






Amino Acid Profile

	Test Code	B2020
	Test Summary	
	Turn-Around-Time (TAT)*	3 days
	Acceptable Sample Types	Dried Blood Spots
	Acceptable Billing Types	Self (patient) Payment Institutional Billing

Indications for Testing

Amino acid profile analysis is indicated when there is a clinical concern for a metabolic condition or in follow up from a positive newborn screen. Signs and symptoms of a metabolic disorder include vomiting, lethargy and failure to thrive. Amino acid profiles can detect amino acid disorders such as PKU, tyrosinemias, maple syrup urine disease, urea cycle disorders, organic acidemias and homocystinuria. To aid in diagnosis, it may be indicated to run an amino acid profile with other metabolic profiles such as acylcarnitine and urine organic acids.

Test Description

An amino acid biochemical profile is performed for detection of amino acid disorders. Other observations made by this assay include hyperalimentation, liver disease, medium chain triglyceride oil administration, presence of EDTA anticoagulants in blood specimen, treatment with benzoate, pyrovalic acid or valproic acid and carnitine update deficiency.

Condition Description

Amino acid metabolism disorders are inherited metabolic disorders that result from defects either in the breakdown of amino acids or in the body's ability to get amino acids into cells. In the United States, newborns are routinely screened for several amino acid disorders. Early diagnosis of these disorders is important to foster a change in medical management to improve outcomes for individuals with these disorders.

Test Methods and Limitations

This test is performed by tandem mass spectrometry. Tandem mass spectrometry (MS/MS) is an analysis technique that utilizes two mass analyzers to target analytes based on mass and charge. This mass filtering process provides highly sensitive and specific measurements of small molecules or peptides from biological samples.

Detailed Sample Requirements

Dried Blood Spots