

# THE AGA KHAN UNIVERSITY HOSPITAL CLINICAL LABORATORIES

## UPDATE

### Dried Blood Spot Metabolic Profile (Amino Acids, Acylcarnitines and Succinylacetone)

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#### INTRODUCTION:

This profile provides semi-quantitative detection of the amino acids, succinylacetone and acyl carnitines. Amino acids are the building blocks of proteins. In order to metabolize the amino acids, special proteins called enzymes are needed. A change or mutation in the gene coding for an enzyme, results in the accumulation or the deficiency of one or more amino acids, which leads to the clinical signs and symptoms of the particular amino acid disorder. Acylcarnitines are responsible for the transportation of long-chain fatty acids into the mitochondria via the carnitine shuttle and their elevated concentrations occur in response to a block in the metabolic pathway of either fatty acids or branched-chain amino acids, thus making it a useful clinical screening tool for the detection of a number of deficiencies in  $\beta$ -oxidation of fatty acids. Succinylacetone is a specific marker for Tyrosinemia type 1, an autosomal recessive condition caused by a deficiency of the enzyme fumarylacetoacetate hydrolase (FAH). The profile test is indicated as a screening method for the early detection of disorders of amino acid, fatty acid metabolism and organic acidemias in newborns. Report will be provided in the form of interpretative comments based on pattern recognition of the amino acids, acyl carnitines and succinylacetone. For confirmation, further diagnostic evaluation may be required.

#### PRINCIPLE:

Liquid chromatography- tandem mass spectrometry (LC-MS/MS)

#### SPECIMEN COLLECTION:

Specimen Type: Dried blood spot filter paper (DBS)

#### Specimen Collection:

- Blood sample is collected from the baby's pricked heel on dried blood spot filter paper (DBS) by trained phlebotomists/nurses.
- The blood spot should be air-dried for at least 3 hours.
- Once dry, the sample should be sent to the laboratory within 24 hours in a separate paper envelope, along with humidity indicator card and desiccant pouch to protect against moisture.

Results: Interpretative report will be provided.

SCHEDULE: Test will be performed twice a month according to the following schedule:

- i) Second Thursday of the month and will be reported on following Monday.
- ii) Fourth Thursday of the month and will be reported on following Monday.  
(Cut-off: Thursday morning 8:00 am)

Note: For critical patients requiring immediate results contact the biochemical genetics laboratory.

**PLEASE FILE FOR QUICK REFERENCE**

