Elevated C3-DC (isolated)

Isolated Elevation of C3-DC

† Routine Labs:
Glucose, electrolytes, blood gas

Assay
Plasma AC
Plasma MMA
Urine OA

Plasma AC – C3-DC high
Plasma MMA - High
Urine OA – Malonic acid & MMA High

Malonic Aciduria

(Optional Confirmatory Testing)
MLYCD Gene Mutations

YES
MCD deficiency

NO
CMAMMA

Plasma AC – Normal
Plasma MMA - Normal
Urine OA - Normal

Normal

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.

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 are also 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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