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ORAL PRESENTATION

Hyperammonemia in neonate

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Hyperammonemia in neonates is a critical metabolic emergency that requires prompt recognition and management to prevent irreversible neurological damage or death. It is characterized by elevated levels of ammonia in the blood, often resulting from inborn errors of metabolism, such as urea cycle disorders (UCDs), organic acidemias, and certain fatty acid oxidation defects. In neonates, the presentation is typically non-specific, including poor feeding, lethargy, vomiting, hypothermia, hypotonia, seizures, and coma, which may easily be mistaken for sepsis or other neonatal conditions.

Early diagnosis is vital, and plasma ammonia levels should be measured in any neonate with unexplained encephalopathy or progressive deterioration. Ammonia levels exceeding 150 µmol/L warrant further investigation, while levels >300 µmol/L are considered severe and require urgent intervention. Initial management focuses on halting protein intake, providing high-energy glucose infusions to prevent catabolism, and initiating ammonia-scavenging agents such as sodium benzoate or sodium phenylacetate. In severe cases or those unresponsive to medical therapy, dialysis (preferably hemodialysis or continuous renal replacement therapy) may be necessary to rapidly reduce ammonia levels.

Definitive diagnosis involves metabolic and genetic testing to identify the underlying disorder, which guides long-term management and genetic counseling. Advances in newborn screening programs have improved early detection of some metabolic conditions, allowing for presymptomatic treatment and better outcomes. However, for many urea cycle and other rare metabolic disorders, clinical vigilance remains essential.

Hyperammonemia in neonates poses a diagnostic and therapeutic challenge due to its rapid progression and high risk of morbidity. A high index of suspicion, timely investigation, and aggressive management are key to improving survival and preserving neurological function.

Biography

Maha Alotaibi is a champion of genetic disease in ministry of health. She has plenty of researches in genetic and metabolic disorders and Ad-Hoc Reviewer. She is training and teaching the medical students from different medical collages and residents in Riyadh. She also participates as a speaker in international and local conferences.

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