

## HYPERAMMONEMIA AND LATE-ONSET UREA CYCLE DISORDERS

## Adult patient (protein avoidance) receiving high-dose IV steroids

**Patient presentation and history:**

A 58-year-old female with recurrent episodes of “asthmatic bronchitis” presented to the hospital with severe wheezing of several days’ duration and a productive cough. The initial diagnosis was viral illness that was exacerbating the patient’s asthma. When the condition worsened, the patient was admitted to the hospital and was administered IV steroids. Five days later the patient developed acute confusion and an expressive aphasia with focal deficits, progressing to complete coma within 48 hours. The patient was thought to have steroid psychosis.

Past history included episodic asthma, allergic rhinitis, and nasal polyps. Previous surgeries included a cholecystectomy, appendectomy, hysterectomy, and a thyroid nodule treated by radiation. The patient had never before received IV steroids. The patient ate a normal diet but tended to avoid large protein intakes.

**Findings:**

- Lumbar puncture, EEG, two head CTs, carotid Doppler test, and cerebral 3-vessel arteriogram: all nondiagnostic
- Temperature and pulse: normal; blood pressure: 142/80 mmHg
- Cerebral edema on CT
- Diffuse slowing but no seizures on EEG
- Citrulline and arginine: very low
- Glutamine: mildly elevated
- Ammonia levels: 120  $\mu\text{mol/L}$ , later rising to 280  $\mu\text{mol/L}$  (normal 11-32  $\mu\text{mol/L}$ )

**Outcome:** After 3+ days of aggressive treatment for hyperammonemia, the patient was discharged from the hospital; a 5-year follow-up showed no residual deficits.

**Final diagnosis:** Carrier for OTC (ornithine transcarbamylase) deficiency—an X-linked urea cycle disorder and the most common UCD, with many family members diagnosed with OTC deficiency, including 4 infant sons who had died as a result of OTC deficiency

**Summary:** The IV steroids caused increased protein turnover, resulting in excessive nitrogen release and 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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