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To Err is Genetics: Diagnosis and Management of Inborn Errors of Metabolism (IEM)

Anju Gupta

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ABSTRACT The colossal number and kaleidoscopic clinical presentation of inborn errors of metabolism present an indomitable task to a clinician. Prevention of death or permanent neurological sequelae in patients with these disorders is dependent on early diagnosis and institution of appropriate therapy. It is therefore necessary for the pediatrician to be familiar with the major signs and symptoms of inborn errors of metabolism and the laboratory studies necessary to arrive at an initial diagnosis. The aim of this review is to demystify this elusive and extremely heterogeneous group of diseases, to promote clinical vigilance in their detection, and to provide a systematic approach to diagnosis when clinical suspicion is aroused.