

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 orotic acid 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 .

Component	(L) Low (H) High
Phosphoserine	0
Phosphoethanolamine	<2
Taurine	20 (L)
Asparagine	53
Serine	49 (L)
Hydroxyproline	16
Glycine	192
Glutamine	481
Aspartic Acid	3
Ethanolamine	14
Histidine	44
Threonine	86
Citrulline	155 (H)
Sarcosine	2
Beta-Alanine	9
Alanine	315
Glutamic Acid	75
1-Methylhistidine	1
3-Methylhistidine	11 (H)
Argininosuccinic Acid	477 (H)
Carnosine	0
Anserine	0
Homocitulline	15 (H)
Arginine	63
Alpha-amino adipic Acid	3 (H)
Gamma-amino-n-butyric Acid	0
Beta-aminoisobutyric Acid	2

Table 1: Amino acid Assay

Alpha-amino-n-butyric Acid	13
Hydroxylysine	0
Proline	136
Ornithine	55
Cystathionine	4
Cystine	26
Lysine	246
Methionine	44
Valine	194
Tyrosine	50
Isoleucine	66
Leucine	117
Phenylalanine	68
Tryptophan	36
Allg-isoleucine	0

Table 1 (contd): Amino acid Assay

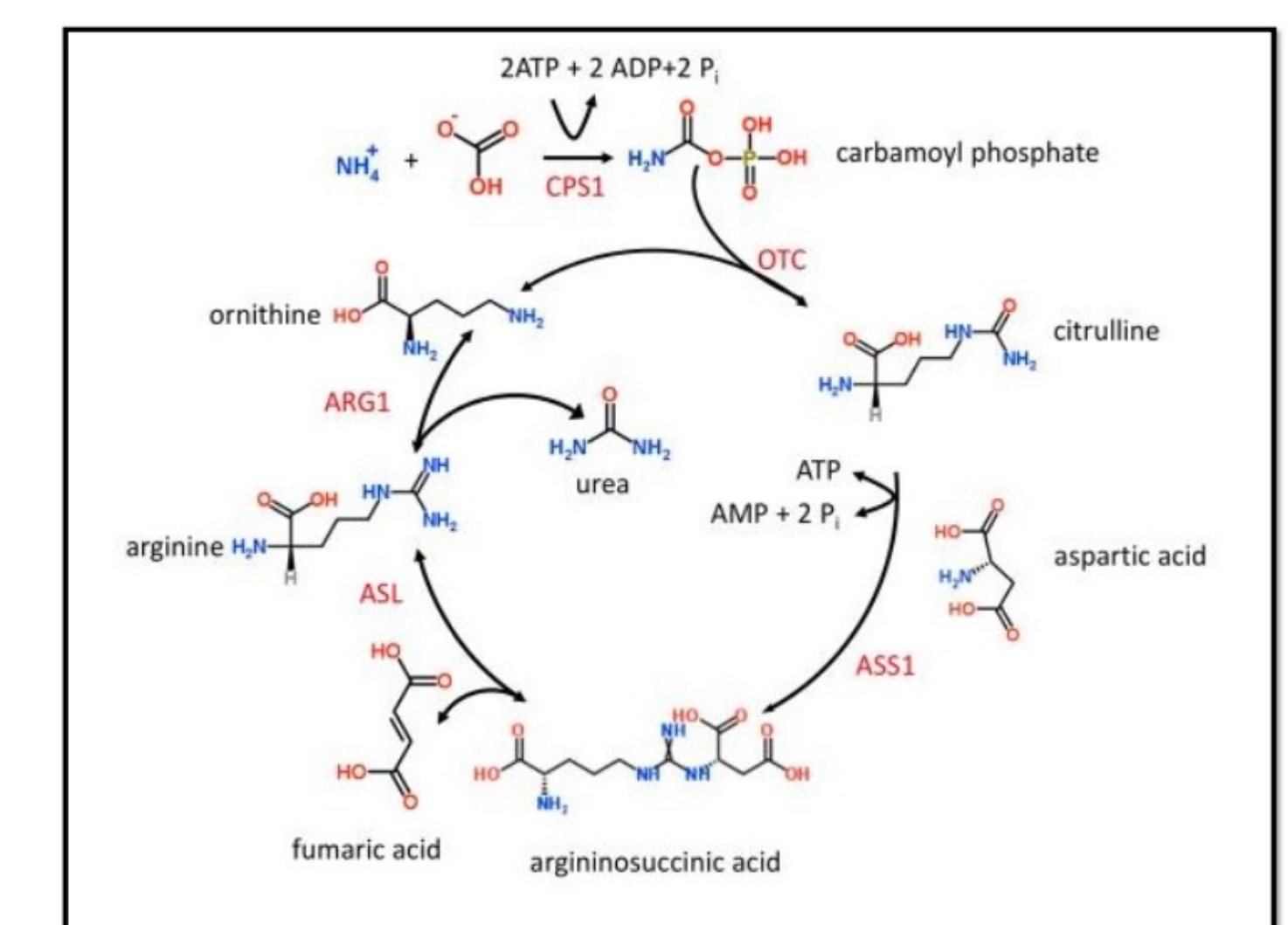


Figure 3: Uric acid cycle



Fig: 1 CT Head imaging on arrival



Fig:2 CT Head showing cerebral edema [elevated ammonia]

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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.