HYPERAMMONEMIA AND LATE-ONSET UREA CYCLE DISORDERS

Adult post-bariatric surgery patient with rapid weight loss

Patient presentation and history: A 34-year-old female was hospitalized 8 months after gastric bypass surgery with a Roux-en-Y procedure. The patient's presenting symptom was weakness, which quickly progressed to uncontrolled status epilepticus and neurological unresponsiveness requiring intubation.

Pre-bariatric surgery, the patient had been morbidly obese at 400 pounds. Rapid weight loss resulted in a current weight of 160 pounds. The patient had been hospitalized many times post-surgery. The patient had a history of dumping syndrome with hypokalemia and hypernatremia, non insulin-dependent diabetes, nephrolithiasis and depression. The patient tended to avoid protein.

Findings:

- Methyl-resistant staph in the sputum; other cultures (viruses, bacteria, fungi) were negative
- Possible fatty liver; possible pneumonia
- Initial aspartate aminotransferase level: elevated
- Initial CT exam: normal; later exam showed some cerebral edema
- Organic acid profile showed no orotic acid and an elevated 5-oxoproline and lactic acid
- Ammonia level: 442 µmol/L (normal 11-32 µmol/L)

Outcome: After remaining unconscious for several days despite receiving aggressive treatment, the patient was removed from life support.

Final diagnosis (post-mortem): CPS1 (carbamoyl phosphate synthetase) deficiency, a rare urea cycle disorder, along with fatty liver infiltrate and multiple kidney stones

Summary: The patient suffered from malabsorption and nutritional disruption. The rapid weight loss resulted in protein catabolism in tissues and excess nitrogen, resulting in hyperammonemia.

Case source: Summar ML, Barr F, Dawling S, Smith W, Lee B, Singh RH, Rhead WJ, Sniderman King L, Christman BW. Unmasked adult-onset urea cycle disorders in the critical care setting. Critical Care Clinics 2005;21:S1-S8.
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