INBORN ERRORS OF METABOLISM - II

NUTRITIONAL MANAGEMENT IN INBORN ERRORS OF METABOLISM - PRINCIPLES AND PRACTICAL PITFALLS

**** Sanjeeva GN***

Abstract: *Inborn Errors of Metabolism constitute a heterogeneous group of genetic disorders, resulting from an enzyme defect in biochemical and metabolic pathways affecting proteins, fats, carbohydrates metabolism or impaired organelle function presenting as complicated medical conditions involving several human organ systems. Their successful long-term clinical outcome is fundamentally dependent upon prompt and precise nutritional intervention. In majority of inborn errors of metabolism, the key therapeutic strategy is nutritional management which primarily is structured around the restriction of toxic substrates, supplementation of deficient products and strict prevention of catabolism. This comprehensive review systematically delineates the key principles of nutritional management, emphasizing the necessity of highly individualized, dynamic and rigorously monitored dietary regimens. Specific emphasis is placed on the tailored nutritional strategies and prevalent practical challenges associated with Maple Syrup Urine Disease, Organic Acidemias, Fatty Acid Oxidation Disorders, Glycogen Storage Diseases, Galactosemia and Urea Cycle Defects. Furthermore, this article articulates critical operational pitfalls pertaining to the reliable procurement of specialized medical foods, accurate biochemical monitoring and ensuring patient adherence in resource-constrained environments. Consequently, an anticipatory and highly coordinated multidisciplinary team approach, incorporating the expertise of the pediatrician, metabolic specialist and specialized dietitian, is deemed indispensable for optimizing somatic growth, neurodevelopmental outcomes and the overall quality of life for affected pediatric populations.*

Keywords: *Inborn errors of metabolism, Nutritional management, Acute metabolic crisis, Specialized medical foods.*

Points to Remember

- ***Acute illness necessitates the immediate cessation of the restricted substrate and aggressive high-dose carbohydrate administration to prevent catabolism and neurotoxicity.***
- ***Monitoring is dynamic and must prioritize tracking the specific toxic metabolite while urgently checking urine ketones for signs of catabolism during illness.***
- ***For FAOD and GSD, the primary therapy is the absolute prevention of fasting, often maintained via strict nocturnal slow-releasing carbohydrate administration.***
- ***The availability of indigenous low cost specialized medical formulae has made the treatment of IEMs accessible in India. However long-term adherence and requirement of dietician, specialized in the management of IEM are the challenges for the successful outcome.***

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* Professor,
Pediatrics, Fellowship in Clinical Genetics,
Indira Gandhi institute of Child Health,
Bangalore.
email: sanju.gn26@gmail.com

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