№ 242

A Case of HYPERAMMONEMIA in a Ornithine Transcarbamylase Deficiency (OTCD) Heterozygous Carrier

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HOW to Define Ornithine Transcarbamylase Deficiency (OTCD) ?

Hereditary metabolic disorder with X-linked recessive inheritance.
The most prevalent genetic disorder within urea cycle disorders **affecting ureagenesis.** (Figure 1)
We would like to discuss on a case of a heterozygous female with OTC deficiency exhibited mild cognitive deficits with impaired functional capability and hyperammonemia.

CASE DESCRIPTION

- A 32 years old female with underlying OTC Heterozygous Carrier with (c.275G>A) gene mutation, presented with **confusion, short term memory loss, nausea and headache** for 2 days, preceded by **fasting in past 3 days**.
- Similar event occurred 4 years ago during her first pregnancy, where this pathogenic genetic mutation had only been detected with a comprehensive genetic workup after she experiencing her first neonatal death.

Physical Examination on arrival:
She was **confused, not orientated to time and place. GCS E4V4M6,** Overall stable haemodynamically but fluctuating consciousness.

Investigations:
Serum Ammonia: 166.1µmol/L *(N: 11 to 51µmol/L)*

- Intravenous hydration was provided at first, subsequently patient's condition improved to her usual self with full consciousness.
- She was then admitted to medical ward with regular dosage of Syrup Lactulose, inpatient dietician referral for low protein diet and monitoring of serum ammonia level.

DISCUSSION

- ✓ Heterozygous females with ornithine OTC deficiency could be either ranged from asymptomatic, intermittently symptomatic, or to recurrent episodes of hyperammonemic coma, depending upon the degree of enzyme deficiency in gene mutation.
- ✓ During birth, OTC deficiency is not included in our routine neonatal screening, therefore the diagnosis is often delayed, or even misdiagnosed as psychiatric disorder.
- ✓ ***Rapid correction of hyperammonemia to less than 200 µmol/L*** is recommended at acute phase to avoid hyperammonemic crisis.
- ✓ **Primary goals in ACUTE cases**: Adequate hydration, Discontinuing protein intake, Reversing catabolism triggers.
- ✓ **In SEVERE hyperammonemic cases:** Parenteral nitrogen scavenger therapy and Continuous venovenous haemodialysis (CVVHD) are recommended.

CONCLUSION

OTCD is a rare and potentially lethal medical condition. Detection of OTC gene mutation early, with collaborative care among interprofessional teams (geneticists, physicians, nurses, dieticians, and pharmacists), aid in holistic management preventing hyperammonemia.

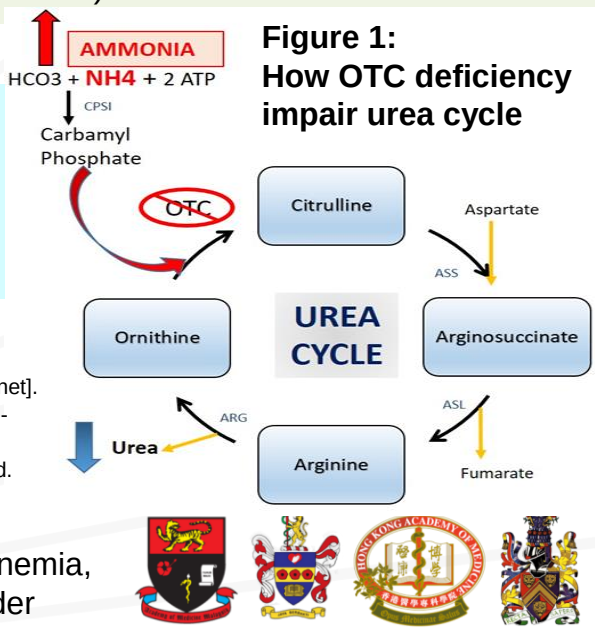

Figure 1: How OTC deficiency impair urea cycle

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