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Considerations on urea cycle disorders: a child with ornithine transcarbamylase deficiency

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Keypoints

Preoperative infusion of 10% dextrose associated with phenylbutirate and arginine should be prescribed to optimize the metabolic state of the patient. Careful premedication and induction to reduce the psychological stress and maintain hemodynamic stability are indicated. Avoid endotracheal intubation and neuromuscular relaxants if possible. Assure a suitable pain control with regional techniques.

Abstract

Ornithine transcarbamylase deficiency is the most common inside the group of the urea cycle disorders. Anesthesic management focus on reduce the patient´s stress along all the surgical process. A proper management of patient´s anxiety, metabolic and pain control during surgery helps to reduce the hyperammonemia on the early postoperative period.

Keywords: Ornithine Carbamoyltransferase Deficiency Disease, Hyperammonemia, Urea Cycle Disorders, Citrulline, Metabolic Diseases, Epidural Analgesia

Case report

The urea cycle disorders are a group of diseases characterized by the abscence of some enzymes involved in ammonium degradation. They are very rare entities, which have an incidence of 1 per 30000 newborn. The most prevalent is the ornithine transcarbamylase (OTC) deficiency. This enzyme is responsible for combining ornithine and carbamoylphosphate aminoacids which produce citrulline. The lack of this enzyme is an inheritance disease x-linked. The disease's course may have an early manifestation or a late one, which depends on the activity levels of this enzyme. These patients usually have a bad prognosis, and a high morbidity and mortality, especially in relation to brain damage. The clinical manifestation of this metabolopathy usually is characterized by vomiting, respiratory distress, headache, lethargy, decrease of the consciousness level, hypotonia, ataxia, seizures and coma. Stressful situations like infections, dehydration states, protein diets, parenteral nutritions, fasting and the surgical stress, may precipitate episodes of encephalopathy due to the accumulation of ammonium, which can aggravate the neurological prognosis. In these cases, the medication tries to stop possible triggers, optimizes caloric intake and increases the

removal of ammonium. Our case was a male of eleven years old, diagnosed with an OTC deficit, who was programmed for a tibial rotational osteotomy. He was admitted to the hospital twelve hours before surgery to optimize the metabolic state through a continuous infusion of 10% dextrose associated with phenylbutirate and arginine. In order to reduce the psychological stress of the child we have used intranasal midazolam as premedication, then, induction was made with intravenous midazolam, ketamine and fentanyl. A laryngeal mask was placed. After that, a lumbar epidural catheter was set for an analgesic control intra and post operatively. A continuous infusion of 0.125% bupivacaine was set up at 2 ml/h. The maintenance was performed with inhalation anesthesia with sevoflurane. The fluid therapy was based on the infusion of 10% dextrose, monitoring biochemistry values periodically, which were normal all the time. During the 90 minutes of surgery, the patient remained hemodynamically stable, normothermic and the oxygen saturation levels and the exhaled CO_2 were kept within the normal range. The eduction was performed in the operating room without incident. The patient was subsequently moved to the pediatric intensive care unit, keeping the administration of phenylbutyrate and arginine infusion. The patient had no neurological, metabolic or laboratory abnormalities within 48 hours of admission, so it was decided to discharge him to pediatric orthopedic hospitalization area.

Discussion and conclusion

The limited literature on anesthesia in these patients coincides with the peroperative management of this case. To sum up, the administration of a right perioperative treatment to avoid a catabolic state, the association of ketamine, midazolam and fentanyl to maintain hemodynamic stability and the use of regional analgesia for pain control. In this case, the placement of a laryngeal mask let us avoid some drugs, like neuromuscular relaxants, and the stress related to the intubation. As mentioned above, the surgical and anesthetical stress may trigger an episode of hyperammonemia, so a right metabolic control is essential during all the process.

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