

HYPERAMMONEMIA AND LATE-ONSET UREA CYCLE DISORDERS

Post-surgical adult trauma patient (self-selected vegetarian)

Patient presentation and history:

A male in his early 30s presented to the ED with multiple fractures following a four-wheeler accident. Post-surgery, the patient became confused and combative. The patient developed tachycardia and generalized tonic-clonic seizures, then progressed to status epilepticus and became comatose.

Past history included a closed head injury at age 5 and subsequent development of a seizure disorder, chronic hypertension, gastroesophageal reflux, and persistent anemia. The patient also had a history of nonspecific psychiatric symptoms, was described as “slow,” and was a self-selected vegetarian.

Findings:

- Lacunar infarct of the left caudate by CT
- Severe brain dysfunction and increased signal in the left front periventricular region by MRI
- Hypertensive (diastolic pressure of 140 mmHg); hyperthermic (reaching 106 degrees F)
- Low blood urea nitrogen
- Ammonia level: 553 $\mu\text{mol/L}$ (normal 11-32 $\mu\text{mol/L}$) at 24 hours into course of coma

Outcome: After aggressive but unsuccessful intervention, the patient was removed from life support.

Final diagnosis (post-mortem): NAGS (N-acetylglutamate synthase) deficiency, a rare urea cycle disorder, determined by liver biopsy with enzymatic testing

Summary: Multiple fractures triggered breakdown and processing of blood that was lost into the tissues surrounding the fractures, releasing large amounts of waste nitrogen that exceeded the clearance capacity of his underlying urea cycle disorder, NAGS deficiency, 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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