Acta Scientific PAEDIATRICS (ISSN: 2581-883X)

Volume 5 Issue 9 September 2022

Editorial

Inborn Errors of Metabolism- Role of Tandem Mass Spectrometry in Newborn Screening

MP Narayanan*

Senior Resident, Department of Biochemistry, All India Institute of Medical Sciences (AIIMS), Rishikesh, Uttarakhand, India

***Corresponding Author:** MP Narayanan, Senior Resident, Department of Biochemistry, All India Institute of Medical Sciences (AIIMS), Rishikesh, Uttarakhand, India.

Received: August 02, 2022

Published: August 04, 2022

Inborn errors of metabolism (IEM) are genetic defects involving disorders of enzyme activities in which the toxic substances accumulates and causes neurologic manifestations. This may lead to death or permanent disability of the child. Newborn screening (NBS) is the detection and early intervention is pre-symptomatic newborns with inborn errors of metabolism to prevent adverse outcomes of the disorder. NBS is done by analysis of biomarkers in blood by a spectrum of analytical methods. The introduction of tandem mass spectrometry for newborn screening is a technological breakthrough which is used across the globe nowadays. The power of NBS is enormously enhanced by tandem mass spectrometry (MS/MS) by the ability to analyze several metabolites simultaneously in the same dried blood spot (DBS) with increased sensitivity and specificity. NBS programs tend to focus on three groups of metabolites: amino acids fatty acid oxidation intermediates and short chain organic acids.

Tandem mass spectrometry (MS/MS) measures the ratio of the mass (m) of a chemical to its charge (z). The ionization techniques like electrospray (ESI) and matrix assisted laser desorption/ionization (MALDI) has revolutionized the application of MS to detect any biological molecule. The second tier testing and advances in proteomics further expanded the scope of MS in diagnosis of IEM. For NBS a tandem mass spectrometer is configured to measure amino acid and acyl carnitines using the information about their mass and fragmentation pattern. Though, the interpretation of these profiles may become complex. In MS/MS ratio of metabolites used to define whether an increased value is due to metabolic derangements or to the clinical and nutritional states of the newborn. Assessment of IEM at the appropriate time after birth is crucial for preventing morbidity and death of the infant [1-4].

The spectrum of inborn errors screened varies in different regions of the world according to their prevalence. It is important to confirm the MS/MS diagnosis by molecular genetic testing and to provide appropriate clinical management of the IEM so that the benefit of early diagnosis can be maximized. Screening centers and special clinics should be established for timely counseling, therapy and rehabilitation of the affected child before starting the NBS.

Bibliography

1. Ma Shujun., *et al*. "Expanded newborn screening for inborn errors of metabolism by tandem mass spectrometry in newborns from Xinxiang city in China". *Journal of Clinical Laboratory Analysis* 34.5 (2020): e23159.

2. De Hora., *et al*. "Implementing steroid profiling by liquid chromatography-tandem mass spectrometry improves newborn screening for congenital adrenal hyperplasia in New Zealand". *Clinical Endocrinology* 94.6 (2021): 904-912.

3. Yang N., *et al*. "Inborn errors of metabolism detectable by tandem mass spectrometry in Beijing". *Journal of Pediatric Endocrinology and Metabolism* 33.5 (2020): 639-645.

4. Vaidyanathan Kannan and Sandhya Gopalakrishnan. "Role of Tandem Mass Spectrometry in Diagnosis and Management of Inborn Errors of Metabolism". *Advanced Studies in Experimental and Clinical Medicine.* Apple Academic Press (2021): 135-155.

Volume 5 Issue 9 September 2022