A Rare Genetic Disorder With Recurrent Hypoglycemic Episodes and Ketonemia Requiring Multidisciplinary Team Management During Pregnancy

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Introduction: Beckwith-Wiedemann syndrome (BWS; OMIM 130650) is a multisystem human genomic imprinting disorder of chromosome 11p15.5. It is characterized by overgrowth and cancer susceptibility and is known to have a highly variable clinical spectrum. The main features include lateralized overgrowth and hypoglycemia due to prolonged hyperinsulinism. Most case reports on BWS are focused on the pediatric population with very scarce data in adults.

Clinical case: A 29-year-old, paralympic swimmer and genetically diagnosed female with BWS had recurrent episodes of hypoglycemia during her second pregnancy. She had a history of six previous first trimester miscarriages, anti-phospholipid syndrome, hypoglycaemic seizures, asthma, and migraines. Pre-pregnancy, she was investigated for a neurological cause of her seizures and an electroencephalogram and brain imaging were unremarkable. She underwent extensive investigations for evaluation of hypoglycemia and was diagnosed as having hyperinsulinemia secondary to pancreatic hypertrophy. Factitious hypoglycemia was also ruled out. Tumor localization studies were all negative. A supervised fast could not be conducted due to the inability of the patient to tolerate the test. Her first viable pregnancy was 2 years back, during which she required a prolonged hospital (including the intensive care unit) admission with multiple in-hospital complications and requiring enteral feeding. During her current pregnancy, she experienced recurrent episodes of severe disabling hypoglycemia resulting in seizures. She was commenced on diazoxide, however, despite that, she continued to have hypoglycemic episodes for which she had to self-inject Glucagon several times. She also had unexplained weight loss with ketonemia undeterred by a high-caloric diet. She refused to be on naso-jejunal feeding or enteral feeding. She self-funded a flash glucose monitor which recorded her blood glucose levels between 2.0 to 3.4 mmol/L during the episodes of hypoglycemia. Dietitians were continually involved in her care to provide nutritional support and to optimize blood glucose control through dietary manipulation.

Conclusion: This was a very interesting and complicated case of a genetic disorder that presented multiple challenges during pregnancy. Management focused on psychological concerns, the severe catabolic state leading to rapid weight loss, recurrent hypoglycemic episodes, ketonemia, nutritional support, and fetal well-being monitoring. A multi-disciplinary team approach to management involved the obstetrics, endocrinology, dietetics, and gastroenterology teams. This resulted in a successful outcome for her pregnancy. Very limited data is available on health issues associated with BWS in adulthood and there is no data available on the obstetric management of women with BWS. Additionally, the unexplained weight loss and ketonemia associated with hypoglycemia make this case unique. Further studies are warranted and national guidelines are required to ensure that women with BWS achieve optimum pregnancy outcomes.

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