

Minnesota Department of Health

Methylmalonic Acidemia (MMA)

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 organic acids
 - 4.0 mL random urine, frozen
 - § Plasma methylmalonic acid
 - 0.5 mL frozen plasma in sodium heparin green top tube

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 administration of TPN.

Clinical summary

Methylmalonic acidemia (MMA) is an autosomal recessive disorder that results from the defective activity of methylmalonyl-CoA mutase, an enzyme involved in breaking down certain amino and fatty acids. There are a number of MMA variants. Some are vitamin B₁₂ responsive and some are non-responsive. 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, dehydration, hyperammonemia, ketonuria, vomiting, hypoglycemia, and failure to thrive.

Affected children require life-long vitamin B₁₂ (for responsive MMA), carnitine, low protein diet, and monitoring by both primary care and specialty providers.

Incidence of MMA: Rare. ~1:48,000; affects all ethnic groups

Clinical expectations

If treated promptly, children with MMA may be able to live healthy lives with normal growth and development.

Metabolic crises can still occur even with treatment. Learning problems, mental retardation, movement disorders, and kidney disease can occur even with treatment.

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

