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INBORN ERRORS OF METABOLISM - I

RECOGNIZING RED FLAGS IN INBORN ERRORS OF METABOLISM - CLINICAL PRESENTATION AND ALERTS

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Abstract: *Inborn errors of metabolism are a heterogeneous group of rare genetic disorders that pose a significant diagnostic challenge due to their non-specific presentation, necessitating a high index of suspicion from healthcare providers. Early recognition of “red flags” is crucial for timely, life-saving intervention. This manuscript discusses the common clinical and laboratory red flags seen in inborn errors of metabolism.*

Keywords: *Inborn errors of metabolism, Children, Hypoglycemia, Hyperammonemia.*

Points to Remember

- *High index of suspicion in IEMs is crucial for timely intervention*
- *Red flags in IEMs are categorized into three main areas - symptoms, signs and laboratory parameters.*
- *The pattern of hypoglycemia with or without ketosis is a critical differentiator.*
- *Hyperammonemia is a medical emergency*
- *Unusual body odours and age-specific neurological symptoms are key clues*

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