

Isovaleric Acidemia (IVA)

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 acylcarnitine analysis
 - 5.0 mL random urine, frozen

False Positives

- Screening result can be impacted by drug therapies containing pivalic acid or 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

Newborn screens cannot be accurately interpreted after drug therapies containing pivalic acid or with administration of TPN.

Clinical summary

Isovaleric acidemia (IVA) is an autosomal recessive disorder that results from the defective activity of isovaleryl-CoA dehydrogenase, an enzyme involved in breaking down leucine and isovaleric acid. Newborns may present acutely in the neonatal period. If an infant is not screened and/or left untreated, symptoms may begin to appear in early infancy and can include metabolic ketoacidosis, sweaty feet odor, dehydration, hyperammonemia, ketonuria, vomiting, hypoglycemia, and failure to thrive.

Affected children require a life-long diet free of glycine, carnitine, and leucine, and monitoring by both primary care and specialty providers.

Incidence of IVA: Rare. ~1:230,000; affects all ethnic groups

Clinical expectations

If treated promptly, children with IVA can live healthy lives with normal growth and development.

Metabolic crises can still occur even with treatment. Repeated metabolic crises can lead to life-long learning problems or mental retardation.

Affected children should be monitored for dietary compliance 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