



Case Report

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Gitelman Syndrome in Maternity Ward an Uncommon Entity

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Abstract

Gitelman syndrome (GS), familial hypokalaemia-hypomagnesemia, is an autosomal recessive salt-losing renal tubulopathy that is characterised by hypomagnesemia, hypocalciuric and secondary aldosteronism, which is responsible hypokalaemia and metabolic alkalosis. The prevalence is estimated at 25 million [1].

In the majority of cases, symptoms do not appear before the age of six years and the disease is usually diagnosed during adolescence or adulthood. Transient periods of muscle weakness and tetany, sometimes accompanied by abdominal pain, vomiting and fever are often seen in GS patients. Paraesthesia, especially in the face, frequently occur. Chondrocalcinosis can be present in some patient.

GS is transmitted as an autosomal recessive trait. Mutations in the solute carrier family12, member 3 gene, SLC12A3, which encodes the thiazide sensitive NaCl cotransporter (NCC) is present. At present, more than 140 different NCC mutations exist. mutations in the CLCNKB gene, encoding the chloride channel ClC-Kb have been identified [2].

Diagnosis is based on the clinical symptoms and biochemical abnormalities (hypokalaemia, metabolic alkalosis, hypomagnesemia and hypercalciuria) [3].

Reaching to diagnosis was difficult, as it was not thought but was due to persistent hypokalaemia in patient, further analysis was done, then Gitelman Syndrome was diagnosed.

Keywords: Gitelman Syndrome, hypokalaemia, hypocalciuria, hypomagnesemia

Case Summary

21 years old primi gravida admitted at around 34 weeks with severe anaemia with gestational hypertension. Antihypertensive were started and blood transfusion was given. In spite of giving anti-hypertensive, blood pressure was not controlled. Her renal function was progressively deteriorating. So, emergency caesarean section was done under spinal analgesia. There were no Intraoperative problem and female child was born. Baby was taken into neonatal intensive care unit in view of low birth weight and preterm birth. Post operatively anti hypertensives were given.

Her hypokalaemia persisted in spite of potassium supplements. On day 11 post caesarean section, pt. developed tingling sensation & weakness in both distal extremities of lower limb and upper limb and positive Trousseau sign was present, suggestive of tetany. On work up she had hypocalcaemia, hypokalaemia, hypocalciuric, hypophosphatemia and electrocardiogram were suggestive of hypokalaemia changes and metabolic alkalosis. Based on association of hypokalaemia, hypomagnesemia, hypophosphatemia, hypocalciuric and metabolic alkalosis the diagnosis of Gitelman Syndrome was established.

Comments

GS is often not diagnosed until late childhood or even adulthood. Clinical manifestations are similar to the prolonged administration of thiazide diuretics like of a salt-losing renal tubulopathy that causes hypokalaemia and metabolic alkalosis [4,5]. To sum up, pregnancy with Gitelman syndrome presents with challenges of electrolyte imbalance, which may require a multidisciplinary approach with obstetricians, endocrinologists, anaesthetists, neonatologists and geneticists for good obstetric and neonatal outcomes.

Acknowledgement

None.

Conflicts of Interest

No Conflicts of Interest.

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