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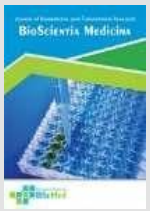

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Bartter Syndrome: A Case Report

Yayik Supriyani[1*], Deka Viotra[2], Putri Deas Hadilofyani[1]

[1]Department of Internal Medicine, Faculty of Medicine, Universitas Andalas/Dr. M. Djamil General Hospital, Padang, Indonesia

[2]Nephrology Division, Department of Internal Medicine, Faculty of Medicine, Universitas Andalas/Dr. M. Djamil General Hospital, Padang, Indonesia

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***Corresponding author:**
Yayik Supriyani

E-mail address:
yayiksupriyani86@yahoo.com

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A B S T R A C T

Background: Bartter's syndrome refers to a group of genetic disorders that affect the renal tubular system, which is responsible for reabsorbing various substances such as sodium, potassium, and chloride from the urine into the blood. This study aimed to present a clinical case related to Bartter syndrome. **Case presentation:** A 52-year-old male patient in the internal medicine department of Dr. M. Djamil General Hospital Padang with the main complaint of weakness in both legs increasing since 1 day ago. On laboratory examination, the patient found potassium 1.7 mmol/L, indicating hypokalemia. Renal function examination showed normal kidney function. Examination of blood gas analysis showed results of metabolic alkalosis. Examination of urine potassium obtained potassium levels of 22 mmol/day, urine osmolarity of 140 mOsm/kgH_2O at serum osmolarity of 274 mOsm/kgH_2O, with TTKG (transtubular potassium gradient) = 28. The patient was diagnosed with Bartter syndrome. Treatment is carried out by administering KSR tablets 3x600 mg orally while monitoring electrolytes regularly. **Conclusion:** This patient has hypokalemia, metabolic alkalosis, normal magnesium and calcium, and hypercalciuria. This patient is diagnosed with Bartter syndrome.

1. Introduction

Bartter's syndrome is a group of rare diseases that affect the kidneys and affect the electrolyte and fluid balance in the body. The syndrome is named after Professor Frederic Bartter, an American nephrologist who first described the symptoms and characteristics of the disease in the 1960s. Bartter's syndrome refers to a group of genetic disorders that affect the renal tubular system, which is responsible for reabsorbing various substances such as sodium, potassium, and chloride from the urine into the blood. Patients with Bartter's Syndrome experience disturbances in various functions of the kidney tubules, which cause disturbances in the balance of electrolytes and fluids in the body. The impact can vary from mild to severe cases, depending on the type and severity of this syndrome in a particular individual.[1,2]

Common symptoms of Bartter's syndrome include electrolyte imbalances such as hypokalemia (low levels of potassium in the blood), hypochloremia (low levels of chloride in the blood), metabolic alkalosis (condition of high blood pH levels), and increased urine production. Other symptoms that may appear include complaints such as muscle weakness, fatigue, cramps, and growth retardation in children. Although Bartter's syndrome is an inherited genetic condition, its symptoms usually appear in childhood or adolescence. The diagnosis is made through a series of laboratory tests, such as blood and urine tests, as well as genetic mapping to identify mutations associated

with this disease. Treatment of Bartter's syndrome aims to manage symptoms and prevent associated complications. This can involve taking electrolyte supplements, such as potassium and magnesium, as well as drugs that reduce the loss of electrolytes in the urine. Therapy can be adjusted based on the severity and type of Bartter Syndrome experienced by the patient.[3-5] This study aimed to present a clinical case related to Bartter syndrome.

2. Case Presentation

A 52-year-old male patient in the internal medicine department of Dr. M. Djamil General Hospital Padang with the main complaint of weakness in both legs increasing since 1 day ago. Weakness in both legs has increased since 1 day ago. Weakness appeared suddenly after the patient worked. Weakness is not followed by weakness in the arms. Complaints are getting worse, so the patient is difficult to walk. Complaints are not accompanied by numbness. Frequent urination since 3 months ago. The patient often wakes up at night to urinate. There is no pain when urinating. No sandy urination. No bloody urination. The patient easily feels hungry and thirsty since 3 months ago. Decreased appetite since 1 month before admission to the hospital. Patients only eat 1-2 times a day, eating only half a serving of food. Weight loss of approximately 10 kg since 1 month ago. Coughing since 2 weeks before entering the hospital, coughing up phlegm, yellowish white. No coughing up blood.

Physical examination found the patient looked moderately ill, with vital signs within normal limits and the patient looked underweight. On examination, the head, face, eyes, ears, throat, and neck were within normal limits. On thoracic examination, there were bronchovesicular breath sounds and loud fine crackles. Examination of the abdomen and extremities was within normal limits. Examination of motor strength found a decrease in motor strength in the lower extremities, which decreased compared to the upper extremities. Sensory examination within normal limits.

On laboratory examination, the patient found potassium 1.7 mmol/L, indicating hypokalemia. Examination of kidney function found levels of urea 9 mg/dl and creatinine 0.4 mg/dl, indicating normal kidney function. Examination of blood gas analysis showed results of metabolic alkalosis with a Ph value of 7.5, a pCO_2 value of 34, a pO_2 value of 119, and an HCO_3- value = 46 mmol/L. Examination of urine potassium obtained potassium levels of 22 mmol/day, urine osmolarity of 140 mOsm/kgH_2O at serum osmolarity of 274 mOsm/kgH_2O, with TTKG (transtubular potassium gradient) = 28, which indicates that hypokalemia is due to potassium leakage in the kidneys. In this patient, the molar ratio of urine calcium and creatinine was 0.22, which indicated hypercalciuria. In this patient, there was high urinary chloride excretion of 74.7 mmol/L. The patient was diagnosed with Bartter syndrome. Treatment is carried out by administering KSR tablets 3x600 mg orally while monitoring electrolytes regularly.

3. Discussion

Periodic diagnosis of paralysis, e.c, hypokalemia e,c, Bartter syndrome based on anamnesis, physical examination, and supporting examinations. In patients from anamnesis found a weakness in the legs 1 day before entering the hospital. The patient has a history of weakness in all four limbs and hypokalemia previously. Hypokalemia is one of the most common electrolyte abnormalities encountered in clinical practice. More than 20% of hospitalized patients are hypokalaemic. Hypokalemia is defined as a serum potassium level of less than 3.5 mmol/L. Hypokalemia may occur in up to 40% of outpatients taking thiazides and diuretics. The kidney is the main regulator of potassium homeostasis, so kidney dysfunction can also cause abnormalities in serum potassium levels. Significant muscle weakness occurs at serum potassium levels below 2.5 mmol/L but may occur at higher levels if the onset is acute. Muscle weakness starts from the lower extremities, continues to involve the trunk and upper extremities, and has the potential

to progress to paralysis. Respiratory muscles can also be affected and cause respiratory failure. Gastrointestinal muscle involvement can cause ileus with associated symptoms of nausea, vomiting, and abdominal distention. Severe hypokalemia can also cause muscle cramps, rhabdomyolysis, and myoglobinuria.[6-8]

Clinical presentation of Bartter syndrome with periodic paralysis is rare, but it has been reported in a 12-year-old boy who suffers from periodic paralysis, and the results of clinical and laboratory examinations support the direction of Bartter syndrome. The patient had periodic bouts of paralysis for 2 years and had a total of 10 attacks, lasting 1 to 3 days. The patient has growth disorders, polyuria, and polydipsia. Laboratory tests show alkalosis, normomagnesemia, hypercalciuria, and hyperaldosteronism. The cause of hypokalemia can be determined from the patient's medical history, namely hypokalemia due to vomiting, diarrhea, or use of diuretics, so further investigation is not necessary. However, in certain cases, the cause is sometimes uncertain, and making the diagnosis can be difficult.[14] Temporary causes of hypokalemia may be due to shifts in cells, whereas ongoing hypokalemia may be manifested by inadequate intake or excessive loss of potassium. Hypokalemia due to excessive loss of potassium can be caused by loss of potassium due to disorders of the kidney or extra-renal. The causes and sources of hypokalemia can be assessed by history taking and physical examination, as well as paying attention to the patient's acid-base status and blood pressure.[9-11]

On laboratory examination, the patient found potassium 1.7 mmol/L, indicating hypokalemia. Examination of kidney function found levels of urea 9 mg/dl and creatinine 0.4 mg/dl, indicating normal kidney function. Examination of blood gas analysis showed results of metabolic alkalosis with a Ph value of 7.5, a pCO_2 value of 34, a pO_2 value of 119, and an HCO_3- value = 46 mmol/L. Examination of urine potassium showed potassium levels of 22 mmol/day, urine osmolarity of 140 mOsm/kgH_2O at serum osmolarity of 274 mOsm/kgH_2O, with TTKG = 28, which indicated that hypokalemia was due to potassium leakage in the kidney. Approaches to distinguish between renal and extrarenal causes of hypokalemia are based on urinary potassium excretion measured in a 24-hour urine sample or random urine potassium concentration values. A urine potassium concentration of more than 15 mmol/day or more than 15 mmol/L indicates a renal cause. Hypokalemia, if the urine potassium concentration is less than 15 mmol/day or 15 mmol/L, indicates that there is an extra-renal cause. The results of a 24-hour urine potassium examination in this patient were 42 mmol/L. These results may indicate that there is a cause in the kidney that results in hypokalemia. Urinary chloride levels can differentiate between causes of hypokalemia due to loss of potassium from the kidneys from metabolic alkalosis. Patients with hypokalemia resulting from renal loss of potassium with metabolic alkalosis can be divided into two groups based on urinary chloride excretion. Possible causes of low urine chloride levels (<10 mmol/L) are vomiting, post hypercapnic alkalosis, and diarrhea.[16] Patients with high urine chloride levels (>20 mmol/L) and low or normal blood pressure can be due to diuretics, Gitelman's syndrome, and Bartter's syndrome. The most common cause of hypokalemia is the use of diuretic therapy, but it can be ruled out from history. In this patient, there was a high urinary chloride excretion of 74.7 mmol/L, which was associated with metabolic alkalosis and low blood pressure.[12-14]

To distinguish Gitelman syndrome, it is important to check the urine for calcium. In patients with Bartter syndrome, there is often hypercalciuria, whereas, in Gitelman's syndrome, there is hypocalciuria. Urine calcium examination can use 24-hour urine or urine at any time, but it is better to use 24-hour urine to measure the ratio of urine calcium and creatinine. The normal value for the ratio of urinary calcium to creatinine is less than 0.14. If it is more than 0.20, it is said to be hypercalciuria. In this patient, the molar ratio of urine calcium and creatinine was 0.22, which indicated hypercalciuria.[15,16]

4. Conclusion

Based on the hypokalemia pathway approach with hypokalemia conditions, metabolic alkalosis, normal magnesium and calcium, and hypercalciuria, this patient is diagnosed with Bartter syndrome.

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