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CASE REPORT
Pediatric Emergency

Bartter syndrome: A case report requiring emergency intervention

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ABSTRACT

Bartter syndrome is a rare autosomal recessive genetic disease characterized by hypokalemia, hypochloremic metabolic alkalosis, and normal blood pressure despite hyperaldosteronism and hyperreninemia. In the treatment of Bartter syndrome, the priority is to eliminate dehydration, correct electrolyte imbalance, provide general support and replacement therapies, and treat any accompanying infection. In this article, a patient diagnosed with Bartter syndrome, aged 3 years and 6 months, was presented to highlight this condition in differential diagnosis and increase awareness.

Keywords: Alkalosis; Bartter syndrome; child; dehydration.

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Bartter sendromu: Acil müdahale gerektiren olgu sunumu

ÖZET

Bartter sendromu, hipokalemi, hipokloremik metabolik alkaloz ve hiperaldosteronizm ile hiperreninemiye rağmen normal kan basıncı ile karakterize otozomal resesif nadir bir genetik hastalıktır. Bartter sendromunun tedavisinde, dehidratasyonun giderilmesi, elektrolit dengesizliğinin düzeltilmesi, genel destek ve replasman tedavileri ile varsa eşlik eden enfeksiyonun tedavisi önceliklidir. Bu yazıda, 3 yaş 6 aylık Bartter sendromu tanısı ile takip edilen hasta, ayırıcı tanıda akılda tutulması ve farkındalığın artırılması amacıyla sunulmuştur.

Anahtar Kelimeler: Alkaloz; Bartter sendromu; çocuk; dehidratasyon.

INTRODUCTION

Bartter syndrome is a rare autosomal recessive genetic disorder characterized by hypokalemia, hypochloremia, metabolic alkalosis, and normal blood pressure despite hyperaldosteronism and hyperreninemia. To date, five types of Bartter syndrome have been identified (1). Clinically, it presents with growth retardation, polyuria, polydipsia, muscle weakness, and dehydration attacks (2). We present a case of Bartter syndrome to highlight the importance of considering it in the differential diagnosis due to the potential for serious complications, such as sudden arrhythmia and arrest secondary to fluid-electrolyte imbalances.

CASE REPORT

A 3-year and 6-month-old male patient presented with complaints of polyuria, polydipsia, excessive salt intake, intermittent vomiting attacks, loss of appetite, and abdominal pain for approximately 9 months. The patient's birth weight was 3,500 grams. At 1 year old, he weighed 10 kg but did not gain weight afterward. Currently, his body weight is 10 kg (<3rd percentile), and his height is 88 cm (<3rd percentile). His medical history is unremarkable, and there is no consanguinity between his parents. Family history reveals kidney stones in the mother and grandmother. During the physical examination, the child appeared to be in good general condition and was playful, but he showed signs of developmental delay. The patient's peak heart rate was 100/min, respiratory rate was 30/min, and blood pressure was 90/60 mmHg. The liver was palpable 2 cm below the costal margin, and Traube's space was open. Other systemic examinations were normal. Laboratory findings revealed hyponatremia (124 mmol/L), hypochloremia (81.7 mmol/L), and hypokalemia (2.9 mmol/L). Blood gas analysis showed a pH of 7.66, actual HCO_3=29.8, and standard HCO_3=33.5. The patient was initiated on maintenance therapy, but metabolic alkalosis and hypochloremia did not improve despite sodium and chloride replacement. Suspecting Bartter syndrome, the patient's spot urine electrolytes were examined. In the spot urine test, sodium was 42 mmol/L, potassium was 7.78 mmol/L, chloride was 37.4 mmol/L, and calcium was 6.8 mg/dL, all of which were high, while magnesium was normal. The patient's blood pressure follow-up was within normal limits. Complete urine analysis showed a pH of 7, density of 1003, and protein++. Serum aldosterone was >50 ng/dL, and after dilution, >300 ng/dL. Serum renin was >500 pg/dL, and after dilution, >720 µg/dL. Renal ultrasound revealed a right kidney measuring 4.5x3.3 cm, a left kidney measuring 7.3x3.6 cm, and grade 1 hydronephrosis in the collecting systems. The patient's biochemical parameters and spot urine electrolytes during clinical follow-up are provided in Tables 1 and 2. Based on these findings, the patient was diagnosed with Bartter syndrome. Replacement therapy was initiated, and he was referred to pediatric nephrology for further management.

DISCUSSION

Bartter syndrome was first described in 1962 by Dr. Frederic Bartter as a condition characterized by hypokalemic hypochloremic metabolic alkalosis, normal blood pressure despite hyperaldosteronism, and growth retardation (3). Over time, it has

Table 1. Serum electrolite levels

	Na+ mmol/L (135–148)	K+ mmol/L (3.5–5.5)	Cl mmol/L (95–108)	Ure mg/dL (10–50)	Cre mg/dL (0–1.2)	Uric acide mg/dL (3.4–7)	Ca++ mg/dL (8.8–10.8)	P mg/dL (3.1–6.2)	Mg++ mg/dL (1.7–2.55)	Total protein g/dL (6.6–8.7)	Albumin g/dL (3.8–5.4)
1th day	124	2.94	81.7	22	0.29	5.1	9.2	3.4	2.01	6.7	3.9
2nd day	128	2.74	86.2	15	0.25	4.3	9.8	3.2	1.98	6.6	4.0
5th day	132	3.67	92	17	0.5	2.6	9.6	3.7	2.0	7.1	4.0
7th day	132	3.25	91	20	0.4	3.2	9.8	4.0	1.8	7.0	4.0
10th day	134	2.89	93	27	0.5	3.2	10.1	3.9	2.0	7.4	4.0

Na+: Sodium; K+: Potassium; Cl: Chloride; Cre: Creatinin; Ca++: Calcium; Mg++: Magnesium.

Table 2. Spot urine electrolite levels

	Na+ mmol/L (40–220) (>30)	K+ mmol/L (2.5–125) (>15)	Cl mmol/L (15–40) (>20)	Ca++ mg/dL (2–6) (>3.5 mg/kg)	Mg++ mg/dL
1th day	42	7.78	37.4	6.7	2.8
3th day	45	7.71	41	6.8	2.7
10th day	49	14			

Na+: Sodium; K+: Potassium; Cl: Chloride; Ca++: Calcium; Mg++: Magnesium.

been determined that this syndrome results from genetic defects that disrupt transport in the Na-K-Cl channels in the ascending limb of the loop of Henle (4) Approximately 200 cases have been reported in the literature. There are five identified types of Bartter syndrome: Types 1, 2, and 4 are characterized by polyhydramnios, prematurity, polyuria, and dehydration in the mother, starting in the prenatal period. Types 1 and 2 are associated with nephrocalcinosis/nephrolithiasis, whereas Type 4 is not but is accompanied by a sensorineural hearing defect (5). Type 3 is the classic form of Bartter syndrome, typically seen in infancy and early childhood. It is characterized by growth retardation, nausea, vomiting, dehydration, and the classic laboratory findings of hypokalemia and hypochloremic metabolic alkalosis (5). Type 5 is the rarest form, characterized by hypocalcemia, hypercalciuria, and low parathyroid hormone (PTH) levels (5).

In Type 3 Bartter syndrome, the disorder is caused by a chloride channel protein defect in the ascending limb of the loop of Henle, specifically due to a mutation in the CLCNKB gene (6). This mutation leads to sodium and chloride loss, reducing intraluminal volume and subsequently activating the renin-angiotensin-aldosterone system (RAAS). Although RAAS aims to promote sodium reuptake, it also causes increased potassium excretion and hydrogen+ ion secretion from the distal loop of Henle (6). This process results in hypochloremic hypokalemic metabolic alkalosis. Symptoms typically present before the age of 6. Diagnosis is often based on a thorough physical examination and laboratory results (7) However, if patients present with symptoms such as nausea, vomiting, and dehydration, and only symptomatic treatment is administered without addressing growth retardation or re-evaluating laboratory values post-treatment, the condition may be overlooked.

Based on the clinical and laboratory findings, our patient appeared to fit the profile of Type 3 classic Bartter syndrome. The patient's low serum sodium, potassium, and chloride levels, alongside normal calcium and magnesium levels, supported this diagnosis. Our patient's magnesium levels were normal, which helped us differentiate this condition from Gitelman syndrome. Despite sodium chloride replacement, the patient's metabolic alkalosis and electrolyte imbalances did not improve, suggesting chloride-unresponsive metabolic alkalosis. The absence of elevated blood pressure, despite high renin and aldosterone levels, ruled out conditions such as renal artery stenosis and primary hyperaldosteronism, steering us toward Bartter syndrome. Additionally, diuretics that affect the ascending loop of Henle could produce similar effects, but in this case, the absence of nephrocalcinosis and hearing loss, combined with the patient's symptom onset and laboratory findings, further supported a diagnosis of Type 3 Bartter syndrome.

In the treatment of Bartter syndrome, the primary goals are to eliminate dehydration, correct electrolyte imbalances, provide general support and replacement therapies, and treat any concomitant infections if present. Indomethacin, a prostaglandin inhibitor, is a key component of treatment (8).

Bartter syndrome is a rare genetic disorder, and early diagnosis and intervention are crucial for effective management and improving outcomes. Although some genetic mutations have been identified, diagnosis is primarily clinical (9). It is especially important to consider Bartter syndrome in cases of milder presentations, such as halted weight gain and hypokalemic metabolic alkalosis.

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Hasta Onamı: Yazılı hasta onamı bu çalışmaya katılan hastaların ailelerinden alınmıştır.

Çıkar Çatışması: Yazarlar çıkar çatışması bildirmemişlerdir.

Yazma Yardımı için Yapay Zeka Kullanımı: Beyan edilmedi.

Mali Destek: Yazarlar bu çalışma için mali destek almadıklarını beyan etmişlerdir.

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