C4 and C5 +/-
Other Acylcarnitines Elevated

Abbreviations/Key
AC = Acylcarnitine
AG = Acylglycine
EE = Ethylmalonic encephalopathy 1
ETF = Electron transfer flavoprotein
GA2 = Glutaric aciduria Type
IVG = Isovaleryl glycine
OA = Organic acid
‡ = When the positive predictive value of screening is sufficiently high, some initiate diagnostic studies that are locally available at the same time as confirmation of the screening result is done.

Optional Confirmatory Testing:
ETF/ETF-QO Assay
ETF/ETF-QO Gene Sequencing

Optional Confirmatory Testing:
ETHE1 Gene Sequencing

Optional Confirmatory Testing:
ETF/ETF-QO Assay
ETF/ETF-QO Gene Sequencing

© American College of Medical Genetics, 2009
(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)