

# Peripartum Management of Carbamoyl Phosphate Synthetase I Deficiency: A Case Across Two Pregnancies

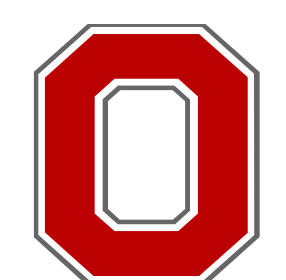
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CPS1 deficiency is an AR urea cycle disorder with hyperammonemia, exacerbated by catabolic states such as labor and postpartum uterine involution.

**Case:** 22-year-old G2P0, cesarean delivery at 34w2d for PPRROM with breech

- **POD2:** agitation requiring intubation; lactate 10, ammonia 228; treated with CVVH and sodium phenylbutyrate; evaluated for portal vein thrombosis
- **Course:** transferred for genetics care to a children's hospital; received IV sodium phenylacetate/benzoate (Ammonul) and arginine
- **Discharge:** low-protein diet and oral glycerol phenylbutyrate
- **Genetics:** compound heterozygous CPS1 mutations (c.1312G>C; p.Ala438Pro and 4274+2T>C splice)



## Pregnancy 2 (Age 30):

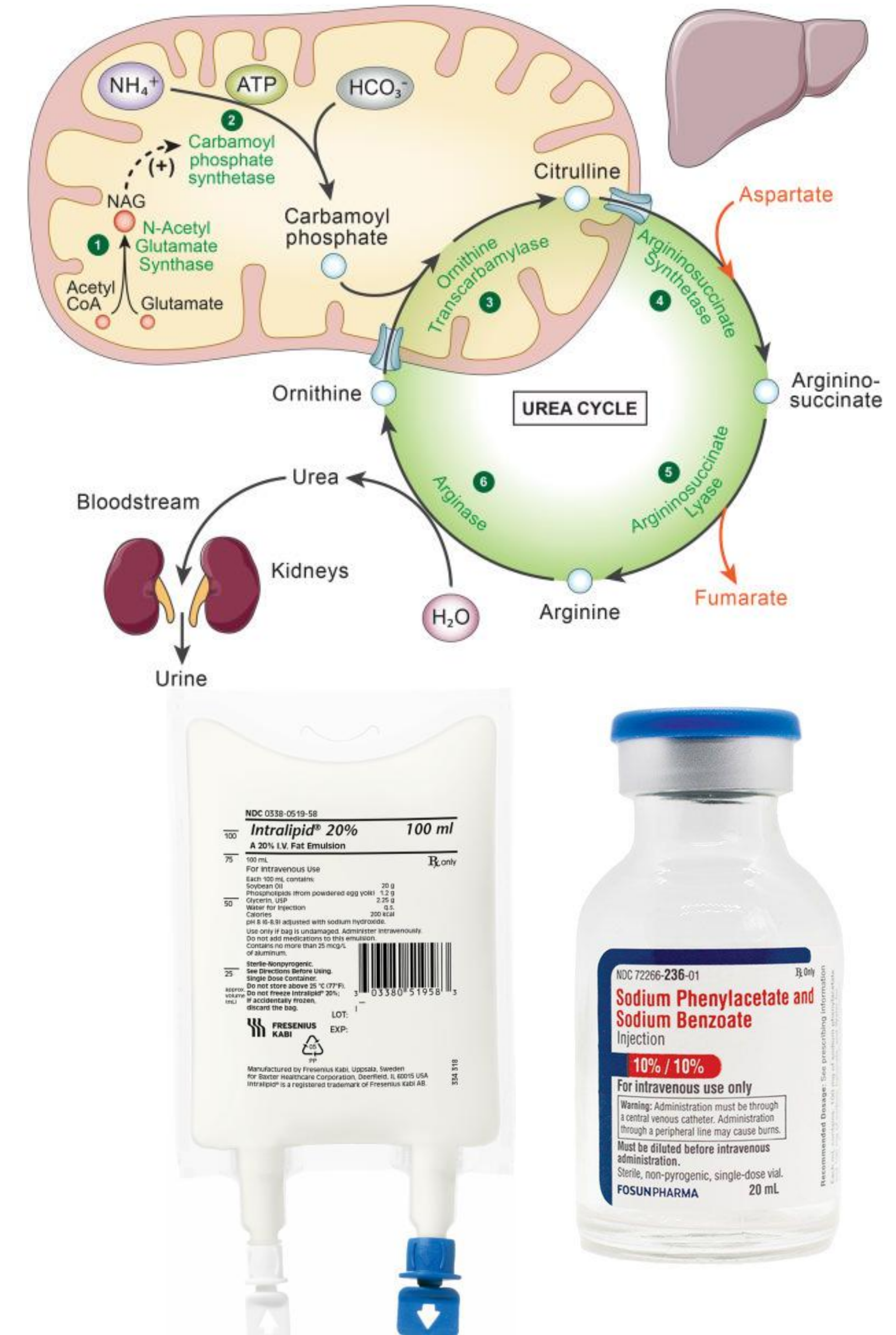
- 37w3d induction, TOLAC; pregnancy complicated by IUGR
- Baseline ammonia 60  $\mu\text{mol/L}$ ; stable (60 - 80) during labor with epidural
- Repeat C-section for NRFHT; intraoperative hemorrhage requiring transfusion

## Postpartum Course:

- Ammonia peak 160  $\mu\text{mol/L}$
- Treated with D10 infusion + 20% Intralipid (2 g/kg) for 24h to promote anabolism. Ammonia improved to 79  $\mu\text{mol/L}$
- POD2 fever, treated as endometritis (clindamycin/gentamicin)
- ED presentation 2 weeks postpartum: lethargy with ammonia 78  $\mu\text{mol/L}$ , improved with hydration

## Outcome:

- Ammonia 77 - 148  $\mu\text{mol/L}$  postpartum; neurologically intact
- Discharged POD4 with ammonia 80  $\mu\text{mol/L}$  on oral therapy



Pregnancy can unmask urea cycle disorders; presentations include encephalopathy, psychosis, hallucinations, vomiting, abdominal pain.

### **Management:**

- Maintain anabolic state with IV D10 and 20% Intralipid (2 g/kg)
- Implement nitrogen scavengers

### **Risk Factors for Hyperammonemia:**

- Labor with intense muscle activity, poor oral intake
- Postpartum uterine involution with increased amino acid load
- Infection, surgery, pain, sleep deprivation
- Steroids, transfusions, albumin due to increased catabolism or protein load
- Breastfeeding increases energy demand (approximately 500 kcal/day); involve a dietician

### **References:**

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