Metabolic Disease: Panel (AA, AR, CN, OA)

**Condition Description**

This panel is a comprehensive evaluation when an unspecified metabolic condition is suspected but not yet confirmed in a patient. The panel includes the following four tests:

**Urine Organic Acid Analysis:** Symptoms of lethargy, coma, hypotonia, seizures, ataxia, vomiting, failure to thrive, metabolic acidosis, etc., may suggest abnormalities of organic acid metabolism. Qualitative and quantitative determination of organic acids is performed by gas chromatography/mass spectrometry. This includes lactic and pyruvic acids.

**Plasma Acylcarnitine Profile:** Plasma acylcarnitine analysis by tandem mass spectrometry 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, as well as some organic acidemias. Plasma acylcarnitine values are compared to age-matched normal values.

**Free and Total Carnitine Concentration:** Carnitine deficiency can be from a primary defect or can be a secondary effect. Primary carnitine deficiency can be the result of several metabolic conditions and can present with hypoketotic hypoglycemia, encephalopathy, hepatomegaly, cardiomyopathy, muscle weakness, or gastrointestinal dysmotility.

Secondary carnitine deficiency results from another metabolic disorder, such as another fatty acid oxidation disorder, or an organic acidemia leading to carnitine depletion secondary to the formation of acylcarnitines for excretion of accumulating byproducts. Secondary carnitine deficiency can present with symptoms of encephalopathy, hypotonia, hepatomegaly, cardiac hypertrophy, failure to thrive, hypoglycemia, ketoacidosis, and hyperammonemia.

**Plasma Amino Acid Analysis:** Plasma amino acid analysis will detect specific amino acid disorders such as phenylketonuria (PKU), maple syrup urine disease (MSUD), urea cycle defects, non-ketotic hyperglycinemia and homocystinuria.

**Indications**

This panel is offered as a comprehensive starting point for patients who present with symptoms of a metabolic disease, such as lethargy, vomiting, and failure to thrive and includes: plasma amino acids, plasma acylcarnitine, plasma carnitine, and urine organic acids.

**Methodology**

- Plasma amino acids: Ion Exchange Chromatography for quantitative plasma and urine amino acids analysis. LC/MS/MS for selective amino acids analysis on filter paper.
- Plasma acylcarnitine: Electrospray Tandem Mass Spectrometry (MS/MS).
- Plasma carnitine: Electrospray Tandem Mass Spectrometry (MS/MS).
- Urine organic acids: Qualitative and quantitative determination of organic acids is performed by gas chromatography/mass spectrometry. This includes lactic and pyruvic acids.

**Reference Range**

- Plasma amino acids: Click here for reference range.
- Plasma acylcarnitine: Click here for reference range.
- Plasma carnitine: Click here for reference range.
- Urine organic acids: Click here for reference range.

**Specimen Requirements**

Additional Specimen Collection/Handling Instructions Required for this Test

For diagnostic purposes, urine should ideally be collected during time of acute illness as abnormal metabolite levels may decrease, sometimes to near normal concentrations, when patient is well.

Submit both of the following specimens

**Type: Plasma**

Specimen Requirements:

In sodium heparin (green top) tube: 2-5 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.

**Type: Urine**

Specimen Requirements:

In a clean container without preservatives: 2-5 ml. Freeze.

Fasting or first void sample is preferable.

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

### Special Instructions

**Both plasma AND urine are required for this panel. Please indicate on the test requisition form any medications or dietary changes.**

### Related Tests

- The components of this panel can be ordered separately when a specific metabolic condition is suspected.
- Plasma acylcarnitine concentration, carnitine concentration & urine organic acid analysis (OA) are used in the diagnosis and evaluation of patients with organic acid or fatty acid disorders.
- Plasma amino acids and urine organic acid analysis (OA) are used in the diagnosis and evaluation of patients with aminoacidopathies.