

Managing a Traumatic Unknown: A caesarean section in a women with potential ornithine transcarbamylase deficiency

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Introduction

The number of women with complex metabolic disorders reaching childbearing age has significantly increased¹. Obstetric anaesthetists, as integral members of the multidisciplinary team, must be prepared to manage such cases throughout their careers. Mismanagement can result in catastrophic outcomes.



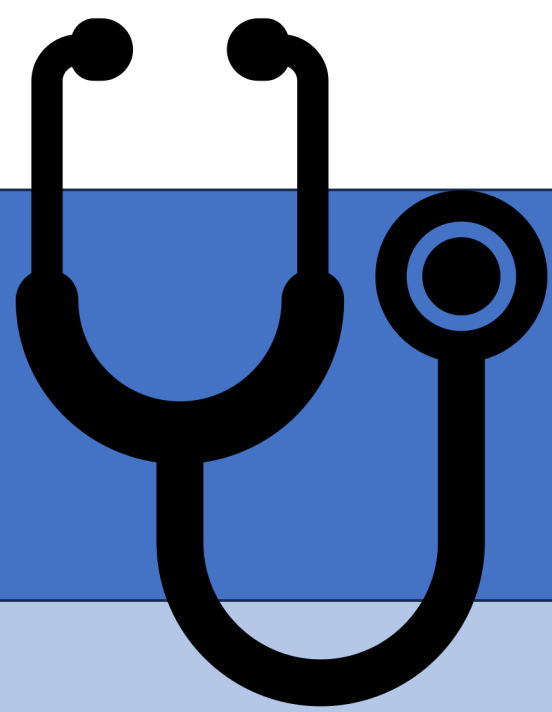
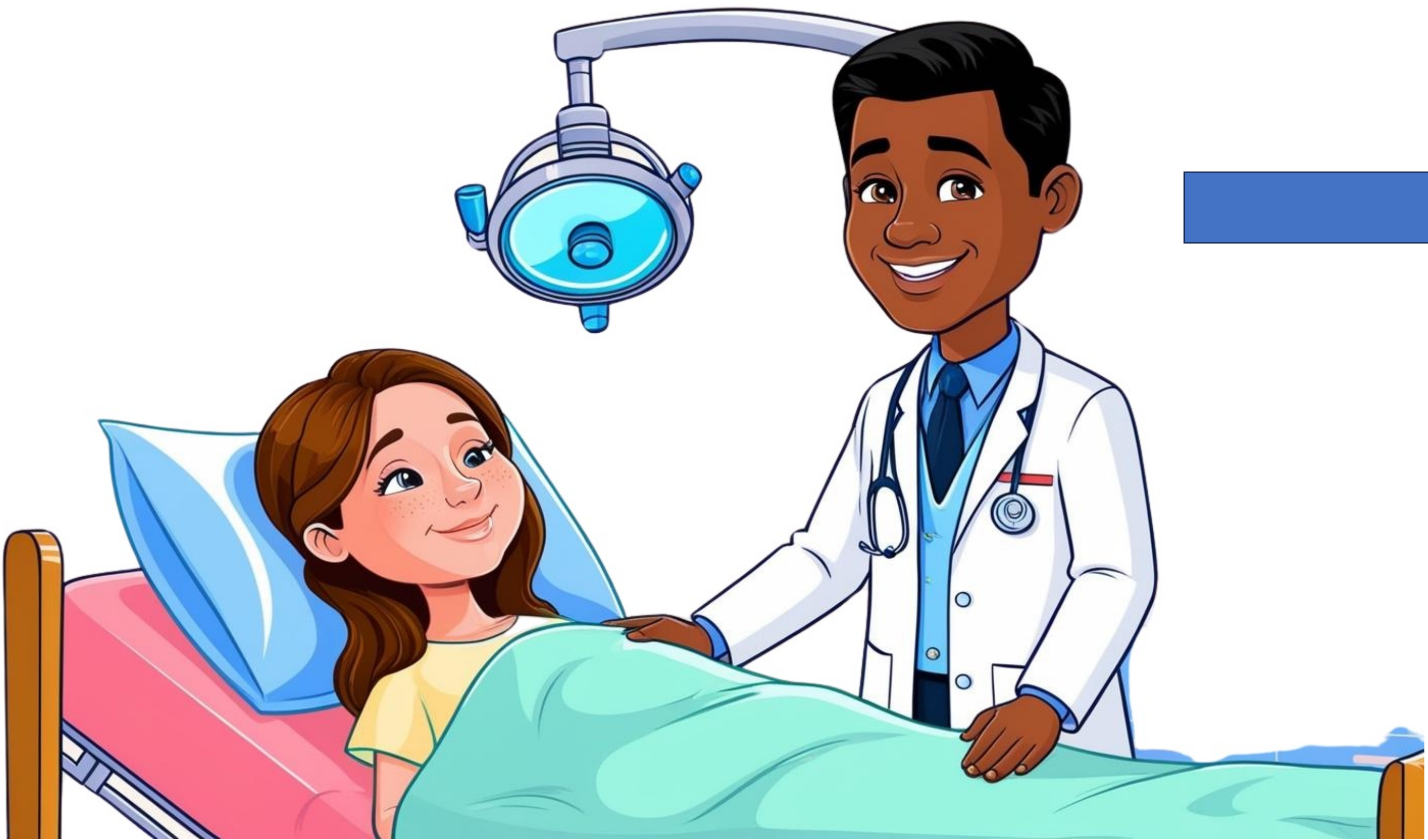
Case Report

A healthy 34-year-old woman presented for an elective caesarean section. Two weeks prior, her sister had experienced rapid deterioration following an instrumental delivery, requiring intensive care for suspected ornithine transcarbamylase deficiency (OTCD). As genetic testing was delayed, this case was treated as presumed OTCD.



Ornithine Transcarbamylse Deficiency

- Genetic metabolic disorder caused by an enzyme deficiency affecting nitrogen conversion to urea.
- OTCD is often asymptomatic, but stressors like pregnancy can trigger hyperammonemic crises.
- Acute neurological, psychiatric or gastrointestinal symptoms during the peripartum period should raise suspicion of OTCD.



Our Practice

- A dedicated, experienced, health professional appointed during the operation as first point of contact between team and patient
- Ensuring the patient is listened to and concerns are addressed promptly and with care
- Multifaceted and multidisciplinary approach to the patient's care pathway emphasizing collaboration among midwifery, obstetric, anaesthetic, and metabolic teams
- Ensuring that follow-up is arranged from clinical and non-clinical aspects

	Pre	Peri	Post
Non-Clinical	Telephone pre-operative assessment Early discussion with metabolic diseases consultant regarding management in the pre-, peri- and postpartum periods	Reduce external stressors	Multidisciplinary post-op review with close involvement of metabolic diseases team Referred to onward management/follow-up from metabolic team
Clinical	Baseline bloods taken for U&Es, ammonia, plasma glucose, quantitative plasma amino acids. Initiated an IV dextrose infusion Ensured supply of ammonia scavenging drugs available.	Invasive blood pressure monitoring Spinal anaesthesia. Tight glycemic control	Admitted to HDU. Rapid re-initiation of food. Half hourly neuro observations. Ammonia, blood gases, U&E's monitored 4 hourly for first 48 hours and 12 hourly for subsequent 5 days. Samples sent for genetic testing.

References:
1. Stepien KM, Langendonk JG, Dao M, Gomes DC, Douillard C, Filipsson K, Glamuzina E, Haverkamp JA, Langeveld M, Lehman A, de Lonlay P. The management and clinical outcomes of pregnancies in women with urea cycle disorders: A review of the literature and results of an international survey. Journal of Inherited Metabolic Disease. 2023 Dec 9.