Acylcarnitine Profile, Quantitative and Qualitative, Plasma

Test Code: AR  
Turnaround time: 7 days  (STAT: 1 day)  
CPT Codes: 82017 x1

Condition Description

Plasma acylcarnitine profile by tandem mass spectrometer provides quantitative evaluations of individual acylcarnitine species in the plasma. Different patterns of the plasma acylcarnitine profile can indicate the diagnosis of fatty acid oxidation disorders (MCAD, VLCAD, SCAD, MAD, LCHAD, and CPTII), as well as some organic acidemias (propionic acidemia, methylmalonic acidemia, isovaleric acidemia, glutaric acidemia type I, 3-methylcrotonyl CoA carboxylase deficiency, B-ketothiolase deficiency, etc). If the above diseases are suspected, this analysis is recommended in conjunction with plasma amino acid analysis and urinary organic acid analysis. Plasma acylcarnitine values are compared to age-matched normal values.

Indications

This test is indicated for:

- Infants with a positive newborn screening result indicative of a metabolic disorder.
- Evaluation of patients with signs of a possible metabolic condition, such as lethargy, vomiting, and failure to thrive.
- Monitoring for individuals diagnosed with a metabolic condition.

Methodology

Electrospray Tandem Mass Spectrometry (MS/MS).

Detection

Test results should be interpreted in light of the patient's clinical and nutritional status.

Reference Range

Please [click here](https://www.eglgenetics.com) for reference range.

Specimen Requirements

Type: Plasma

Specimen Requirements:

In sodium heparin (green top) tube: 1-2 ml

Sample should be collected while fasting or 2-4 hours postprandial.

Centrifuge to separate plasma and freeze.

Specimen Collection and Shipping: Ship frozen sample on dry ice with overnight delivery.

Special Instructions

Please indicate any medication or dietary changes on the test requisition.

Related Tests

- [Carnitine Concentration (CN) - Plasma](https://www.eglgenetics.com)
- [Organic Acid Analysis (OA) - Urine](https://www.eglgenetics.com)

Note: These tests are used in the diagnosis and evaluation of patients with organic acid or fatty acid disorders.