Case Report

Intensive care management of Gitelman syndrome: A rare case

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ABSTRACT

Gitelman syndrome (GS) is an autosomal-recessive disorder distinguished by hypokalemia, hypomagnesemia, and hypocalciuria. Elderly people and women of childbearing age are highly affected by GC. Not much evidence is known about its effects on maternal and fetal outcomes. GS is caused by mutations in the thiazide-sensitive Na-Cl cotransporter gene. Due to its rarity and lack of knowledge, it is susceptible to misdiagnosis or being overlooked. In our case, the patient suffered from recurrent hypokalemia, hypomagnesemia, hypoclorhaemia, and hypocalciuria with hypotension. After taking proper medication, the patient recovered slowly, and during counseling, the patient was provided a diet chart by nutritionists to avoid recurrent electrolyte imbalances.

Key words: Hypocalciuria, Hypochloremia, Hypokalemia, Hypotension

CASE REPORT

A 55-year-old male with recurrent hypokalemia was admitted to the general medicine department with complaints of muscle weakness, dizziness, low blood pressure of 108/72 mmHg (systolic blood pressure was always below 110 mmHg with positive anamnesis), polyuria, and nocturia. His medical history was not significant, and he had a 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, for the last 6 months, he has 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 mmHg, 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; pCO₂ 38 mmHg). In biochemical analysis, the report confirmed hypokalemia (2.42 mmol/L), hypomagnesemia (0.35 mmol/L), and hypochloremia (89 mmol/L). Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal. Further investigation found raised plasma-active renin (2719 fmol/L/s), normal aldosteronemia (89 mmol/L), Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal. Further investigation found raised plasma-active renin (2719 fmol/L/s), normal aldosteronemia (89 mmol/L), Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal. Further investigation found raised plasma-active renin (2719 fmol/L/s), normal aldosteronemia (89 mmol/L), Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal. Further investigation found raised plasma-active renin (2719 fmol/L/s), normal aldosteronemia (89 mmol/L), Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal. Further investigation found raised plasma-active renin (2719 fmol/L/s), normal aldosteronemia (89 mmol/L), Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal. Further investigation found raised plasma-active renin (2719 fmol/L/s), normal aldosteronemia (89 mmol/L), Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal. Further investigation found raised plasma-active renin (2719 fmol/L/s), normal aldosteronemia (89 mmol/L), Urea (58 mg/dL), Serum creatinine (2.8 gm/dL), serum sodium level (123.4 mmol/L), and remaining ionogram were normal.
The estimated glomerular filtrate rate was 120 mL/min/1.73 m², and the potassium transtubular gradient was 11.6. Other tests such as renal ultrasonography and renal and adrenal CT indicate normal kidney function (Table 1). A correlation between hypokalemia, hypomagnesemia, metabolic alkalosis, hypocalciuria, and low blood pressure led to the diagnosis of GS.

The patient took medication according to the prescribed manner, which includes magnesium aspartate 1230 mg once a day (OD), Tolvaptan tablet 15 mg OD, potassium chloride capsule 600 mg twice daily (BD), and started taking magnesium aspartate or 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 and high-potassium diet by nutritionists. 20 days later, the patient’s serum potassium and magnesium levels were 2.9 mmol/L and 0.52 mmol/L, respectively. The patient recovered slowly with the proper care of the healthcare team.

DISCUSSION

A frequent clinical issue with potentially fatal symptoms is chronic hypokalemia, long-term symptomatic hypotension, hypomagnesemia, 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 of 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 modest hypomagnesemia, Bartter syndrome is considered implausible. On the basis of laboratory test reports, a final diagnosis of GS was made.

GS is an autosomal recessive renal tubulopathy that occurs due to the presence of an inactivating mutation in the gene SLC12A3. Since sodium-chloride cotransporter is sensitive to thiazides and found in the epithelial cells of renal DCT, it is encoded by the gene in the vast majority of instances [6]. Its distinguishing characteristics are secondary hyperaldosteronism, hypomagnesemia, and hypocalciuria, which cause hypokalemia and metabolic alkalosis. Hypomagnesemia can inhibit parathyroid hormone secretion [7].

Table 1: The biochemical parameters of the patient during initial presentation

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Value</th>
<th>Normal range</th>
</tr>
</thead>
<tbody>
<tr>
<td>Serum potassium, mmol/L</td>
<td>2.42</td>
<td>3.6–5.4</td>
</tr>
<tr>
<td>Serum magnesium, mmol/L</td>
<td>0.35</td>
<td>0.7–1.0</td>
</tr>
<tr>
<td>Serum sodium level, mmol/L</td>
<td>123.4</td>
<td>135–145</td>
</tr>
<tr>
<td>Blood urea, mg/dL</td>
<td>58</td>
<td>10–50</td>
</tr>
<tr>
<td>Blood calcium level mmol/L</td>
<td>0.73</td>
<td>2.5–7.5</td>
</tr>
<tr>
<td>Serum creatinine, g/dL</td>
<td>2.8</td>
<td>0.6–1.6</td>
</tr>
<tr>
<td>Plasma renin activity, fmol/L</td>
<td>2719</td>
<td>100–1,500</td>
</tr>
<tr>
<td>Serum chloride, mmol/L</td>
<td>89</td>
<td>96–108</td>
</tr>
<tr>
<td>Urine sodium, mmol/L</td>
<td>126.5</td>
<td>20–110</td>
</tr>
<tr>
<td>Urine chloride, mmol/L</td>
<td>166</td>
<td>55–125</td>
</tr>
</tbody>
</table>

The clinical signs are similar to those using thiazide diuretics for a long time [8]. Clinical signs and biochemical abnormalities, such as hypomagnesemia, 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 suggestions, 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].

CONCLUSION

This case report features a patient who has the usual GS symptoms of hypomagnesemia, persistent hypokalemia, and metabolic alkalosis. Inconsistently, the patient was taking oral magnesium and potassium supplements over the course of 6 months. A frequent rise in urinary sodium and chloride excretion and normal or mild hypomagnesemia are considered implausible. When thiazide diuretics are used for a prolonged period of time, the clinical symptoms are the same. During patient counseling, nutritionists provided a diet chart to patients 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. We strengthened ion supplementation, and the patient gradually achieved a full recovery.

ACKNOWLEDGMENT

The patient’s confidentiality was maintained. The Institutional Ethics Committee (IEC) of Institute for Medical Sciences and Research Centre, Jaipur National University, does not require IEC approval for this present case. All patient data and records were de-identified and anonymously collected before analysis.

REFERENCES


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