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Neonatal Hyperammonemia Mimicking Early-Onset Sepsis: A Case Report of Hyperammonemic Encephalopathy Due to a Urea Cycle Disorder

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Abstract

Background: *Metabolically defective classic infantile hyperammonemia is an infrequent but substantial diagnosis that can be obscured and lead to rapid development of cerebral edema with vasogenic brain injury when not identified early. In newborns, urea cycle disorders [1] are the most frequent cause of severe hyperammonemia. These disorders manifest at a similar stage of echocardiographic development as sepsis and very-early-onset infection (VUE) [2].*

Case Presentation: *A lethargic term male infant with poor feeding and tachypnea at 36 h of life was suspected to have early-onset neonatal sepsis. Despite being placed on antibiotics, the baby's neurological status gradually declined. Tests revealed they had a very high level of serum ammonia (428 µmol/L) at a time when it should have been 15 to 45 µmol/L, and an MRI of the brain showed swelling in all dimensions, labeled "hyperammonemic encephalopathy." She would immediately start metabolic management with protein restriction, intravenous glucose delivery, ammonia-scavenging therapy, and urgent hemodialysis. This resulted in an early biochemical remission, with the clinical status being stable. A urea cycle disorder was revealed by a later metabolic test.*

Conclusion: *This case highlights the diagnostic challenge of distinguishing metabolic disorders from neonatal sepsis. Early measurement of serum ammonia levels should be considered in neonates presenting with unexplained neurological deterioration or poor response to antibiotic therapy. Prompt recognition and treatment are essential to prevent severe neurological complications.*

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Introduction

Neonatal hyperammonemia hypersensitive care clinical medical emergency rapid diagnosis and management Elevated ammonia levels are highly neurotoxic,

and cerebral edema, seizures, coma, and irreversible neurologic damage can occur without urgent intervention. UCDs are a group of inherited metabolic disorders characterized by enzyme deficiencies that impede ammonia detoxification in the liver, and they are the most common cause of severe hyperammonemia in neonates [3-5].

Case Presentation

A term male neonate was delivered at 39 weeks of gestation via spontaneous vaginal delivery to a 27-year-old primigravida mother. Pregnancy and antenatal screening were unremarkable. Birth weight was 3.2 kg, and Apgar scores were 8 and 9 at one and five minutes, respectively.

The infant remained clinically stable during the first 24 hours of life.

At approximately 36 hours of life, the neonate developed:

- Poor Feeding
- Irritability
- Tachypnea
- Progressive Lethargy

The infant was transferred to the Neonatal Intensive Care Unit (NICU) for further evaluation.

Clinical Examination

Vital signs on admission were:

Parameter	Value
Heart rate	170 bpm
Respiratory rate	72/min
Temperature	37.6°C
Oxygen saturation	94% room air
Blood pressure	60/34 mmHg

Neurological examination revealed **hypotonia, weak suck reflex, and decreased spontaneous movements**. No dysmorphic features were noted.

Given the clinical presentation, early-onset neonatal sepsis was suspected, and empirical intravenous antibiotics (ampicillin and gentamicin) were initiated after blood cultures were obtained.

Laboratory Findings

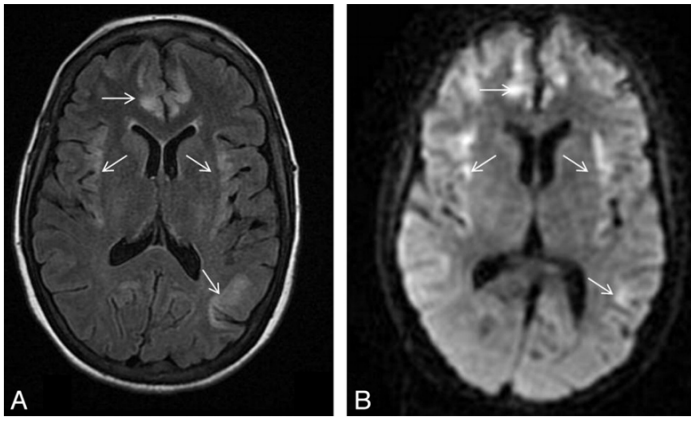
Initial laboratory investigations showed:

Test	Result
White blood cell count	13,500/mm³
C-reactive protein	3 mg/L
Blood glucose	76 mg/dL
Blood gas analysis	Respiratory alkalosis

Despite treatment, the infant's neurological status deteriorated with increasing lethargy and decreased responsiveness.

Serum ammonia was measured and found to be:
428 µmol/L (normal <100 µmol/L)
This confirmed **severe hyperammonemia**

Neuroimaging



Brain MRI demonstrated **Diffuse Cerebral Edema involving Cortical and Subcortical Regions**, consistent with **Hyperammonemic Encephalopathy**.

Clinical Timeline

Figure 1: Timeline of Clinical Events in the Neonate with Hyperammonemia

1. Birth at 39 weeks - Healthy Term Neonate
2. 36 hours - Poor Feeding, Lethargy, Tachypnea
3. Day 2 - Neurological Deterioration, Ammonia Level 428 µmol/L
4. Day 2-3- Metabolic Therapy Initiated
5. Day 3 - Hemodialysis performed with rapid improvement

Management

Immediate treatment was initiated to rapidly lower ammonia levels.

Metabolic Stabilization

- Discontinuation of protein intake
- Intravenous glucose infusion
- Lipid infusion for caloric support

Ammonia-Scavenging Therapy

- Sodium benzoate
- Sodium phenylacetate
- Intravenous arginine

Dialysis

Because ammonia levels remained elevated above 300 µmol/L, urgent hemodialysis was performed.

Ammonia Trend

Time	Ammonia Level
Admission	428 µmol/L
After treatment	290 µmol/L
After dialysis	120 µmol/L
Day 3	72 µmol/L

Outcome

Following dialysis and metabolic therapy, the infant's neurological status improved gradually. The patient was extubated on day 5 of life.

Subsequent metabolic testing confirmed a Urea Cycle Disorder, and the infant was discharged with specialized metabolic follow-up and dietary management.

Discussion

Primarily concerning hyperammonemic neurotoxicity. Ammonia crosses the blood–brain barrier, and astrocytes metabolize it to glutamine, resulting in osmotic swelling (cerebral edema). This general mechanism accounts for the acute neurological deterioration of affected neonates.

Disorders of urea metabolism are still the most frequent cause of severe hyperammonemia in newborns. Genetic defects of such enzymes, e.g., ornithine transcarbamylase (OTC) or carbamoyl phosphate synthetase I (CPS1), physiologically oversee normal ammonia biotransformation in the liver.

Hyperbetalipoproteinemia and neonatal sepsis, two of the main diseases in our continuum, are diseases we want to detect through their pathophysiological relationship. The ideality staging of subcarinal lesions is akin to that of GGO, with the caveat that an increased maximum effect at that persistence was additionally observed. In a young, well individual, all options need to be explored, and sarcoidosis should always be considered in the background. This requires an open lung biopsy or bronchoscopy with bronchial washing, but in this case, transbronchial methods could also work.

However, several clinical clues may suggest a metabolic disorder:

- **Respiratory Alkalosis** on blood gas analysis
- Rapid Neurological Deterioration
- Lack of response to Antibiotic Therapy

Treatment focuses on three main principles:

1. Reducing Ammonia Production
2. Enhancing Nitrogen Excretion
3. Rapid removal of ammonia through dialysis

Hemodialysis remains the most effective therapy for rapidly lowering ammonia levels in severe cases.

Delayed diagnosis is associated with poor neurological outcomes, highlighting the importance of early ammonia measurement in neonates with unexplained encephalopathy.

Hyperammonaemia should be considered as an unusual etiology of encephalopathy in tactile infants and because a late diagnosis will always induce a poor neurologic outcome, it must be routinely searched with ammonia measurement whenever unexplained encephalopathy occurs.

Clinical Learning Points

- Neonatal Hyperammonemia may initially mimic early-onset neonatal sepsis.
- Respiratory Alkalosis is an important diagnostic clue.
- Serum Ammonia should be measured in neonates with unexplained neurological deterioration.
- Hemodialysis is the most effective therapy for severe hyperammonemia.

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