ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

ACT Sheet

Newborn Screening ACT Sheet

[Elevated C3-DC Acylcarnitine]

Malonic Acidemia

Differential Diagnosis: None.

Condition Description: Malonic acidemia is caused by deficiency of malonyl-CoA decarboxylase, which disrupts the regulation of fatty acid synthesis and breakdown and leads to elevated malonic acid and malonylcarnitine (C3-DC). This disorder has a variable phenotype and may present during the neonatal period through adulthood.

You Should Take the Following IMMEDIATE Actions:

- Inform family of the newborn screening result.
- Ascertain clinical status (poor feeding, lethargy, seizures).
- Consult with pediatric metabolic specialist the same day.
- Evaluate the newborn (poor feeding, respiratory distress, lethargy, seizures, hypoglycemia). If any of these findings are present or if the newborn is ill, transport to a hospital for further treatment in consultation with the metabolic specialist.
- Initiate confirmatory/diagnostic testing as recommended by the specialist.
- Provide family with basic information about malonic acidemia and its management.

- Report final diagnostic outcome to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitines: C3-DC is elevated in malonic acidemia. Urine organic acids demonstrate elevated malonic acid. Molecular genetic testing can confirm the diagnosis.

Clinical Considerations: Malonic acidemia can present acutely in the neonatal period with respiratory distress, seizures, hypoglycemia, metabolic acidosis and lethargy. More commonly, malonic acidemia presents during infancy or later childhood with developmental delay, seizures, failure to thrive, hypotonia, hypoglycemia, metabolic acidosis, and cardiomyopathy. Treatment is supportive and directed at promoting normal growth and development.

Additional Information:

How to Communicate Newborn Screening Results
Emergency Protocols (New England Consortium of Metabolic Programs)
Gene Reviews
Medline Plus
Condition Information for Families- HRSA Newborn Screening Clearinghouse

Referral (local, state, regional, and national):

Find a Genetics Clinic Directory
Genetic Testing Registry

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State and Other Resources

State Newborn Screening Program

Newborn Screening Program, Utah Department of Health and Human Services
801-584-8256, newbornscreening.health.utah.gov/

Genetics/Metabolic Consultants

Pediatric Medical Genetics, University of Utah Department of Pediatrics
801-213-3599, healthcare.utah.edu/pediatrics/programs-services/genetics.php

Information for Clinicians and Families

Utah Medical Home Portal (see Newborn Disorders and Parents & Families sections)
ut.medicalhomeportal.org/newborn/malonic-acidemia

Parent/Family Support

Organic Acidemia Association
www.oaanews.org/ma.html

National Resources (with web addresses)

Additional Information

How to Communicate Newborn Screening Results
www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf

Emergency Protocols (New England Consortium of Metabolic Programs)
www.newenglandconsortium.org/methyl

Gene Reviews
www.ncbi.nlm.nih.gov/books/NBK1231/

Medline Plus
medlineplus.gov/genetics/condition/malonyl-coa-decarboxylase-deficiency/

Condition Information for Families-HRSA Newborn Screening Clearinghouse
newbornscreening.hrsa.gov/conditions/malonyl-coa-decarboxylase-deficiency

Referral (local, state, regional and national)

Find a Genetics Clinic Directory
clinics.acmg.net

Genetic Testing Registry
www.ncbi.nlm.nih.gov/gtr/