

Critically High Plasma Ammonia in an Adolescent Girl

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CASE DESCRIPTION

A 12-year-old female patient presented with nonspecific symptoms including fatigue, fever, and headache persisting for approximately 4 months. Plasma ammonia was 230 µg/dL (135 µmol/L) (critical value >187 µg/dL or 110 µmol/L). The patient was referred to a pediatric emergency center for urgent evaluation of hyperammonemia. Plasma and urine amino acid testing was performed to investigate the possibility of a urea cycle disorder; however, all results were normal. Over the following 2 weeks, the patient was treated with intravenous sodium benzoate/sodium phenylacetate and L-arginine to reduce the blood ammonia concentration. However, the patient's ammonia concentration never dropped below 94 µg/dL (55 µmol/L) (upper limit of reference interval, 80 µg/dL or 47 µmol/L), with concentrations as high as 296 µg/dL (174 µmol/L) with inconsistent associations between ammonia concentrations and headaches. During the course of treatment, the patient suffered substantial side effects of intravenous therapy and underwent numerous costly clinical and laboratory investigations. Amino acid investigations were repeated but results remained normal. Given the large fluctuations in blood ammonia concentrations with inconsistent response to intravenous medications, the metabolic service contacted the laboratory to inquire about a possible preanalytical or analytical cause. One week before this query, a community physician also contacted the laboratory regarding hyperammonemia in several patients. However, no obvious preanalytical or analytical errors were discovered despite numerous audits of preanalytical conditions and review of quality control data.

QUESTIONS TO CONSIDER

- What is plasma ammonia and why is it important?
- What are the physiologic causes of hyperammonemia?
- What is the appropriate treatment for hyperammonemia?
- What are the preanalytical causes of hyperammonemia?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the December 2016 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol62/issue12> and follow the link to the Clinical Case Study and Commentaries.

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