

## Case Report

# Adult-Onset Bartter Syndrome: A Case Report

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**Abstract****Introduction**

Bartter syndrome is a rare genetically inherited salt-wasting disorder that is typically seen in children and neonates with association to many morbidities. We present a case of Bartter syndrome in an adult who showed excellent response to treatment.

**Case presentation**

The patient was a 23-year-old male presenting with polyuria, polydipsia, nocturia, and fatigue, especially within the lower limbs for the last two years but no history of vomiting. He was clinically diagnosed with Bartter syndrome and received conservative management with a good response.

**Conclusion**

Bartter syndrome is a rare disease. It has a wide range of clinical presentations. It can be diagnosed clinically and confirmed by genetic testing. Conservative management has a good clinical outcome.

**1. Introduction**

Bartter syndrome (BS) is a rare disease of the renal tubules which consequently results in decreased reabsorption of salt (1). There are two different types of classification for the disease which are either phenotypic or genotypic. Phenotypically, the disease is divided into either neonatal(antenatal) or classical BS

which presents later in life. Genotypically, the disease has 5 different subtypes with the majority having a pediatric onset and presentation apart from types 3 and 5 (2). The disease has a unique characterization consisting of polyuria, polydipsia, hypercalciuria, hypokalemia, hyperaldosteronism, metabolic alkalosis, and blood pressure that could be either normal or low (3). The clinical features usually have a varying severity with a

large range of symptoms; However, the presentation in the neonatal age group is usually polyhydramnios, hypovolemia, and sometimes even failure to thrive while polyuria and fatigue are the common symptoms in an adult diagnosed with BS (2). The disease usually presents in either infants or in early childhood and rarely so in late childhood and adulthood (4). The incidence of Bartter is very rare and it is found in 1/1,000,000 people (5). It is worth mentioning that Gitelman syndrome has a very similar clinical course to BS and is sometimes even mentioned with the Bartter subtypes. They both have hypokalemia and metabolic alkalosis with elevated renin and aldosterone making it difficult to distinguish them clinically or on the bases of laboratory manifestations (6). Limited data are available on the sex predominate of the disease but there was a male-to-female ratio of 6:1 in a case series study of childhood Bartter's syndrome (7).

In this study we present a case of adult-onset Bartter syndrome which didn't respond to supportive care such as analgesics at the time of presentation, however, he showed a dramatic response to the treatment of choice and all his symptoms disappeared. Due to the rarity of the condition, our aim is to enrich the literature with information regarding the disease and how it is managed.

## 2. Case Presentation

### 2.1. Patient information

A 23-year-old male presented with a history of polyuria, polydipsia, nocturia, muscle weakness, spasm, and fatigue especially confined to the lower limbs however no vomiting. He had these symptoms for the past two years and did not respond to analgesics and painkillers. He denied usage of any diuretics and urine electrolyte testing was done to confirm this.

### 2.2. Clinical findings

Cardiovascular, respiratory, and gastrointestinal system examinations were all normal with normal vital signs; However, neurological examination showed weakness of the lower limbs.

### 2.3. Diagnostic assessment

Complete blood count (CBC), Erythrocyte sedimentation rate (ESR), Blood glucose, Renal function test, Liver function test, lipid profile, and Thyroid function tests were all normal. There was a decrease in both serum Potassium (2.8Mmol/L) and serum Chloride (95Mmol/L). Tests were ordered to check for Aldosterone and Renin which showed normal serum Renin and serum Aldosterone with a normal unaltered serum ratio of Renin/Aldosterone. The PH study revealed metabolic alkalosis.

There was an increase in total urine volume (7.2 L). A 24-hour urine examination revealed an increase in urine Calcium (670 mg/24 h), increase in urine Chloride (1173 mmol/24 h), increase in urine Potassium (91.8 mmol/24 h) and an increase in urine Sodium (1037 mmol/24 h) but with normal 24-hour urine Magnesium and Creatinine. Urine osmolarity was also seen to decrease (199.0 mOsm/kg) in a random urine examination.

There was an increase in Parathyroid hormone (163.6 pg/ml), however, an MRI scan of the brain showed no abnormality. The patient also had normal Vit D3 and normal neck ultrasound.

### 2.4. Therapeutic intervention

Potassium effervescent tabs (KALINOR) 1 (potassium citrate) (1x1) (1.56gm of potassium & 2.5gm of citrate) were given to replace the lost potassium and reverse the hypokalemia. Aldosterone antagonists such as Spironolactone tabs (25mg, 1x1) were also given since it works to oppose the Renin Angiotensin Aldosterone System (RAAS) while also minimizing the potassium loss to keep it within the physiological range. Lisinopril tab (5mg tab 1x1) as an angiotensin-converting enzyme inhibitor (ACEI) was prescribed to further oppose the RAAS system. Naproxen capsule (500mg, 1x2) was also given for two weeks to help relief the pain he had. The patient also received mineral replacements such as Magnesium effervescent tab (200mg, 1x1), vitamin D3 tabs (2.5µg, 1x1).

### 2.5. Follow-up

The patient showed a dramatic response to the treatments given and all his symptoms were resolved so he continued on the treatment.

## 4. Discussion

Bartter syndrome is a salt-wasting disorder that is usually inherited in an autosomal recessive fashion (2). The disease is typically found in children and neonatal age groups but this does not exclude adolescents, it just makes them much more rare like the current case. BS is associated with a defect in the thick ascending limb due to mutations coding for either the potassium-sodium chloride cotransporter (NKCC2) or the potassium channel (ROMK) (2). The disease has an incidence of 1 per 1,000,000 people, making Gitelman a much more common disorder (5). Bartter syndrome has a varying sexual predominance in which some studies have a male predominance like the study done in India for childhood Bartter syndrome whereas Abdel-al et al. had a series with female predominance (7,8). There even have been studies with no gender predominancy such as the series published by Dillon et al. (9). In a study done in Korea of 54 patients that were either clinically or genetically diagnosed with Bartter syndrome, the median age of onset was 5 months with a range of 0-271 months (10).

The clinical presentation is usually different in neonates when compared to adults. Neonates present with symptoms such as polyuria, polydipsia, dehydration, constipation, sensorineural deafness, eyes that are big and protrude with growth delays, and failure to thrive (5). This is a little different from the way adults usually present since they tend to have more fatigue and weakness with the common polyuria and polydipsia due to excessive salt loss (11). The current patient presented with the expected polyuria, polydipsia, and weakness but also had nocturia and muscle spasm. Interestingly, the fatigue in this patient was found all over his body, however, it seemed to have a greater impact on his lower limbs.

The diagnostic approach is based on a combination of the clinical picture and electrolyte investigation in both serum and urine. Renin and Aldosterone levels can also be helpful as they tend to get elevated in BS mainly due to the activation of the RAAS system as a response to the hypovolemia associated with the polyuria present in Bartter syndrome. Hypokalemia and hypochloremia are usually present in all cases and can help guide the clinician toward the diagnosis of BS. Elevated levels of calcium in a 24-hour urine examination are also very essential as it helps exclude Gitelman syndrome which is associated with low calcium excretion. With this in mind, genetic testing can also be used for confirmation of diagnosis (2). A genetic test was not performed for this case since the patient refused to give consent for genetic testing when asked about it.

Due to the different types of BS present and their different clinical presentations, different clinicians take alternative therapeutic approaches based on their own experience, understanding of the pathophysiology, and the patient's clinical presentation. Since hypokalemia is the predominant finding, most treatments do involve potassium chloride replacement given with prostaglandin inhibitors and potassium sparing diuretics such as Spironolactone. Spironolactone does not only help keep potassium within the physiological range but also antagonizes Aldosterone which can be elevated in many cases of BS (2). This patient, apart from receiving potassium replacement and Spironolactone, also received Lisinopril as an angiotensin-converting enzyme inhibitor to further oppose the Renin-Angiotensin-Aldosterone-System (RAAS).

The prognosis is generally good assuming there is strict adherence to the treatment of choice provided by the clinician, however, some patients do develop chronic kidney disease as a result of long-term hypokalemia (2). Our limitation in this case is that the diagnosis was made solely on the bases of clinical presentation as our patient had most of the symptoms typically seen in an adult-onset BS, however, no genetic testing was done.

## 5. Conclusion

Bartter syndrome is a rare disease. It has a wide range of clinical presentations. It can be diagnosed clinically and confirmed by genetic testing. Conservative management has a good clinical outcome.

## Declarations

**Conflicts of interest:** The author(s) have no conflicts of interest to disclose.

**Ethical approval:** Not applicable, as meta-analyses do not require ethical approval.

**Patient consent** (participation and publication): Not applicable.

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**Authors' contributions:** SMG participated in data collection; BAA designed the study; ASA, FHK participated in preparing the manuscript; SAM, SFA, ZBN, DTG and HRA critically revised the manuscript; FHK, SMG confirmed the authenticity of the data; all authors approved the final version of the manuscript.

**Data availability statement:** Note applicable.

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