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THE AGA KHAN UNIVERSITY HOSPITAL CLINICAL LABORATORIES

UPDATE
URINE SUCCINYLACETONE

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INTRODUCTION:
The tyrosine metabolite succinylacetone is produced exclusively in individuals with tyrosinemia type 1 due to inherited deficiency of enzyme fumarylacetoacetate hydrolase (FAH), the final enzyme in tyrosine catabolic pathway. Detection of succinylacetone in urine is critical for the diagnosis of tyrosinemia type 1 in symptomatic infants with jaundice and liver failure or following positive results on newborn screening for tyrosinemia.

PRINCIPLE:
The test is performed by selected ion monitoring gas chromatography mass spectrometry (SIM-GC/MS).

SPECIMEN COLLECTION:
- Collect 2-5 ml random urine specimen in a sterilized tube, bottle or container with no preservative
- **Minimum acceptable volume of urine is 0.5 ml (500µl)**
- Freeze sample (-20°C) if it cannot be sent to the laboratory within 4 hours.
- Transport sample frozen in dry ice to prevent bacterial overgrowth and loss of volatile substances. Succinylacetone is photosensitive; therefore protect the specimen from light by wrapping it with dark plastic bag.

UNACCEPTABLE CONDITIONS: Unfrozen Sample >24 hrs.

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

SCHEDULE:
Test will be performed on every Wednesday, reporting will be on next Wednesday.

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.