCAD develop symptoms.

Episodes of metabolic crises, developmental delays, muscle weakness, and other health problems may still develop even with treatment. Some children may remain asymptomatic.

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