

Patient Name: MOCK REPORT: Positive Isovaleric AcidemiaAccession#: R10000
Birthdate: 3/2/2020
MRN#:
Specimen: Plasma
Collected: 11/1/2021
Reported: 01/21/2022**Expanded Acylcarnitine Panel by LC-MS/MS****RESULTS**

ANALYTE	REFERENCE RANGE	RESULT	FLAG
carnitine (C0)	(12.7 to 69.1)	11.192	L
deoxycarnitine (DXY)	(0.126 to 2.155)	0.466	
trimethylamine-N-oxide (TMAO)	(0.060 to 13.034)	89.047	H
trimethyllysine (TML)	(0.173 to 1.118)	0.395	
acetylcarnitine (C2)	(1.626 to 25.827)	1.907	
propionylcarnitine (C3)	(0.11 to 1.208)	0.177	
acrylylcarnitine (C3:1)	(0 to 0.001)	0.000	
isobutyrylcarnitine (C4)	(0.018 to 0.31)	0.028	
butyrylcarnitine (C4)	(0.009 to 0.302)	0.044	
tiglylcarnitine (C5:1)	(0.001 to 0.024)	0.008	
3-methylcrotonylcarnitine (C5:1)	(0 to 0.002)	0.000	
pivaloylcarnitine (C5)	(0 to 0.014)	0.000	
2-methylbutyrylcarnitine (C5)	(0.008 to 0.093)	0.000	L
isovalerylcarnitine (C5)	(0.01 to 0.161)	15.302	H
valerylcarnitine (C5)	(0 to 0.010)	0.000	
hexanoylcarnitine (C6)	(0.008 to 0.165)	0.018	
heptanoylcarnitine (C7)	(0 to 0.001)	0.000	
octenoylcarnitine (C8:1)	(0.076 to 0.622)	0.179	
octanoylcarnitine (C8)	(0.016 to 0.403)	0.032	
nonanoylcarnitine (C9)	(0.002 to 0.033)	0.009	
decadienoylcarnitine (C10:2)	(0.001 to 0.027)	0.009	
decenoylcarnitine (C10:1)	(0.026 to 0.311)	0.061	
decanoylcarnitine (C10)	(0.012 to 0.631)	0.043	
dodecenoylcarnitine (C12:1)	(0.004 to 0.234)	0.039	
dodecanoylcarnitine (C12)	(0.002 to 0.175)	0.023	
tetradecadienoylcarnitine (C14:2)	(0.004 to 0.352)	0.038	
tetradecenoylcarnitine (C14:1)	(0.012 to 0.451)	0.042	
tetradecanoylcarnitine (C14)	(0.003 to 0.116)	0.018	
hexadecenoylcarnitine (C16:1)	(0.006 to 0.127)	0.023	

hexacecanoylcarnitine (C16)	(0.020 to 0.253)	0.081	
heptadecanoylcarnitine (C17)	(0 to 0.003)	0.007	H
octadecadienoylcarnitine (C18:2)	(0.025 to 0.275)	0.141	
cis-9-octadecenoylcarnitine (C18:1)	(0.029 to 0.346)	0.107	
octadecanoylcarnitine (C18)	(0.010 to 0.097)	0.026	
hydroxy(iso)butyrylcarnitine (C4:OH)	(0.010 to 0.644)	0.054	
3-hydroxyisovalerylcarnitine (C5:OH)	(0.004 to 0.101)	0.007	
pimelyl/hydroxyoctanoylcarnitine (C7:DC / C8:OH)	(0.001 to 0.014)	0.000	
3-hydroxydecanoylcarnitine (C10:OH)	(0.003 to 0.079)	0.012	
hydroxydodecenoylcarnitine (C12:1:OH)	(0 to 0.020)	0.003	
hydroxydodecanoylcarnitine (C12:OH)	(0.002 to 0.043)	0.005	
hydroxytetradecanoylcarnitine (C14:OH)	(0 to 0.017)	0.007	
hydroxyhexadecenoylcarnitine (C16:1:OH)	(0 to 0.014)	0.006	
3-hydroxyhexadecanoylcarnitine (C16:OH)	(0 to 0.033)	0.000	
hydroxyoctadecenoylcarnitine (C18:1:OH)	(0 to 0.032)	0.000	
hydroxyoctadecanoylcarnitine (C18:OH)	(0 to 0.007)	0.000	
malonylcarnitine (C3:DC)	(0 to 0.025)	0.000	
fumarylcarnitine (C4:1:DC)	(0 to 0.001)	0.000	
methylmalonylcarnitine (C4:DC)	(0.006 to 0.048)	0.017	
succinylcarnitine (C4:DC)	(0.007 to 0.057)	0.015	
glutaconylcarnitine (C5:1:DC)	(0 to 0.001)	0.000	
glutaryl carnitine (C5:DC)	(0.006 to 0.075)	0.015	
methylglutaconylcarnitine (C6:1:DC)	(0 to 0.015)	0.000	
adipoylcarnitine (C6:DC)	(0.002 to 0.204)	0.019	
3-methylglutaryl carnitine (C6:DC)	(0.003 to 0.084)	0.000	L
suberoylcarnitine (C8:DC)	(0.002 to 0.257)	0.011	
sebacoylcarnitine (C10:DC)	(0 to 0.036)	0.002	
hexadecanedioylcarnitine (C16:DC)	(0 to 0.050)	0.006	
octadecanedioylcarnitine (C18:1:DC)	(0 to 0.041)	0.007	
octadecanedioylcarnitine (C18:DC)	(0 to 0.028)	0.008	
eicosanedioylcarnitine (C20:DC)	(0 to 0.004)	0.005	H
docosanedioylcarnitine (C22:DC)	(0 to 0.002)	0.000	
C3/C2 (RATIO)	(0 to 0.150)	0.093	
C8/C10 (RATIO)	(0 to 2)	0.752	
C14:1/C12:1 (RATIO)	(0 to 3)	1.076	
C0/(C16+C18) (RATIO)	(48 to 1,586)	104.601	
(C16+C18:1)/C2 (RATIO)	(0 to 0.150)	0.099	
TML/DXY (RATIO)	(0 to 3.3)	0.848	

*values in micromoles/L

INTERPRETATION

MOCK report illustrating utility of acylcarnitine isomer separation. C5 elevations are shown to be caused by the C5 isomer isovalerylcarnitine thus providing confident diagnosis of isovaleric acidemia.

ASSAY INFORMATION**Method**

Analysis is performed by liquid chromatography tandem mass spectrometry (LC-MS/MS) on underivatized specimens.

For more information visit: <https://medicine.iu.edu/iubgl>

Limitations/Disclaimer

False negative results can occur in rare situations when diet, treatment or secondary carnitine depletion causes acylcarnitine levels to appear normal in an affected individual.

This test was developed and its performance characteristics determined by Indiana University Biochemical Genetics Laboratory. It has not been cleared or approved by the U.S. Food and Drug Administration. This test is used for clinical purposes. It should not be regarded as investigational or for research. The laboratory is certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA '88) as qualified to perform high complexity clinical laboratory testing. CLIA# 15D0647198 • CAP# 1678930

ELECTRONICALLY SIGNED BY

Marcus J. Miller PhD FACMG, Director of the IU Biochemical Genetics Laboratory, 01/21/2022