

SBCAD**2-Methylbutyryl-CoA Dehydrogenase Deficiency****Action required**

Contact metabolic specialist today.

Issues to discuss with metabolic specialist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or metabolic clinic
 - § Plasma acylcarnitines
 - 0.1 mL frozen plasma in a sodium heparin green top tube
 - § Plasma carnitines
 - 0.5 mL frozen plasma in a sodium heparin green top tube
 - § Urine acylglycines
 - 5.0 mL random urine, frozen

False Positives

- Common
- Screening result can be impacted if an infant is on carnitine supplementation or on medications containing pivalic 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. 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 administration of TPN.

Clinical summary

Short branched chain acyl-CoA dehydrogenase (SBCAD) deficiency is an autosomal recessive disorder that results from the defective activity of 2-methylbutyryl-CoA dehydrogenase, the enzyme responsible for the breakdown of isoleucine.

Generally, most infants with SBCAD are of Hmong descent and are asymptomatic.

Infants from other ethnic groups have shown lethargy, hypoglycemia, and developmental delay.

Symptomatic children identified by newborn screening may be given carnitine and should be monitored by both primary care and specialty providers.

Incidence: ~ 1/100,000; more prevalent in Hmong individuals (1:500)

Clinical expectations

Though the clinical spectrum of SBCAD is variable, children with SBCAD can be expected to be clinically stable and have normal development.

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