THE AGA KHAN UNIVERSITY HOSPITAL
CLINICAL LABORATORIES

UPDATE
RAPID QUANTITATIVE ANALYSIS OF PLASMA BRANCHED-CHAIN AMINO ACIDS FOR MAPLE SYRUP URINE DISEASE

VOL. XX1, NO. 11, 2014

INTRODUCTION:
Maple Syrup Urine Disease (MSUD) is an inherited metabolic disorder caused by a deficiency of the branched-chain alpha-keto acid dehydrogenase complex leading to a buildup of the branched-chain amino acids (leucine, isoleucine and valine) and their toxic by-products (ketoacids) in the blood and urine. Infants usually present with sweet-smelling urine, with an odor similar to that of maple syrup. Quantitative amino acid analysis of these three branched-chain amino acids is crucial for the diagnosis and monitoring of MSUD.

As compared to 22 plasma amino acid quantification, this shortened program allows the separation and quantification of the four branched chain amino acids using norleucine as the internal standard. In addition to this, alloisoleucine is also identified and used as a specific and sensitive diagnostic marker. This alloisoleucine is derived from the amino acid leucine in various variants of MSUD except in E3-Deficiency with lactic acidosis, in which alloisoleucine levels are low or absent.

PRINCIPLE:
The test is performed by Cation-Exchange HPLC (Biochrom 30+).

SPECIMEN COLLECTION:
- Fasting sample is required, at least 2 hours in infants, 4 hours in children and 8 hours in adults.
- In adults 3-4 ml and in children 1-2 ml of blood, (Lithium heparin containing tube, green top) is required.
- Minimum acceptable volume of separated plasma is 0.2 ml (200 µl).
- Sample should be kept at room temperature for 4 hours. If there is delay than centrifuge and separate the plasma immediately into a clean plain container and store at -20° C.
- Transport specimen in dry ice to the laboratory.

UNACCEPTABLE CONDITIONS: Hemolyzed specimen.

SCHEDULE: Reporting will be 7 days after receiving the sample.

NOTE:
- It is essential to fill in the request form related to inherited metabolic disease provided at the reception of AKU Clinical Laboratory, Collection Points and Consulting Clinics.
- Instruct patient's attendant (parents or guardians) to provide previous reports related to inherited metabolic disease if available.
- Encourage the patients to get their test charged against the medical record number each time so that a laboratory record related to patient can be readily available.

PLEASE FILE FOR QUICK REFERENCE

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