DOI:10.38173/RST.2021.21.1.8:91-95

| | |
|---|---|
| **Title:** | *BARTTER SYNDROME: A RARE RENAL TUBULOPATHY* |
| **Authors:** | Amelia GENUNCHE-DUMITRESCU<br>Daniela BADEA<br>Marius Cristian MARINAȘ<br>Daniela NEAGOE<br>Tiberiu-Stefaniță ȚENEA-COJAN<br>Aurelian-Adrian BADEA |

**Section:** MEDICINE

**Issue:** 1(21)/2021

| | |
|---|---|
| **Received:** 11 January 2021 | **Revised:** 19 February 2021 |
| **Accepted:** 6 March 2021 | **Available Online:** 15 March 2021 |

Paper available online HERE

RST Research and Science Today
WWW.RSTJOURNAL.COM
SPRING 2021
No. 1(21)/2021
ISSN-P: 2247-4455 / ISSN-E: 2285-9632

Medical Sciences

# BARTTER SYNDROME: A RARE RENAL TUBULOPATHY

**Amelia GENUNCHE-DUMITRESCU**[1]
**Daniela BADEA**[2]
**Marius Cristian MARINAȘ**[3]
**Daniela NEAGOE**[4]
**Tiberiu-Stefaniță ȚENEA-COJAN**[5]
**Aurelian-Adrian BADEA**[6]

**ABSTRACT:**
*BARTTER SYNDROME IS AN AUTOSOMAL RECESIVE DISORDER ASSOCIATED WITH BIOCHEMICAL ABNORMALITIES SUCH AS HYPOKALEMIA AND ELEVATED RENIN, ALDOSTERON LEVEL AND METABOLIC ALKALOSIS. THE CLINICAL MANIFESTATION SUCH AS ANOREXIA, CONSTIPATION, NAUSEA, MUSCULAR HYPOTONIA ARE DETERMINED BY HYPOKALEMIA. PROGNOSIS IS GENERALLY GOOD WITH EARLY STABILIZATION OF METABOLIC ABNORMALITIES. LIFE LONG MEDICATION IS NEEDED IN THIS DISEASE.*

**KEY WORDS:** BARTTER SYNDROME, HYPOKALEMIA

## INTRODUCTION

Barter syndrome is a genetic disorder which consist in a defect in the thick ascending limb of the loop of Henle. This syndrome was caracterized by low potassium levels (hypokalemia), hyperreninemia and hyperaldosteronism with or without hypomagnesemia and in alkalosis. Blood presure is normal or low in these patients. Also this diagnosis includes high urinary potassium, hyperplasia of the juxtaglomerular apparatus on kidney biopsy. The clinical manifestations are: leg cramps, weakness, constipation, myalgia, vomiting and abnormal heart rhytm.

[1] Clinical Hospital of Emergency Craiova, 1st Medical Clinic, University of Medicine and Pharmacy Craiova, Romania ;
[2] Clinical Hospital of Emergency Craiova, Department of Physiology, University of Medicine and Pharmacy Craiova, Romania;
[3] Clinical Hospital of Emergency Craiova, Department of Anatomy, University of Medicine and Pharmacy Craiova, Romania;
[4] Clinical Hospital of Emergency Craiova, 1st Medical Clinic, University of Medicine and Pharmacy Craiova, Romania ;
[5] CFR Hospital, Surgery Department, University of Medicine and Pharmacy Craiova, Romania;
[6] Clinical Hospital of Emergency Bucharest, Resident physician, Romania ;

## CASE REPORT

We are reporting a case of a 39 year-old female who was admited in our hospital. She had significant nausea, vomiting, polydipsia, polyuria, muscle weakness and pain. The patient has a history of vomiting and muscle cramps and she had been admitted in various hospitals for dehydration episodes. During this period she received antiacids and anti-emetics but vomiting persisted. Her mother related persistent electrolyte abnormalities during the time. Physical examination revealed a patient with growth retardation, not conjunctival pallor or icterus. Lung and heart ascultation did not show any abnormalities. The patient blood pressure was 90/60 mmHg which is low and the pulse rate 84 b/min ritmic. Chvostek's and Trousseau's signs were positive.

Laboratory investigations included normal values of blood count, hemoglobin, liver function test, blood sugar and renal functions. The analyses of serum electrolytes consist in persistent hypokalemia (3 mEq/L), hyponatremia (125 mEq/L) and hypocloremia (64 mEq/L) and normal magnesium level (2,34 mg/dl). Her arterial blood gas analysis showed metabolic alkalosis, Ph 7,58, PO2 94 mmHg, PCO2 35 mmHg and $HCO3^-$ 36,6 mmol/L. Also she had elevated renin and aldosteron (256,10 pg/ml) levels. Urine biochemical analysis showed high levels of sodium (87 mmol/l), potassium (38 mmol/l) and chloride (136,2 mmol/l). The proteine urinar was 745 mg/24 hours (N-0-300) and the urine culture showed no infection.

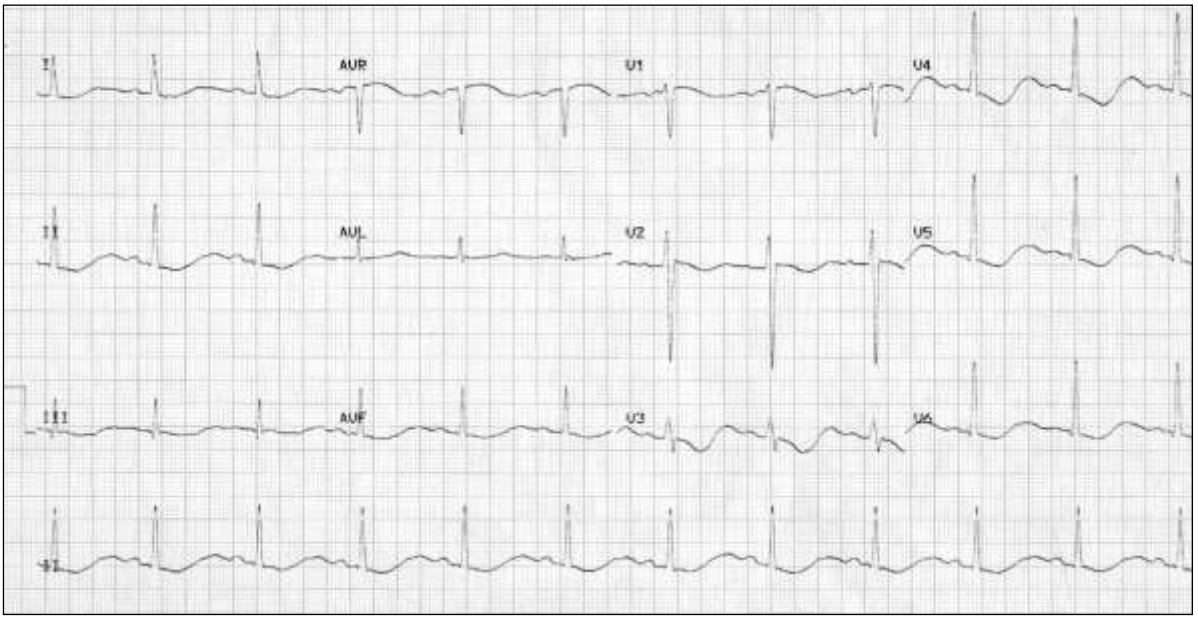


Fig. 1. Electrocardiography

Electrocardiography showed PR prolongation, ST segment depression, T wave flattening and large U wave.

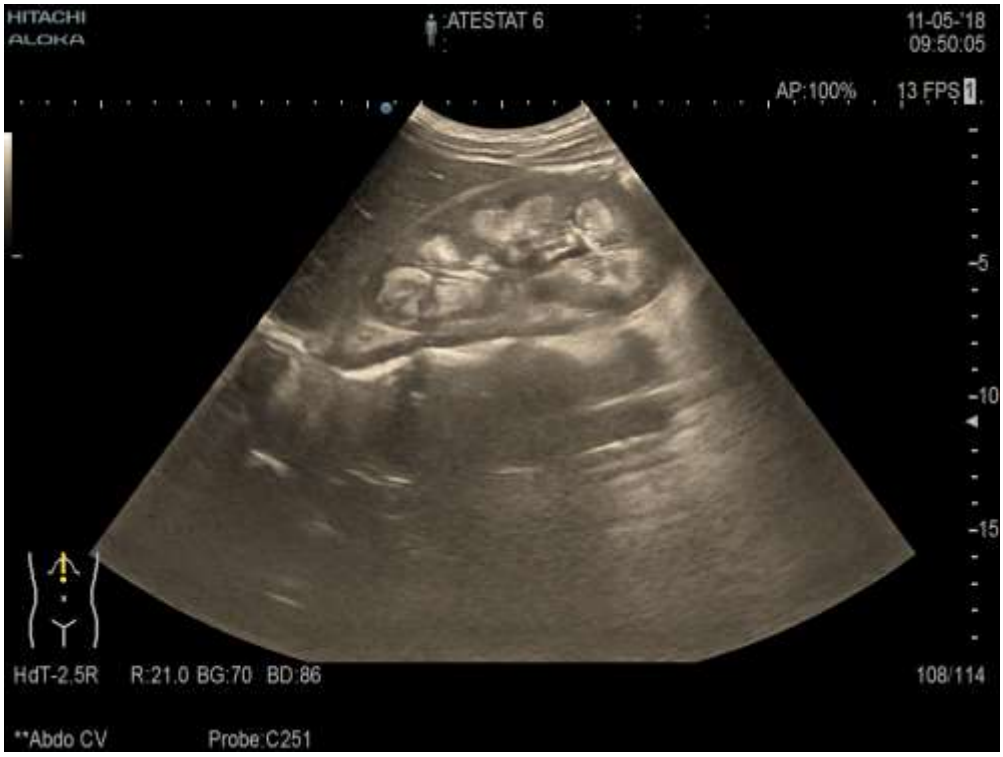


Fig. 2. Ultrasonography of kidney

The abdominal ultrasound discribed normal sized kidneys and increased diffuse echogenicity, hyperechoic pyramids and rare calcareous interstitial deposits.

## DISCUSSION

Hypokalemia is defined as a serum potassium level of less than 3,5 mEg/l (3,5 mmol/l), normal potassium levels are between 3,5 and 5 mEg/l. The causes of hypokalemia include: vomiting, diarrhea, medication like furosemide and steroids, diabetes insipidus, pancreatic fistulae and the presence of an adenoma. Bartter syndrome is a maladie with autosomal-recessively inherited and characterized by the association of hypokalemia, hypochloremia, metabolic alkalosis, growth retardation and the activation of the renin-aldosterone axis[7]. Bartter syndrome is classified into five types. Type I and II are the most severe disorders. They are characterized by polyhydramnios during pregnancy and premature birth[8],[9].

These patients present nephrocalcinosis which contributes to the late development of kidney insufficiency. Bartter syndrome type III is present later in life and manifests with dehydration, electrolyte imbalance, polyuria, polydipsia, vomiting, and growth retardation [10]. Bartter syndrome type IV is associated with sensorineural deafness. In contrast, patients with

[7] Bartter FC, Pronove P, Gill JR Jr, Mac Cardle RC. *Hyperplasia of the juxtaglomerular complex with hyperaldosteronism and hypokalemic alkalosis: a new syndrome*, The American Journal of Medicine (1962) **33**:811–28.
[8] Seyberth HW, Schlingmann KP. *Bartter- and Gitelman-like syndromes: salt-losing tubulopathies with loop or DCT defects*, Pediatric Nephrology, 2011;26:1789–1802.
[9] Peters M, Jeck N, Reinalter S, et al. *Clinical presentation of genetically defined patients with hypokalemic salt-losing tubulopathies*. The American Journal of Medicine , 2002;112:183–190.
[10] Rodriguez-Soriano J., *Bartter and related syndromes: the puzzle is almost solved*, Pediatric Nephrology (1998) **12**:315–27;

Gitelman which is another syndrome that has low serum magnesium levels with hypocalciuria with the absence of signs of overt volume depletion[11]. Gitelman syndrome is a recessive salt-losing tubulopathy caused by the SLC12A3 gene mutation. SLC12A3 gene encodes the thiazide-sensitive transporter NCCT (sodium chloride co-transporter). NCCT is located in the distal convoluted tubular cells (DCC), which are responsible for 7–10% of electrolyte tubular absorption. This condition is sometimes confused with Bartter syndrome.
We excluded other causes of hypokalemia such as chronic loop diuretic use, laxative abuse, endocrine causes and enema abuse. In our patient we determined ACTH, TSH, FT4, FT3 and cortyzol hormones which are in normal values. We performed computed tomography of the brain and abdomen in order to rule out an endocrine pathology (such as primary aldosteronism, rennin secreting tumors).

Upper endoscopy does not reveal any kind of subacute obstruction or gastroesophageal reflux causing recurrent vomiting. Treatment is generally focused on the repair of hypokalemia by the inhibition of the rennin-angiotensin-aldosteron or the prostaglandin-kinin system. The treatment in our case consists in correcting deshydratation and electrolyte disorders. In the hospital we administered intravenous normal saline along with potassium, 40 mEq per liter over 4 hours and potassium-sparing diuretic Spironolactonum 25 mg, three times a day (with blood pressure control). During this time we monitorized the potassium levels. We recomand to eat foods rich in potassium such as: leafy green vegetables, tomatoes, bananas and avocado.

## CONCLUSION

The persistent decreased values of kalemia in the absence of other causes such as diarrhea, vomiting, diuretics abuse and endocrine diseases represent the possibility of Bartter syndrome. The early treatment has improved the prognosis and prevented various complications such as hypokalemic nephropaties and chronic renal failure.

[11] Brenner B, Levine S. *Inherited disorders of renal tubule*, Brenner and Rector's The kidney, 8th ed. Philadelphia, PA: WB Saunders; 2007:1411-1414 [chapter 40].

## REFERENCES

1. **G. Graziani, C. Fedeli, L. Moroni, L. Cosmai, S. Badalamenti, C. Ponticelli**, *Gitelman syndrome: pathophysiological and clinical aspects*, QJM, 103 (2010), pp. 741-748
2. **Vezzoli G, Arcidiacono T, Paloschi V, et al**. *Autosomal dominant hypocalcemia with mild type 5 Bartter syndrome*, Journal of Nephrology, 2006;19:525-528.
3. **Zaffanello M, Taranta A, Palma A, Bettinelli A, Marseglia GL, Emma F**. "*Type IV Bartter syndrome: report of two new cases"*, Pediatric Nephrology, 2006; **21** (6): 766–70.
4. **Deschênes G, Fila M.** *Primary molecular disorders and secondary biological adaptations in Bartter syndrome*, International Journal of Nephrology, 2011;2011:1–8.
5. **Kleta R, Basoglu C, Kuwertz-Bröking E.** *New treatment options for Bartter's syndrome*, The New England Journal of Medicine, (2000) **343**:661–2.
6. **Knoers NVAM, Starremans PGJF, Monnens LAH**: *Hypokalemic tubular disorders*, Oxford Textbook in Clinical Nephrology. Edited by: Davidson AM, Cameron JS, Grunfeld J-P, Ponticelli C, Ritz E, Winearls CG, van Ypersele C. 2005, Oxford University Press, 995-1004.