SCREENING OF INBORN ERRORS OF METABOLISM AMONG EGYPTIAN PEDIATRIC INTENSIVE CARE UNIT (PICU) PATIENTS BY TANDEM MASS SPECTROMETRY

Elkaffas R., ElGayar D., Halawa E., Sheta M.

ABSTRACT

Background: Inborn errors of metabolism (IEM) are not an uncommon entity presenting to the pediatric intensive care unit (PICU). Several of these diseases constitute acute emergencies amenable to life-saving therapy. An earlier recognition of these IEM has the potential to reduce morbidity and mortality rates among children in PICU. Expanded metabolic screening by Tandem Mass Spectrometry (MS/MS) using dried blood spot technique proved to be an efficient and rapid method in early detection of these cases.

Objective*:* This cross sectional study aims to detect the incidence of IEM disorders detectable by MS/MS among critically ill Egyptian infants and children admitted to the PICU at Cairo University Children Hospital (CUCH) for early detection, proper treatment and prevention of devastating neurological outcomes through reviewing their clinical, biochemical and neuroradiological data. Thus, highlighting the importance of the application of newborn screening programs by MS/MS in Egypt.

***Subjects & Methods*:** Forty critically ill patients with metabolic presentations admitted to Cairo University Children Hospital PICU, were selected from a total of 297 PICU admissions in the period between May 2010 to October 2010 and underwent metabolic screening by MS/MS for detection of inborn errors of aminoacids, organic acids and fatty acids metabolism. Dried blood spots (DBS) were extracted into a methanol solution with deuterium-labeled internal standards, derivatized with butanolic-HCI then submitted to MS/MS analysis and urine samples were collected for the need of confirmatory tests. All cases had been subjected to full history taking, clinical examination, routine and metabolic screening investigations**.**

Results*:* Among the 40 selected cases, MS/MS detected IEM in 15% (6/40) of the cases. While, among all PICU admissions during the study period, the incidence was 2% (6/297). These Six cases were, one case Tyrosinemia type I, two` cases MSUD, one case Propionic acidemia, one case CPT1 deficiency and one case Glutaric aciduria type II. Metabolic acidosis was the commonest finding (80%), hyperammonemia was found in 9 cases (22.5%) and hypoglycemia in 10 cases (25%). Neurological deficits in the form of seizures (45%), encephalopathy with disturbed conscious level and need for mechanical ventilation (35%) were the dominant presentation among the study group.

Conclusion*:* IEM is not uncommon in PICU, thus, screening of the PICU cases presenting with a picture suggestive of IEM by MS/MS using dried blood spot technique helps in early and effective therapeutic intervention to prevent devastating outcomes. Expanded metabolic screening by MS/MS is a rapid and efficient method in the early detection of IEM in PICU, however, confirmatory tests such as GC/MS profiling are mandatory particularly for clinically suspected cases with normal MS/MS profile.

***Keywords*:** Tandem Mass Spectrometry, Inborn errors of metabolism, Pediatric intensive care unit.