

Inborn Error of Metabolism –An Indian Perspective

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ABSTRACT Inborn errors of metabolism (IEM) constitute a diverse heterogeneous group of disorders with protean clinical manifestation presenting mainly in the pediatric population. Though individually rare, together they constitute a significant percentage of children seen in genetic and neurology clinics. With the recent advancement of knowledge of genetic engineering and awareness of clinical presentation of IEM, early diagnosis and treatment facilities has dramatically changed the whole perspective of morbidity and mortality of infants (Burton 1998). This review focuses on selected IEMs and highlights those seen in the neonatal period. Data from Indian centers are presented. It also emphasizes principles of management in these difficult disorders in the context of developing country (Kumta 2005).