HYPERAMMONEMIA AND LATE-ONSET UREA CYCLE DISORDERS

Post-partum adult female with uncharacteristic behavior

Patient presentation and history:

A 38-year-old female presented at the hospital 5 days postpartum (first pregnancy) with acute onset of behavioral symptoms. The pregnancy was uncomplicated, but breech presentation led to a C-section delivery. The previous day the patient vomited twice and that night awoke confused, with no recognition of her husband and no recollection of childbirth. The patient became uncharacteristically aggressive and violent.

On arrival at the hospital, the patient was disoriented, agitated, and unable to follow commands, but the exam revealed no focal neurological signs. The patient was taking analgesics for post-op pain and had no significant past medical history of substance abuse or psychiatric history. By the next morning, the patient's clinical status had completely normalized. A provisional diagnosis of post-partum psychosis was made.

Later that evening the patient relapsed, becoming disoriented with fluctuating responses to cognitive tasks and profound amnesia for recent events. The patient also had marked verbal and motor perseveration and subtle asterixis. The patient's condition deteriorated, with progressively declining responsiveness and prominent asterixis.

Findings:

- **At presentation:**
  - Blood tests showed mild abnormalities, many possibly explained by recent pregnancy
  - Negative for renal and hepatic dysfunction other than mildly elevated serum alkaline phosphatase
  - CT brain scan: normal
- **At relapse <24 hours later:**
  - Brain MRI: normal; EEG showed generalized delta slowing
  - Plasma ammonia level: 292 µmol/L (normal 11-32 µmol/L)
  - Respiratory alkalosis confirmed by arterial blood gases
  - Plasma arginine and citrulline: low
  - Urinary orotate: elevated
  - Repeat ammonia level: 382 µmol/L (normal 11-32 µmol/L)

Outcome: Treatment for hyperammonemia was initiated after the repeat plasma ammonia test. The patient was discharged after several days and remained well at a 12-month follow-up visit.

Final diagnosis: OTC (ornithine transcarbamylase) deficiency—an X-linked urea cycle disorder and the most common UCD

Summary: The increased catabolic protein load following involution of the uterus during the post-partum period caused metabolic decompensation and hyperammonemia.

Case source: Blair NF, Cremer PD, Tchan MC. Urea cycle disorders: a life-threatening yet treatable cause of metabolic encephalopathy in adults. *Practical Neurology* 2015;15:45-48.

http://bit.ly/2IbasbX