Acylcarnitine Dried Blood Spots (DBS) 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 vary in 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 acylcarnitines 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 to 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.

While acylcarnitines are usually analyzed in plasma for general genetic testing, newborn screening programs use dried blood spots (DBS) as the standard specimen for testing due to easy sample collection, transportation, and storage.

Our lab offers a sensitive and accurate MS/MS assay for DBS 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:
Dried blood spots on Whatman 903 filter paper

Volume:
One full spot (minimum)

Specimen Preparation:
Completely fill one or more spots on Whatman 903 filter paper; allow blood to air dry for 3 hours before transporting. Refrigerate.

Unacceptable Specimens:
Samples transported before blood is dry; water on blood spots.

Stability:
Ambient: 1 week
Refrigerated: 2 weeks
Frozen: 1 month

Methodology:
Tandem mass spectrometry (MS/MS)

Reporting Units:
Quantitative: mcmol/L

Reference Interval:
See report

Shipping Conditions:
Refrigerated (cold pack), 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 Code:
82017

Contact Information:
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Shipping Address:
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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.