Hospital Universitario La Paz
Hospital Carlos III
Hospital Cantoblanco

eahp european association of hospital pharmacists

4CPS-063

LATE-ONSET ORNITHINE TRANSCARBAMYLASE DEFICIENCY IN A PAEDIATRIC PATIENT: SUCCESSFUL MANAGEMENT OF A METABOLIC EMERGENCY — A CASE REPORT

[1]ME Ibáñez Ronco, [1]AB Arancón Pardo, [1]C Jiménez Nuñez,[1]A Herrero Ambrosio
[1] Hospital Universitario La Paz, Pharmacy Department, Madrid, Spain

BACKGROUND AND IMPORTANCE

Ornithine transcarbamylase deficiency (OTCD) is a rare X-linked urea cycle disorder causing impaired ammonia detoxification; late-onset forms are frequently misdiagnosed due to non-specific symptoms, delaying treatment and increasing the risk of severe metabolic encephalopathy or death.

AIM AND OBJECTIVES

To describe the clinical management of a paediatric patient with late-onset OTCD and highlight the therapeutic role of hospital pharmacists in metabolic decompensations.

MATERIALS AND METHODS

An 11-year-old male with no relevant family history was admitted with:

- Vomiting
- Somnolence
- Hyperammonaemia (480 µmol/L)
- Hyperlactacidaemia
- Hepatic dysfunction

On admission, he received:

- Hydroxocobalamin
- Biotin
- Cerebral oedema management
- Continuous Veno-Venous Hemodiafiltration (CVVHDF) for rapid ammonia reduction

Initial pharmacological therapy included:

- Sodium benzoate with sodium phenylacetate (alternated with phenylbutyrate)
- Arginine
- Carnitine
- Carglumic acid
- L-citrulline
- Paromomycin
- Lactulose

The hospital pharmacist validated parenteral nutrition with protein restriction, compensated by higher carbohydrate intake to promote anabolism and protein synthesis, and provided basal fat to meet energy needs without overloading metabolism or causing acidosis/stress.

RESULTS

- ✓ CVVHDF rapidly reduced ammonia levels with clinical improvement.
- ✓ Therapy was adjusted for outpatient management, maintaining phenylbutyrate, citrulline, and strict protein control.
- ✓ Diagnosis was supported by metabolic findings:
  ↑ urinary orotic acid + ↑ glutamine + ↓ citrulline ⟶ clinical suspicion of OTCD.

- ✓ Genetic testing later confirmed a hemizygote OTC gene mutation.
- ✓ Maternal testing revealed mutation G4A1V3, despite an uncomplicated pregnancy and delivery.
- ✓ The patient remained clinically stable with no further decompensations.

CONCLUSION AND RELEVANCE

OTCD can occur beyond the neonatal period with non-specific symptoms, delaying diagnosis. Early multidisciplinary intervention, including hospital pharmacists—who play a key role in the rapid initiation and adjustment of therapy and parenteral nutrition—is essential to secure favourable outcomes and avoid irreversible neurological damage.