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CASE REPORT

Adult Gitelman Syndrome: Case Report and Review of the Literature

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ABSTRACT

Gitelman Syndrome, the most frequently detected hereditary tubulopathy in adults, was first described by Gitelman et al. in 1966 in 3 adults who presented with tetany associated with hypokalemic metabolic alkalosis, hypomagnesemia, and hypocalciuria. The estimated prevalence of Gitelman syndrome, which is considered as a variant of Bartter syndrome with autosomal recessive inheritance, is reported as 1:40,000. In this article, a case diagnosed with Gitelman Syndrome, who was hospitalized many times, including the intensive care unit, with symptoms such as constipation, weakness, nausea and vomiting since childhood is presented.

One of the most prevalent inherited renal tubular diseases is known as Gitelman syndrome (GS), which is brought on by mutations that inactivates located on chromosome 16(16q13) in the SLC12A3 gene, which codes for the sodium chloride cotransporter (NCC) that is expressed in the apical membrane of distal convoluted tubule (DCT) cells and is thiazide sensitive. Gitelman variant of Bartter syndrome is seen in adolescence and adulthood and often has a milder clinical course than Bartter Syndrome. Although the cases are generally asymptomatic, symptoms such as fatigue, muscle weakness, cramps, tetany, fatigue, nocturia and polydipsia are common. Polyuria and nocturia are common due to nephrogenic diabetes insipidus due to hypokalemia. Growth retardation is also common in these cases.

CASE REPORT

A 28-year-old female patient was hospitalized for further examination and evaluation after her application to the emergency department due to increased complaints of fatigue, nausea and vomiting, which she stated for about 10 years. The patient, who did not have any known additional disease and no medication that she used constantly, stated that she had constipation and oral intake disorder throughout her life, and that her vomiting had increased especially in the last 4-5 days. The patient, who stated that he had a history of intensive care hospitalization 3 months ago with similar complaints, had sinus rhythm in electrocardiography, his QT interval was normal, and no significant U wave was observed. In the renal ultrasound, the parenchymal

echo was reported as bilateral grade I-II increased, while the parenchymal echo was normal in the thyroid ultrasound, no nodules and pathological lymphadenopathy were observed.

In laboratory evaluation, Ca = 8.0 mg/dL, p = 1.9 mg/dL, K = 1.8 mg/dL, Na = 128 mg/dL, Creatinine = 0.46 mg/dL, Mg = 1.52 mg/ dL was observed as Parathormone (PTH) = 32.3 mU/L, while pH = 7.58, pCO2 = 71.9 and HCO3 = 67.9 in blood gas. Autoimmune markers p-anca, c-anca and Anti-dsDNA tests, and hormone levels such as Cortisol, FSH, LH, prolactin, Anti-TPO, Anti-TG and Thyroglobulin were reported within normal limits. 24-hour urine Na = 27.6 mmol, Ca= 47.64 mg, and Cl = 20.39 mmol.

In the case, the patient was diagnosed with Gitelman Syndrome, in spite of the existing cachexia, with normal breast development, genital hair growth, no growth retardation, the characteristic phenotype of Bartter Syndrome (triangular face, large eyes), and the dramatic improvement of his complaints with the potassium and magnesium treatment given. After IV replacements, oral magnesium potassium preparations and PPI were prescribed, and she was discharged.

DISCUSSION

The diagnosis of Gitelman syndrome is mainly based on clinical, biochemical and molecular findings. The disease is often confused with Bartter syndrome. Gitelman syndrome is distinguished from Bartter syndrome by the milder clinical presentation, absence of polyuria, normal or slightly decreased tubular concentration ability, decreased urinary calcium excretion, decreased serum magnesium, and absence of maternal polyhydramnios or prematuration [1-3]. Although chondrocalcinosis is rarely seen in Gitelman syndrome, it is not seen in Bartter syndrome. In addition, rare arrhythmias due to electrolyte imbalance may occur. Chronic hypomagnesemia has been blamed for the development of chondrocalcinosis in a small number of patients. Persistent hypomagnesemia suppresses PTH secretion and may cause chondrocalcinosis by impairing the function of enzymes such as alkaline phosphatase, which regulates pyrophosphate concentration in the extracellular space [4].

Progression to chronic renal disease is rare and few cases have been reported so far. Blood pressure is usually normal. Urinary potassium and magnesium excretion is decreased. Urinary calcium is usually below 2 mg/kg. Renal functions, urinary PgE2 and cAMP are normal. Thiazide diuretics similarly suppress the Na/Cl cotransporter in the distal tube. All findings seen with long-term thiazide use are similar to those found in Gitelman's Syndrome [5]. However, hypomagnesemia, which is an important finding in Gitelman Syndrome, is rarely seen in thiazide use. Despite all clinical and laboratory findings, it is often difficult to diagnose and differentially diagnose Bartter Syndrome and Gitelman Syndrome. Molecular genetic studies have made significant progress in the differential diagnosis of these syndromes [1].

Differential diagnosis

In the differential diagnosis, especially Bartter syndrome (type 3), use of diuretics and laxatives, and chronic vomiting should be considered [1]. Rarely, cisplatin use and autoimmune diseases may also cause Gitelman syndrome-like findings. Diuretic and laxative use, chronic vomiting can be detected with a careful history taking and physical examination.

Treatment

The cases are usually asymptomatic and do not require treatment. The long-term prognosis is good. The goal of treatment is to correct electrolyte abnormalities and symptoms [6]. In these patients, it is possible to improve the quality of life with magnesium replacement. With regular magnesium treatment, hypomagnesemia, hypokalemia and hypocalciuria improve, tetany and acid-base imbalance are prevented. Magnesium chloride, magnesium aspartate or magnesium lactate salts are divided into 3-4 parts per day and given in a total dose of 4-5 mg/kg/day. Hypomagnesemia may occur as a result of urinary potassium loss. Potassium requirement may be as high as 10 mg/kg/day [5]. In general, MgCl is preferred because of chlorine loss. A high salt diet is recommended. The tendency to chondrocalcinosis is also controlled with magnesium therapy. Diarrhea is the most common side effect of magnesium replacement. Potassium and prostaglandin suppressors are generally not needed, but some patients may require potassium replacement. Aldosterone antagonists (amiloride and spironolactone) can be used to correct the serum potassium level. In potassium deficiency, hypokalemia may be resistant to replacement with potassium salts, in which case it is important to replace magnesium first [5, 6].

CONCLUSION

Patients with fatigue, weakness, muscle pain and cramps should be evaluated for electrolyte imbalance, and patients with hypomagnesemia and hypocalciuria should be investigated for Gittelman Syndrome.

Authorship Contributions

Conception: DSC., NK., F.İ. Design: DSC., F.i., N.K. Supervision: NK Funding: None, Data Collection or Processing: DSC., Fİ. Analysis or Interpretation: DSC., F.İ., N.K. Literature Review: DSC., F.İ, N.K., Writer: DSC., N.K. Critical Review: N.K.

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Conflict of Interest

No conflict of interest was declared by the authors.

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