Conditional Description

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

Indications

This test is indicated in the case of:

- Patients experiencing a metabolic crisis.
- Evaluation of patients with signs and symptoms of a possible metabolic condition, such as lethargy, vomiting, and failure to thrive.
- Monitoring known metabolic patients who have been hospitalized and for which a rapid analysis is essential.
- Infants with a positive newborn screening result indicative of a metabolic disorder.

Methodology

Electrospray Tandem Mass Spectrometry (MS/MS).

Detection

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

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 medications or dietary changes on the test requisition form.

STAT TESTING MUST BE COORDINATED WITH AND PREAPPROVED BY ONE OF THE DIRECTORS IN THE BIOCHEMICAL GENETICS LABORATORY. Please call 855-831-7447.

Related Tests

- STAT Organic Acids Quantitative Analysis (BOAST)
- STAT Amino Acids Analysis, Plasma (BAAST)