INBORN ERRORS OF METABOLISM - II

LONG-TERM FOLLOW-UP AND NEURODEVELOPMENTAL OUTCOMES IN CHILDREN WITH INBORN ERRORS OF METABOLISM

***Umamaheswari Balakrishnan**
****Rangan Srinivasaraghavan**

Abstract : *Inborn errors of metabolism affect 1 in 1000-3000 live births. Without universal newborn screening in India, diagnostic timing varies widely, delaying intervention. Many children experience substantial neurodevelopmental burden despite metabolic stabilization. Outcomes vary widely depending on disorder type, timing of diagnosis, metabolic control and access to care. Effective follow-up requires regular biochemical and clinical monitoring, structured neurodevelopmental assessment using WHO International Classification of Functioning, Disability and Health frameworks, coordinated metabolic clinics and active parental involvement. Strengthening such systems in resource-limited settings is essential to translate survival into meaningful developmental gains. This narrative review outlines a structured follow-up program for children with inborn errors of metabolism, including clinical and biochemical monitoring, neurodevelopmental services, parental roles and establishment of dedicated metabolic clinics.*

Keywords : *Treatable IEMs, Follow up, Metabolic clinic, Biochemical monitoring, Growth, Neurodevelopmental outcome.*

Points to Remember

- ***Neurodevelopmental outcomes in IEMs are variable and influenced by disease type, timing of diagnosis, metabolic control, and access to care.***
- ***Multidisciplinary, longitudinal follow-up is essential for optimizing developmental outcomes. General pediatrician should understand the importance of long term follow-up.***
- ***An ICF-guided neurodevelopmental approach emphasizes functional abilities, participation and quality of life beyond biochemical control.***
- ***Strengthening follow-up systems and continued research are crucial to improving long-term outcomes, particularly in resource-limited settings.***

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* Professor and Head of Neonatology,
Sri Ramachandra Medical College and Research Institute,
Sri Ramachandra Institute of Higher Education and Research (SRIHER), Chennai.
email : drumarajakumar@gmail.com

** Associate Professor,
Developmental Pediatrics Unit,
Christian Medical College, Vellore.

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