

Acylcarnitine Plasma Test Information

Carnitine and its fatty acyl esters (acylcarnitines) are essential compounds involved in fatty acid metabolism. Acylcarnitines are produced by the action of carnitine acyltransferases and have various chain lengths depending on their cellular location and metabolic purposes. Acylcarnitines are responsible for the transportation of long-chain fatty acids (LC-FA) into the mitochondria via the Carnitine Shuttle, and elevated acylcarnitine concentrations in human whole blood or plasma occur in response to a block in the metabolic pathway of either fatty acids or branched-chain amino acids. Hence, acylcarnitine profile analysis (ACP) is a useful clinical screening tool for the detection of a number of deficiencies in β -oxidation of fatty acids.

Tandem mass spectrometry (MS/MS) is the gold-standard method for ACP. Acylcarnitines are extracted by organic solvents (either Acetonitrile or Methanol) and then derivatized into their butyl ester forms. These derivatives are analyzed predominantly by flow-injection and parent ion scan analysis for the product ion of $m/z=85$, which is the butyl group common to all acylcarnitines. Defects in fatty acid β -oxidation are usually determined by pattern recognition of abnormal acylcarnitines profiles between normal and suspected patients.

Our lab offers a sensitive and accurate MS/MS assay for plasma acylcarnitines analysis. The results are reported to and interpreted by American Board of Medical Genetics and Genomics (ABMGG)-certified biochemical geneticists in the Division of Human Genetics at Cincinnati Children's Hospital Medical Center (CCHMC). For more information, call the lab at 513-636-4203.

Sample Type:

Plasma (Dark green – sodium heparin)

Volume:

1.5 mL

0.5 mL (minimum)

Specimen Preparation:

Spin/pour off, freeze ASAP.

Unacceptable Specimens:

Ambient samples, red cells when pouring off sample, or grossly-hemolyzed specimens.

Stability:

Refrigerated: 12 hours

Frozen: 1 month

Methodology:

Tandem mass spectrometry (MS/MS)

Reporting Units:

Quantitative: $\mu\text{mol/L}$

Reference Interval:

See report

Shipping Conditions:

Frozen (dry ice), next day.

Testing Schedule:

Once per week (for testing outside this schedule, please call 513-636-4203). **Turnaround time** is typically 7 – 10 days.

CPT Codes:

82017

Contact Information:

Clinical Mass Spectrometry

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Shipping Address:

Clinical Mass Spectrometry Facility, MLC 7019

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Cincinnati Children's Hospital Medical Center

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Cincinnati, Ohio 45229-3039

References:

1. Rinaldo P. et al. Inborn errors of amino acid, organic acid, and fatty acid metabolism. In: Burtis CA, Ashwood ER, Bruns DE, editors. Tietz textbook of clinical chemistry and molecular diagnostics, 4th ed. Philadelphia: WB Saunders, 2005: 2207-2247.
2. Chace DH. et al. A biochemical perspective on the use of tandem mass spectrometry for newborn screening and clinical testing. Clin. Biochem. 2005 (38) 2996-309.