

Newborn Screening ACT Sheet [Elevated C3-DC Acylcarnitine] Malonic Aciduria

Differential Diagnosis: Malonyl-CoA decarboxylase deficiency (malonic aciduria).

Condition Description: Malonic aciduria is caused by deficiency of malonyl-CoA decarboxylase which converts intramitochondrial malonyl-CoA to acetyl-CoA. This results in an increase in malonic acid and its derivatives.

YOU SHOULD TAKE THE FOLLOWING ACTIONS IMMEDIATELY:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
 - Consultation/referral with a pediatric metabolic specialist.
 - Immediately evaluate infant; check blood glucose and urine ketones and, if elevated or infant is ill, initiate treatment as recommended by metabolic specialist and transport IMMEDIATELY to tertiary center with metabolic specialist.
 - Initiate timely confirmatory/diagnostic testing as recommended by consultant.
 - Educate family about signs, symptoms and need for urgent treatment of metabolic acidosis and hypoglycemia.
 - Report findings to newborn screening program.
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Diagnostic Evaluation: Plasma acylcarnitine analysis will confirm increased C3-DC and urine organic acid analysis will show increased malonic acid.

Clinical Considerations: Malonic aciduria may present acutely in the neonate. The presentation can include hypoglycemia, lactic acidosis, and marked lethargy. More commonly, malonic aciduria presents during infancy or later childhood with developmental delay, seizures, vomiting, failure to thrive, hypotonia, hypoglycemia, metabolic acidosis, and cardiomyopathy. Treatment may be beneficial.

Additional Information:

[Genetics Home Reference](#)
[OMIM](#)

Referral (local, state, regional and national):

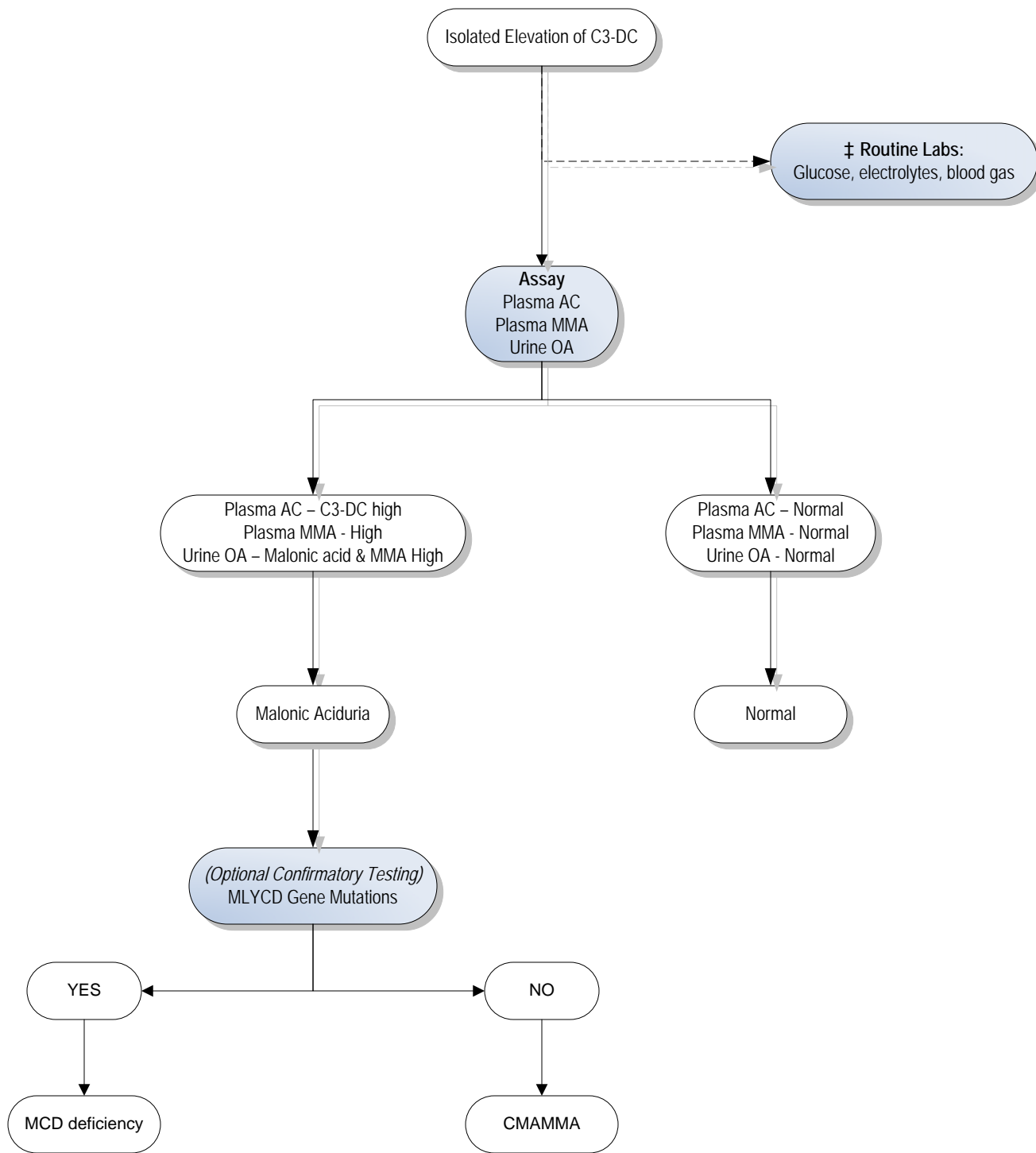
[Testing](#)
[Clinical Services](#)
[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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Elevated C3-DC (isolated)



Abbreviations:

- AC = acylcarnitines
- CMAMMA = combined malonic and methylmalonic aciduria with normal MCD activity
- DC = dicarboxylic
- MCD = malonyl-CoA decarboxylase
- MMA = methylmalonic acid
- OA = organic acids

‡ = When the positive predictive value of screening is sufficiently high and the risk to the infant is high, some initiate diagnostic studies that are locally available at the same time as confirmation of the screening result is done.

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