“Sweet Smelling” urine in pregnancy: First reported case in East Coast Malaysia with successful outcome

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ABSTRACT

Introduction: Maple Syrup Urine Disease (MSUD) is a rare autosomal recessive genetic disorder caused by a deficiency of enzymes responsible for breaking down the branched-chain amino acids leucine, isoleucine, and valine (BCAAs). This results in the accumulation of these amino acids in the body, leading to a characteristic sweet-smelling urine and a toxic build-up in the blood, which can cause severe metabolic complications. Management includes emergency treatment, lifelong dietary restrictions under expert supervision, nutritional supplementation, and, in severe cases, a liver transplant to restore enzyme function. **Case Description:** A young patient, G1P0 at 36 weeks and 2 days, with a history of MSUD, well-controlled epilepsy, and intellectual disability, presented with PPROM and a non-reassuring CTG. She had a late antenatal booking at 27 weeks and was under the care of both genetics and high-risk pregnancy clinics. The foetus was SGA with normal Doppler studies. Based on the clinical findings, an emergency LSCS was performed. During postpartum, the patient was closely monitored in the high-dependency unit, with strict nutritional management to prevent metabolic decompensation. Both mother and baby remained stable and were discharged in good condition, with a follow-up appointment at the genetics clinic in four weeks. **Discussion:** MSUD is typically diagnosed through newborn screening and managed with a strict diet to limit BCAA intake. However, pregnancy in women with MSUD presents additional challenges due to increased metabolic demands, particularly in the third trimester, making it more difficult to maintain stable BCAA levels. Pregnant women with Maple MSUD are at an increased risk of metabolic crises, which can lead to symptoms such as lethargy, vomiting, and neurological deterioration. If left untreated, these complications may result in maternal death or long-term disability. PPC is crucial to ensure optimal amino acid control before pregnancy, particularly to prevent elevated leucine levels, which have been associated with congenital abnormalities and poor foetal outcomes. Fortunately, our patient maintained well-controlled amino acid levels throughout her pregnancy, reducing the risk of complications. Elevated BCAAs in the maternal bloodstream can cross the placenta, potentially leading to neurodevelopmental complications in the foetus, including cognitive impairment and developmental delays. Additionally, poorly controlled MSUD in pregnant women is associated with an increased risk of foetal growth restriction and congenital malformations, such as congenital heart defects. With careful management, neonatal outcomes can be significantly improved. However, affected infants may still require specialised care after birth to prevent metabolic crises and ensure stable amino acid levels.