Patient presentation and history:
A female—born at 39 weeks after an uneventful pregnancy—began to have symptoms at approximately 60 hours of age. The patient was irritable and hypertonic with tachypnea and tachycardia. The patient’s condition worsened to deep coma with dominant circulatory failure, tachycardia, and hypotension. At approximately 150 hours of age, the patient, on artificial ventilation, was admitted to a neonatal intensive care unit.

Findings:
• At 60 hours of age:
  - Respiratory alkalosis; mild polycythemia
• At 150 hours of age:
  - Cardiac failure: negative
  - Sepsis screening: negative
  - Glutamine, alanine, methionine, glycine, and lysine: elevated
  - Serum citrulline: undetectable; serum arginine: low to low-normal
  - No urinary argininosuccinic acid or its anhydrides
  - Urinary orotic acid: extremely elevated
  - Plasma ammonia level: 981 µmol/L (normal <100 µmol/L)

Outcome: Aggressive intervention reversed most clinical symptoms and normalized most biochemical findings within a few days. In her 7th month, the patient was in the 75th percentile for weight and 5th percentile for head circumference. The patient’s psychomotor development corresponded to 3-4 months of age, indicating moderately severe retardation.

Final diagnosis: OTC (ornithine transcarbamylase) deficiency—an X-linked urea cycle disorder and the most common UCD, based on clinical course, biochemical tests, and subsequent DNA analysis

Summary: Neonatal-onset of OTC deficiency caused severe, rapidly progressive hyperammonemia and associated clinical manifestations, with signs of irreversible brain damage.