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THE AGA KHAN UNIVERSITY HOSPITAL CLINICAL LABORATORIES

UPDATE
CSF AMINO ACID (QUANTITATIVE)

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INTRODUCTION:
Quantitative amino acid analysis in CSF is critical to the differential diagnosis of neonatal onset seizures. Patient often present with life-threatening illness between 12 and 72 hours of age. The test is used to confirm the diagnosis of disease related to amino acid metabolism such as non-ketotic-hyperglycinaemia (NKH) and serine deficiency disorder.

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

SPECIMEN COLLECTION:
- 0.5 ml (500 µl) CSF, clear in sterile container (e.g. microtubes)1 and 2 without any preservative.
- **Minimum acceptable volume is 0.2 ml (200 µl).**
- Ideally CSF should be collected at a time when symptoms are most severe
- Send together with 2 ml whole blood lithium heparinized tube for plasma amino acid if suspecting non-ketotic-hyperglycinaemia (NKH), and serine deficiency disorders. Separate the plasma immediately within 4 hours.
- Transport sample frozen in dry ice to the laboratory.

UNACCEPTABLE CONDITIONS: Blood stained CSF.

SPECIAL PRECAUTION: Biohazard specimen; to be handled with care.

SCHEDULE:
Reporting will be 10 days after receiving the sample.

NOTE:
- It is essential to fill in the request form related to inborn error of metabolism (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 initial laboratory number each time so that a laboratory record related to patient can be readily available.

PLEASE FILE FOR QUICK REFERENCE