Rare Adult-Onset of Bartter Syndrome: A Case Report

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ABSTRACT

Background: Bartter syndrome is an autosomal recessive disorder of the renal system affecting the ascending part of the loop of Henle which causes hypochloremia, hypokalemia, and metabolic alkalosis. Clinical features consist of transient muscle paralysis, vomiting, paresis, polydipsia, constipation, and salt cravings. Bartter syndrome most commonly presents in early age, but scarce information is available about the long-term management, prognosis, and remission from the disease. Adult-onset cases of bartter syndrome are very rare. Better in-depth characterization of various electrolyte abnormalities, in patients of Bartter syndrome using a multitude of parameters, may help us to widen the horizon of our current knowledge and understanding of Bartter syndrome in adult patients.

Case report: Here we report a rare case of a 40-year-old adult male patient with Bartter syndrome presenting with signs and symptoms of persistent hypokalemia, extreme fatigue, and weakness. The patient was prescribed to take potassium chloride syrup 40 mEq/day and indomethacin 50 mg tablet twice a day, to reduce potassium loss.

Conclusion: Treatment with potassium chloride and indomethacin showed a significant reduction in muscle weakness.

Keywords: Acute hypokalemic paralysis, Bartter syndrome, Hypokalemia, Metabolic alkalosis.

INTRODUCTION

Bartter syndrome is a rare clinical condition leading to salt wasting. This is one of the infrequent causes of persistent hypokalemic metabolic alkalosis. Prevalence of Bartter syndrome is 1 in 1,000,000 which signifies that it is a significant barrier in reaching an accurate diagnosis and achieving therapeutic interventions.[1] It can be classified clinically and genetically with five variants of diversified phenotypes.[2] It has five different subtypes according to genetic abnormalities with type III having a defect in chloride protein channel causing decreased chloride efflux and altering the transepithelial voltage gradient which decreases Na-k-Cl resorption.[3] It is the most common subtype and clinically being the most difficult to distinguish it from Gittleman syndrome, another disease of impaired tubulopathy. There is hyperplasia of juxtaglomerular complex, elevated levels of renin and aldosterone with
subsequent exaggerated potassium loss and metabolic alkalosis. This causes activation of renin angiotensin aldosterone system causing hypokalaemia which is considered a primary cause of prostaglandins, kallikrein, and bradykinin abnormalities. The intimate relationship between excessive prostaglandins as signalling molecules and resulting salt wasting is the mainstay in the pathogenesis of Bartter Syndrome. It is known to commonly occur in early childhood age or adolescence. However, adult-onset of the condition is very rare and mainly due to chronic hypokalaemia alkalosis. The rarity of the disease manifesting in an adult itself raises the doubt of reaching a diagnosis along with the number of undiagnosed infants progressing to adulthood with no clinical manifestations or any serious illness precipitating it. Furthermore, due to a lack of affordability and advancement in the tertiary care hospital, no genetic studies are usually performed in these patients. The most devastating complication of hypokalaemia is its wide range of cardiovascular abnormalities predisposing the patient to various arrhythmias and sudden cardiac death. This makes it more imperative for the physicians or health care providers to understand the condition better to achieve an early diagnosis of Bartter syndrome and initiation of treatment.

**CASE REPORT**

A 40-year-old man visited out-patient ward of the Sir Sayajirao General (SSG) Hospital, Vadodara for routine health follow check-up and presented the chief complaint of extreme fatigue and weakness during the check-up. The patient experienced an episode of acute paralysis in both arms and legs in July 2019 and spontaneously emitted from it with appropriate bed rest and fluid intake. The patient’s first episode was when he was 17 years old and subsequently was in remission without visiting the hospital. He denied any ingestion of alcohol, smoking, diuretics, laxative, liquorice, or any other medications. The patient frequently complained about weakness and fatigue with episodic acute paralysis. His urine frequency was 8-9 times in the day and 4-5 times in the night with symptoms of excessive thirst and salt cravings. The patient complained of frequent vomiting since last month and has experienced significant weight loss with no prior family history of the condition or hospitalizations history in the past. The physical condition, appearance, and vitals of the patient were within normal limits. Analysis of laboratory findings showed persistent hypokalaemia with metabolic alkalosis. A comparative analysis of the laboratory findings of the patient in the last two years is presented in (Figure 1) below.
Figure 1: Comparative analysis of the laboratory findings of the patient in the last two years.

The results of arterial blood gas analysis performed on admission are provided in Table 1 below.

Table 1: Results of Arterial Blood Gas Analysis Performed on Admission

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>pH</td>
<td>7.48</td>
</tr>
<tr>
<td>Partial pressure of CO₂ (PaCO₂)</td>
<td>46.8 mmHg</td>
</tr>
<tr>
<td>Partial pressure of O₂ (PaO₂)</td>
<td>88 mmHg</td>
</tr>
<tr>
<td>Oxygen saturation</td>
<td>98%</td>
</tr>
<tr>
<td>Serum bicarbonate</td>
<td>33.5 mEq/L</td>
</tr>
<tr>
<td>Base deficit</td>
<td>2 mEq/L</td>
</tr>
</tbody>
</table>
Electrocardiography studies showed characteristic changes of hypokalaemia such as prolonged PR, flattened T wave, and prominent U waves. Urinary specific gravity was 1.010 and urine osmolality was 212.5 mOsm/kg with serum osmolality being 292.19 mOsm/kg.

His urinary calcium/creatinine ratio was more than 0.2, with urine calcium levels 8.6 g/dL and urine creatinine 11.26 mg/dL. Due to high cost, plasma renin and aldosterone were not tested. The patient’s parathyroid levels, thyroid levels, and hemogram studies were within normal limits. There was no evidence of proteinuria, haematuria, or any urinary sediments. Computer Tomography (CT) scan findings showed no evidence of any focal abnormalities. Few calculi of average size (3-4mm) were found at the poles of right and left kidneys, with mild fullness of the pelvicalyceal system on the left side. Ultrasonography (USG) findings showed no abnormalities in the liver, spleen, and pancreas. Magnetic Resonance Imaging (MRI) of brain and spine showed no abnormalities. The patient was advised to take high potassium diet and drink coconut water 3-4 times a day to restore electrolyte loss from time to time. He was also prescribed with potassium chloride syrup 40 mEq/day and indomethacin 50 mg tablet twice a day, to reduce potassium loss. Treatment with potassium chloride and indomethacin showed significant reduction in muscle weakness. Complete normalization of potassium levels was not aimed along with lack of data on the universal realistic target value. After 2 years of treatment, no side effects of indomethacin were observed, and further follow-ups will be maintained. Compliance of the patient has been variable and was not maintained even though personal and family education was provided. Off-label use of indomethacin decreased the frequency of monthly acute hypokalaemia crisis to none. Magnesium levels were also monitored periodically for better assessment of the electrolyte abnormalities. Aim was to improve the general condition of the patient and to avoid any aggravating factors like diarrhoea and vomiting which resulted in hypokalaemia crisis. Overall patient’s potassium level was maintained above 3 mEq/L without any signs and symptoms of hypokalaemia, however significant weight loss of approximately 5 kg was noticed at the end of one year. The patient was advised to maintain regular follow-up check-ups of every 6 months to one year to assess the long-term outcome of the therapy.

**CONCLUSION**

In summary, we are reporting a case of a 40-year-old adult presenting with a severe episode of acute hypokalaemic paralysis on multiple occasions, accompanied by periods of recurrent vomiting, altered bowel, and bladder frequency, muscle weakness and generalized fatigue. Analysis of the the laboratory findings showed that the patient had persistent hypokalaemia with metabolic alkalosis and blood pressure within normal limits. Furthermore, the urine chloride was found to be high with no history of any diuretics or other medications in the past. CT scan findings of nephrocalcinosis suggested hypercalciuria differentiating it with Gitelman Syndrome. Based on the clinical and laboratory findings, the patient was diagnosed with Bartter syndrome and prescribed with potassium chloride syrup 40 mEq/day and indomethacin 50 mg tablet twice a day. Treatment with potassium chloride and indomethacin showed significant reduction in muscle weakness. The patient’s magnesium and potassium levels were monitored regularly, and the patient was advised for regular follow-up check-ups every 6 months to one year to assess the long-term outcome of the therapy.
The limited number of studies and the voids in the knowledge, need to be filled with more significant scientific evidence that would help provide the physicians or health care providers with a better understanding of the prognosis of Bartter syndrome which still tends to be a mystery.

Patient education is an important aspect to be stressed on, as with any lifelong chronic illness. Education about signs and symptoms necessitating a visit to the hospital such as palpitations, syncope, severe dehydration, tetany, paraesthesia, paralysis, excessive vomiting, etc. is imperative.

REFERENCES