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GITELMAN SYNDROME- RECURRENT HYPOKALEMIC PARALYSIS IN LATE MIDDLE AGE

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ABSTRACT

Hypokalemia is a very common electrolyte disturbance noticed in the hospitals. Hypokalemic paralysis is a less recognized, reversible cause of profound hypokalemia. Gitelman syndrome- one of the causes for renal loss of potassium is an autosomal recessive, salt losing nephropathy, also known as familial hypokalemia hypomagnesemia. It is defined by metabolic alkalosis, hypomagnesemia, and low urinary calcium excretion. With defect in Na- Cl cotransport in distal tubules, it is one of the most common inherited renal tubulopathies.[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.[2] In this case report, we present a 48-year-old male patient who was admitted to our hospital with symptoms of muscle weakness, cramps and numbness, on evaluation, diagnosed as Gitelman Syndrome.

CASE REPORT

We present the case of a 48-year-old gentleman, who presented with quadriparesis, perioral numbness, intermittent joint stiffness, present for the past 1 week. History of sudden onset of breathlessness, which brought the patient to our emergency room at Saveetha medical college and hospital. These complaints along with history of polyuria and nocturia have been present for the past 10 years, requiring multiple hospital visits, records of which were not available. Born to healthy parents of non-consanguineous marriage, with 2 other male siblings of the age 55 and 52 years, respectively, who also had similar complaints of quadriparesis and joint stiffness, on and off, were informed of some kidney disease and are on treatment for the same; details of which weren't available. On physical examination, he had a BP of 90/60mmHg, with signs of moderate dehydration, was anxious, with tachycardia and tachypnoeic with oxygen saturation of 98% at room air, with respiratory rate of 28 cycles per minute.

Trousseau sign and Chvostek sign were positive, cardiopulmonary examination was normal. On blood gas analysis, metabolic alkalosis was observed (pH-7.526, pCo2-29.2, HCO3- 24.3). Biochemical analysis revealed hypokalaemia- 2.5mEq/l (3.5 to 4.5mEq/l), hypomagnesemia-0.5mg/dl (1.3 to 2.1mEq/l),Serum Ca^{2+}- 7.3mg/dl (8.5 to 10.5 mg/dl) , Serum Urea- 53mg/dl (6 to 40 mg/dl), Creatinine- 3mg/dl (0.34 to 1.21 mg/dl) , Serum osmolality-279mOsm/kg (275-295mOsm/kg), Urine osmolality- 353mOsm/kg (50-1200 mOsm/kg), Urine PCR- 0.098, Spot K^{+} 15.9 (25-125mEq/day), 24 hours urinary Ca^{2+} -93mg/dl (100-300mg/dl), Ca^{2+}/Cr ratio- 0.2. Liver function tests, complete hemogram were within the physiological range. Electrocardiogram showed Normal sinus rhythm, T wave inversion with prominent U waves. Renal ultrasound was normal.

The observation of hypokalemia, hypomagnesemia, hypocalciuria, metabolic alkalosis and low normal blood pressure, fit into the diagnosis of Gitelman syndrome.

TREATMENT

Patient was treated with Potassium, Magnesium, Calcium supplements, parenteral and oral, till dyselectrolytemia was corrected. Patient symptomatically improved, renal parameters normalised.

Was encouraged to consume high potassium and calcium diet. He remains symptom free and normokalemic after 6 months of follow up.

DISCUSSION

Gitelman syndrome is usually asymptomatic or presents with symptoms such as muscle weakness, fatigue, salt craving, thirst, nocturia, constipation,vomiting, cramps, and carpopedal spasm or tetany episodes triggered by hypomagnesemia and hypokalemia.Our patient presented with complaints of perioral numbness, tingling sensation and weakness of all the 4 limbs,and intermittent joint stiffness in the form of carpopedal spasm. Gitelman syndrome is a biallelic inactivating mutation of the *SLC12A3* gene, located on chromosome 16, encoding for thiazide-sensitive sodium chloride cotransporter (NCCT), located in the apical membrane of the distal convoluted tubule (DCT). [3,4]

The differential diagnosis suspected clinically before obtaining ABG and other investigations were, channelopathies, myopathies, dyselectrolytemia. Ultrasound of the kidneys was done to rule out , defects of the urinary tract, identify kidney casts, nephrocalcinosis, and nephrolithiasis. Bartter syndrome is the most important genetic disorder to consider in the differential diagnosis of Gitelman syndrome, but the presentation in adult, with low magnesium levels, and hypocalciuria, made Bartter syndrome improbable.

Gitelman syndrome mimics the symptoms of usage of thiazide diuretics, while Bartter's syndrome, that of loop diuretics.[5] However in both the syndromes patients present with hypokalemia, hypomagnesemia and metabolic alkalosis. The distinguishing feature being presence of hypocalciuria in Gitelman syndrome.[6,7]

DNA mutation analysis is the gold standard for diagnosis.[8] Our patient refused genetic workup. Through this case report we would like to emphasis, that electrolyte imbalance should be considered in patients presenting with muscle weakness and fatigue, and irrespective of the age of presentation, it is important to remember salt losing tubulopathies as differentials for recurrent dyselectrolytemia. Although, genetic analysis was not performed in our case, the diagnosis was achieved through clinical and laboratory testing, as suggested by some case reports from the literature.[9,10]

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