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Clinical Case of Successful Correction of Blood Electrolytes and Acid-Base Balance in Gitelman Syndrome

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Authors' contributions

This work was carried out in collaboration among all authors. Authors SD, SK and YK conceived the study and were involved in patient therapy monitoring and analysis. Authors SK and MS performed the literature search. Authors YK and SD drafted the manuscript. Authors MS and SK reviewed the manuscript and did the necessary changes. All authors read and approved the final manuscript.

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Case Report

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ABSTRACT

Gitelman Syndrome (GS) is an autosomal-recessive disorder distinguished by hypokalemia, hypomagnesaemia, and hypocalciuria. Elderly people and women of childbearing age are highly affected by GC. There isn't much evidence known about its effects on maternal and fetal outcomes. There is a high incidence of hypotension and unexpected cardiac arrest. Normal growth is generally seen in GS patients, but growth may be delayed in severe hypokalemia and hypomagnesemia. Surprisingly, some patients are asymptomatic at all, with the exception of chondrocalcinosis, which manifests in adults and causes swelling, localized heat, and joint tenderness. GS is caused by mutations in the thiazide-sensitive Na-Cl cotransporter gene. Due to

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its rarity and lack of knowledge, It is susceptible to misdiagnosis or being overlooked. In this case, the patient was suffering from recurrent hypokalemia, hypomagnesemia, hypochloraemia, and hypocalciuria with hypotension. After taking proper medication patients recovered slowly and during patient counseling provided diet chart by nutritionists to avoid recurrent electrolyte imbalance.

Keywords: Hypokalemia; hypochloraemia; hypocalciuria; hypotension.

1. INTRODUCTION

Gitelman syndrome (GS) is a rare autosomal-recessive disorder, first described in 1966 by Gitelman et al. GS also known as Gitelman's version of Bartter's Syndrome (BS), is a hereditary tubulopathy which considers salt-losing that runs in the autosomal-recessive family [1]. Estimated data from several genomic databases, GS affects around 1 in 40,000 individuals, despite the fact that Asians may be more prone to it [2]. Diagnosis of GS, on the basis of laboratory value characterized by hypokalemia, hypomagnesemia, metabolic alkalosis, and hypocalciuria [3]. Low serum potassium level is the characteristic of hypokalemia. Hypokalemia mainly represents electrolyte imbalance. When potassium levels are below 2.5 mEq/L than severe, life-threatening neurological and cardiac problems may appear. It can be due to various causes. Hypokalemia cannot be corrected just by lowering potassium levels, which may lead to recurrence and severe complications [4]. Although BS (Barter's syndrome) and GS (Gitelman's syndrome) share several phenotypic characteristics, GS can be distinguished by having hypomagnesemia and hypocalciuria. Hypomagnesemia may be present, and hypocalciuria is exceedingly variable. It occurred due to the presence of an inactivating mutation in the gene SLC12A3, on chromosome 16, which codes for the thiazide-sensitive sodium-chloride cotransporter and the magnesium transporter on the apical membrane of the Distal Convoluted Tubule (DCT). GS is a benign tubulopathy with the symptoms are mild weakness, weariness, salt seeking, thirst, and nocturia to muscle weakness, paralysis, paresthesias, or the symptoms of neuromuscular excitability, such as tetany, and infrequent seizures but some time it may have no symptom. A few GS patients may be at risk for cardiac arrhythmias, but overall, the prognosis of patients with this syndrome is excellent. Though, some patient's daily activities may be negatively impacted by the severity of fatigue. It is quite uncommon for this syndrome to deteriorate and progress to renal insufficiency in GS [5].

2. CASE PRESENTATION

This is a case of 55-year-old male with recurrent hypokalemia, admitted to the general medicine department. The patient presented with complaints of muscle weakness, dizziness and low blood pressure of 108/72, systolic blood pressure was always below 110 mmHg with positive anamnesis, and no further symptoms of the urinary tract, he only mentioned polyuria and nocturia. His medical history was not significant with negative family history. He denied using any drugs other than vitamin supplements intake. When the patient was being treated in a hospital for community-acquired pneumonia at the age of 53, this illness was first discovered. Although the last six months he had been taking oral magnesium and potassium supplements inconsistently. During the physical examination, he showed normal hydration, skin and mucosa coloration, a blood pressure of 108/68 mm Hg, and a regular pulse frequency of 80 beats/min. The cardiopulmonary assessment was normal with no evidence of peripheral edema. The remaining physical checkup was normal. A metabolic alkalosis was presented during the gas analysis (pH 7.435; HCO3 33.1 mmol/l; PaCO2 46 mm Hg)and bradypnea present with 11 breaths/min (NR <12 breaths/min). In biochemical analysis, the report confirm hypokalemia (2.42 mmol/l), hypomagnesemia (0.35 mmol/l), and hypochloraemia (89 mmol/l). urea (58mg/dl), Serum creatinine (2.4 gm/dl), serum sodium level 123.4 mmol/l and remainder ionogram was normal. Further investigation found raised plasma-active renin (2719 fmol/l/sec), with normal aldosteronaemia (16.7 ng/ml in orthostatic; NR 4–31), hypocalciuria (0.73 mmol/l; NR 2.5–7.5) and a greater amount of urine excretion of sodium (126.5 mmol/l; NR 20-110) and chloride (166 mmol/l; 55-125). The estimated glomerular filtrate rate (GFR) was 120 ml/min/1.73 m2 and the potassium transtubular gradient was 11.6 and other tests like Renal ultrasonography, and renal and adrenal CT indicate normal kidney function. A correlation between hypokalemia, hypomagnesemia, metabolic alkalosis, hypocalciuria, and low blood pressure led to the diagnosis of GS.

2.1 Treatment

The patient took medication according to the prescribed manner, magnesium aspartate 1230 mg once a day (OD), Tolvaptan tablet 15 mg once a day (OD), potassium chloride capsule 600 mg twice daily (BD), and started taking magnesium aspartate/potassium aspartate 250/250 mg orally four times per day (qid) from admission time. During counseling, The patient was advised to keep up a high-sodium, high-potassium along with balance protein diet by nutritionists. 20 days later patient's serum potassium levels of 2.9 mmol/l and magnesium levels of 0.52 mmol/l, serum sodium level 134.5, serum chloride 92 mmol/l and serum creatinine 1.9 gm/dl. The patient recovered slowly with the proper care of the healthcare team.

3. DISCUSSION

A frequent clinical issue with potentially fatal symptoms is chronic hypokalemia. long-term symptomatic hypotension, hypomagnesaemia, and metabolic alkalosis. Our patient is affected by these conditions. The two main diagnoses in this situation—vomiting and diuretic abuse—were ruled out by measuring high urinary chloride excretion and having no history of using diuretics, respectively. The genetic syndromes Gitelman and Bartter were the only remaining differential diagnoses. Due to its more severe phenotype, earlier start, frequent rise in urinary calcium excretion, and normal or modestly hypomagnesaemia, Bartter syndrome is considered implausible. On the basis of laboratory test reports final diagnosis was GS. It is an autosomal recessive renal tubulopathy that causes salt loss. Sodium-chloride cotransporter sensitive to thiazides, which is found in the epithelial cells of the renal distal convoluted tubule (DCT), and it is encoded by the gene in the vast majority of instances [6]. Its distinguishing characteristics, secondary hyperaldosteronism, hypomagnesaemia, and hypocalciuria, cause hypokalemia and metabolic alkalosis. Hypomagnesemia can inhibit parathyroid hormone secretion [7]. The clinical signs are the same as when thiazide diuretics are used for a long time [8]. Clinical signs and biochemical abnormalities, such as hypomagnesaemia, hypokalemia, metabolic alkalosis, and hypocalciuria, are used to make the diagnosis of GS. This type of patient responds to thiazide with a blunted natriuretic response, but responds quickly to furosemide, indicating that the abnormality is at the level of the distal tubule. Analysis of the GS gene's DNA for mutations may help to confirm the diagnosis [9]. According to global consensus suggestion, blood potassium and magnesium levels should be maintained at or above 3.0 mmol/L and > 0.6 mmol/L, respectively in GS patients [10-11]. Most asymptomatic patients do not receive treatment, and they only sometimes have ambulatory monitoring. Renal insufficiency can progress but it is incredibly uncommon [12].

Table 1. During the initial presentation, biochemical parameters

Test	Value	Normal range
Serum potassium, mmol/l	2.42	3.6–5.4
Serum magnesium, mmol/l	0.35	0.7–1.0
Serum Sodium level, mmol/l	123.4	135–145
Blood urea, mg/dl	58	10–50
Blood calcium level mmol/l	0.73	2.5-7.5
Serum creatinine gm/dl	2.4	0.6–1.6
Plasma renin activity, fmol/l/s	2,719	100–1,500
Serum Chloride mmol/l	89	96-108
Urine Sodium mmol/l	126.5	20-110
Urine Chloride mmol/l	166	55-125

Table 2. Biochemical parameters after 20 days of treatment

Test	Value	Normal range
Serum potassium, mmol/l	2.9	3.6–5.4
Serum magnesium, mmol/l	0.52	0.7–1.0
Serum Sodium level, mmol/l	134.5	135–145
Serum chloride mmol/l	92	96-108
Serum creatinine gm/dl	1.9	0.6–1.6

4. CONCLUSION

This case report features a patient who diagnosed with GS symptoms of hypomagnesemia, persistent hypokalemia, and metabolic alkalosis. Inconsistently, patient was taking oral magnesium and potassium supplements over the previous six months. Frequent rise in urinary sodium and chloride excretion, and normal or mild hypomagnesaemia is considered implausible. When thiazide diuretics are used for a prolonged period of time, then the clinical symptoms are the same. During patient counseling provided diet chart by nutritionists to avoid recurrent electrolyte imbalance. After 20 days, his serum potassium and magnesium levels were 2.9 mmol/l and 0.52 mmol/l, respectively. Administration of ionic supplements improved the patient's quality of life by eliminating basic blood electrolyte deficiencies and correcting acid-base balance indices.

CONSENT

Written informed consent was obtained from the patient for the publication of this case report.

ETHICAL APPROVAL

As per international standards or university standards written ethical approval has been collected and preserved by the author(s).

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COMPETING INTERESTS

Authors have declared that no competing interests exist.

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