

INBORN ERRORS OF METABOLISM - II

EMERGENCY MANAGEMENT OF METABOLIC CRISES IN INTENSIVE CARE SETTINGS

(Acute Decompensation Protocols, Hyperammonemia, Hypoglycemia, Metabolic acidosis)

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Abstract: Acute metabolic crises in Inborn Errors of Metabolism (IEM) are life-threatening emergencies warranting prompt management to optimize outcomes. These crises typically manifest as sudden deterioration following a healthy interval in neonates and triggered by stressors like infection or fasting in children. Management includes immediate stabilization followed by specific therapeutic strategies: substrate reduction by withholding feeding for 48 hours; toxic metabolite elimination; provision of adequate calories and deficient nutrients and enhancement of enzyme activity by cofactor supplementation. Clinical protocols recommend restarting enteral feeding with breast milk or special formula milk within 48 hours, barring specific contraindications. This review comprehensively details these essential principles and strategies for managing IEM crisis.

Points to Remember

- *Early recognition and timely intensive care for metabolic crises in IEM are crucial for preventing mortality.*
- *Initial investigations of 'GALAKS' - glucose, arterial blood gases, lactate, ammonia, ketones, and urine reducing substances help in guiding further approach.*
- *Stabilization with airway, breathing, circulation support and correction of hypoglycemia, seizures, and metabolic acidosis form the foundation of acute management.*
- *Substrate reduction is the key and is achieved by nil per oral route, adequate glucose infusion to prevent catabolism and initiation of special formulas if the IEM diagnosis is known.*
- *Elimination of toxic metabolites should be done by scavengers like sodium benzoate in hyperammonemia and followed by extracorporeal detoxification techniques.*
- *Supplementation with cofactors and vitamins, such as biotin, thiamine and riboflavin will help in specific IEMs.*
- *Prevention of recurrent crises is by multidisciplinary long-term management involving metabolic specialist, pediatrician, dietician and neurologist.*
- *Genetic counselling and metabolic autopsy in critical end stage cases is important for preventing recurrence of the disorder in future pregnancies.*

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