INTRODUCTION

- The urea cycle is a series of catalytic steps for the metabolism of waste products (i.e., ammonia).
- Deficiency in any of these enzymes is referred as Urea Cycle Disease (UCD).
- Elevation of ammonia is a common finding of UCD.
- Argininosuccinate lyase deficiency (ASLD) is a subtype of UCD.
- There are two phenotypic presentations:
  - Neonatal ASLD ➔ Complete absence of ASL enzyme.
  - Late onset ASLD ➔ Partial absence of the ASL enzyme.
- ASLD of late onset manifests as transient hyperammonemia triggered by an acute stressor.
- Hyperammonemia can lead to life-threatening cerebral edema.
- Here we present the case of a patient who developed hyperacute elevations of ammonia in the setting of fatal underlying ASLD.

CASE SUMMARY

- A 51-year-old male presented with septic shock secondary to right obstructive uropathy s/p extraction of a ureteral calculi and stent placement.
- Blood cultures grew Corynebacterium and he was placed on empiric piperacillin-tazobactam.
- Initial liver enzymes were AST 24 U/L, ALT 35 U/L, and ammonia level was elevated at 99 umol/L.
- The patient remained unresponsive over the first 24 hours postop.
- Follow up workup showed acute increase of ammonia to 509 umol/L, AST at 327 U/L, and ALT at 325 U/L.
- A head CT showed diffuse cerebral edema without herniation or hemorrhage.
- Based on these acute findings, a metabolic panel was sent out for suspected urea cycle disease.
- Results were significant for elevated urine ornithine 8.9 mmol/molCr, plasma citrulline 155 nmol/mL, and argininosuccinic acid 477 nmol/mL.
- These findings confirmed hyperammonemia in the setting of ASLD.
- Normalization of ammonia and improvement of cerebral edema was achieved via CRRT and lactulose.

DISCUSSION

- Hyperammonemia should be considered in ICU patients when there is acute mental status change without a clear cause.
- Undiagnosed UCD should be considered in older patients presenting with nonhepatic elevated ammonia.
- Rapid correction of ammonia levels with dialysis is pertinent and safe. Correction of hyperammonemia can reverse cerebral edema without causing drastic fluids shifts.

CONTACT INFORMATION:
Email: Lepes22@ecu.edu
Phone: (704) 900-7559