Isolated C5 Elevation

Urine OA
Urine AG
Plasma AC

Plasma C5 - High
Urine IVG - High
Urine MBG - Normal
Urine PIV - Normal

Plasma C5 - High
Urine IVG - Normal
Urine MBG - Normal
Urine PIV - Normal

Plasma C5 - Normal
Urine IVG - Normal
Urine MBG - Normal
Urine PIV - Normal

Plasma C5 - High
Urine IVG - Normal
Urine MBG - High
Urine PIV - Normal

Plasma C5 - High
Urine IVG - Normal
Urine MBG - High
Urine PIV - High/Normal

Plasma C5 - High
Urine IVG - Normal
Urine MBG - Normal
Urine PIV - Normal

Plasma C5 - High
Urine IVG - Normal
Urine MBG - High
Urine PIV - Normal

Plasma C5 - High
Urine IVG - Normal
Urine MBG - High
Urine PIV - Normal

Plasma C5 - Normal
Urine IVG - Normal
Urine MBG - Normal
Urine PIV - Normal

Plasma C5 - Normal
Urine IVG - Normal
Urine MBG - High
Urine PIV - Normal

Plasma C5 - Normal
Urine IVG - Normal
Urine MBG - High
Urine PIV - Normal

Plasma C5 - Normal
Urine IVG - Normal
Urine MBG - High
Urine PIV - Normal

In vitro study of BCAA catabolism in cultured fibroblasts/lymphocytes

IVG - High
MBG - Normal

IVG - Normal
MBG - Normal

IVG - High
MBG - High

IVG - Normal
MBG - Normal

IVG - Normal
MBG - Normal

IVG - Normal
MBG - Normal

Optional Confirmatory Testing:
IVD Gene Sequencing

SBCAD def.
Ruled out IVA and SBCAD def.

False positive. No further action required

Abbreviations
AC = Acylcarnitine
AG = Acylglycine
IVA = Isovaleric acidemia
IVD = Isovaleryl-CoA dehydrogenase
IVG = Isovalerylglucine
MBG = 3-methylbutylglycine
OA = Organic acid
PIV = Pivalic acid (antibiotic)
SBCAD = short/branched chain Acyl-CoA dehydrogenase

Key
‡ = 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.

Actions are shown in shaded boxes; results are in the unshaded boxes.

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.