ADOLESCENT IN ICU WITH CONVULSIONS, CEREBRAL EDEMA, AND ALKALOSIS: USE OF HEMODIAFILTRATION IN THE MANAGEMENT OF LEUCINOSIS

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INTRODUCTION: Most aminoacid catabolic activity occurs in the liver but branched chain amino acids (BCAAs), including leucine, isoleucine, and valine, are also catalyzed in nonhepatic tissues, especially cardiac muscle, neurons, and kidneys. The literature is scarce regarding the management of some severe manifestations of maple syrup urine disease (MSUD). It is a metabolic disorder of an autosomal recessive nature resulting from the deficiency in the enzymatic complex responsible for the catabolism of branched-chain amino acids leucine, isoleucine and valine, leading to the accumulation of toxic ketoacids and mainly producing muscular and cerebral toxicity. Situations of high endogenous catabolism such as fever, infections, exercise, trauma or surgery, causes individuals with MSUD to present acute deterioration due to acute leucine intoxication. We describe a case with good clinical evolution after performing hemodiafiltration.

CASE REPORT: A 14-year-old male was diagnosed with MSUD in the neonatal period. He had previous cognitive development delay, with control of the disease through protein restriction and free of branched amino acids, in addition to supplementation with maltodextrin, glucose polymer, L-valine, and l-isoleucine. One week before admission he presented a history of upper airway infection and poor adherence to food restriction, followed by several episodes of vomiting, diarrhea and lowering of the level of consciousness. He evolved with neurological worsening (GCS 8), hypotonia and need for transfer to the ICU due to suspected metabolic decompensation. Laboratory tests showed normal renal function, mild metabolic alkalosis, and normal lactate. and hemogram with no evidence of infection. CT scan showed cerebral edema. Serum aminoacid levels (HPLC chromatography method) of BCAAs were: leucine=1758 μM/L (normal: 49-216); isoleucine= 627 μM /L (normal: 22-107); valine 927.5 μM/L (normal: 74-321). Diuresis was normal (1.25 mL/kg/h). He received specific management for intracranial hypertension and underwent continuous venous hemodiafiltration for rapid removal of toxic metabolites. There was a neurological improvement after 24 hours of therapy and on the third day, the serum amino acid level was within normal limits.

DISCUSSION: In adolescents, elevated levels of leucine (leucinosis) can produce epigastric pain, anorexia, nausea and vomiting, muscle fatigue, altered level of consciousness, psychiatric symptoms, change in movement and ataxia, sometimes simulating acute decompensation similar to Wernicke's encephalopathy (Brackburn et al, 2017).
Some centers describe the use of hemodialysis or hemofiltration to remove the BCAAs from the extracellular compartment. This isolated measure is not able to reestablish the accumulated protein balance. Recently, altered catabolism of branched-chain amino acids (BCAAs) has been linked to physiological changes in the myocardium that may lead to heart failure (Huang, 2011). The prognosis is unfavorable if early treatment is not instituted since neonatal diagnosis, based primarily on measures of dietary leucine restriction, inhibition of protein catabolism, BRAA-free nutritional formulas and judicious supplementation with isoleucine and valine, in addition to frequent clinical and biochemistry controls.

CONCLUSION: The management of patients with decompensated leucinosis who present with higher age is a challenge, because it usually occurs in an adult ICU setting, with professionals unfamiliar with a typically pediatric condition. Leucinosis is a potentially fatal condition that requires the institution of appropriate early treatment for the removal of toxic metabolites. It is necessary to know the indication, prescription and maintenance of non-renal dialysis therapy in these patients, in specific situations.