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Case report

BARTTER'S SYNDROME

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ABSTRACT

OBJECTIVE- Bartter's syndrome is a rare genetic disorder characterized by renal salt wasting, hypokalemia and metabolic alkalosis. **CASE REPORT**: A 32 years old male patient presented to casualty with complaints of generalised weakness and chest pain of 15 days duration. Patient had pallor, with loss of buccal pad of fat, skin turgor was lost, and extremities were cold. PR-90bpm, feeble, BP-50/40mmHg. History of similar complaints 1 month back for which he was hospitalised for 15 days and was not completely relieved of symptoms. The differential diagnosis of metabolic alkalosis with hypokalemia without hypertension are; 1. Bartter's syndrome, 2. Gitelman's syndrome, 3. Surreptitious vomiting and 4. Diuretic abuse. **CONCLUSION:** Acquired Bartter's and Gitelman's like syndromes have been described with autoimmune disorders, sarcoidosis and various drugs. This patient could have a yet undetected or unknown mutation of a protein which has a structural or a functional role in renal tubular cells (of Loop of Henle).

KEY WORDS: bartter's syndrome, renal salt wasting, hypokalemia, metabolic alkalosis, sarcoidosis.

INTRODUCTION

Bartter's syndrome is a rare genetic disorder[1], where in there are specific defects in the channels governing the normal functioning of kidney, leading to abnormal loss of electrolytes including sodium, potassium, chloride and calcium etc. Bartter's syndrome is characterised by renal salt wasting, hypokalaemia and metabolic alkalosis [1]. Bartter's syndrome is classified into 5 types based on the channel involved due to specific mutation in the genes coding for that specific channel. BS type 1 is linked to mutations of the gene SLC12A1 (Solute carrier family 12 sodium/potassium/chloride transporters, member 1) on chromosome 15 (15q15-q21.1). BS type 2 is linked to a gene called KCNJ1 (mapped to chromosome 11q21-25), BS type 3 is linked to the gene ClCNKb (mapped to chromosome 1p36) while BS type 4 is linked to gene BSND (mapped to chromosome 1p31). BS type 5 is associated with activating mutations of the CASR gene (mapped to chromosome 3q13.3-q21)[1].

CASE REPORT

A 32 years old male patient presented to casualty with complaints of generalised weakness and chest pain of 15 days duration. The chest pain was diffuse dull aching type, non radiating, not associated with vomiting or syncope. Patient had history of similar complaints 1 month back for which he was hospitalised for 15 days in other hospital and was not completely relieved of symptoms. All other family members are keeping good health. Patient gives history of consumption of alcohol for 10 years, 1-2 quarters per day. On examination patient had pallor, with loss of buccal pad of fat, skin turgor was lost, and extremities were cold. PR-90 bpm, feeble, BP-50/40 mmHg. Patient was conscious and well oriented. Patient had hepatomegaly and periumbilical tenderness on deep palpation.

Treatment was started with IV Fluids of 3L on day one following which BP was 64/48 mmHg, and later Dopamine infusion was started @ 3ml/hr for two days following which dose was tapered and stopped with BP of 118/70 mmHg.

Complete blood count and renal profile was done depicted in table one, revealing hypokalaemia and hyponatraemia with raised creatinine and urea on day one, as severe hypokalaemia (K^+ 2.0) was present on day 1 correction was started with an infusion of 1 ampule of KCl in 100ml NS over 4 hours, following which potassium levels was 2.6 mg/dl on day two and potassium was continued with oral KCl

supplements of 15 ml thrice a day and potassium rich diet, even then potassium was on lower side on day three i.e, 2.6mg/dl .

Table 1 also reveals hypocalcaemia and normal magnesium, to evaluate for the persistence of hypokalemia urinary osmolality (273 mosm/kg), urinary sodium (46 mmol/24hrs) and urinary potassium (20.4 mmol/24hrs) were done and depicted in table one.

Table 1. Blood investigations on Day 1, Day 2, and Day 3.

DAY OF ADMISSION		DAY 1	DAY 2
TOTAL COUNT	16700 cells/cumm		
RBC	3.54 millions/cumm		
HEMOGLOBIN	9.4 gm%		
PLATELET COUNT	3.19Lkh		
ESR	70mm at 1st hour		
RBS	80mg/dl		
CREATININE	3.5 mg/dl		
S. MAGNESIUM	1.6 mg/dl		
S. SODIUM	121 mg/dl	130 mg/dl	126 mg/dl
S. POTASSIUM	2.0 mg/dl	2.6 mg/dl	2.6 mg/dl
U. OSMOLALITY	273.0 mOsm/kg		
U. SODIUM	46.0 mmol/day		
U. POTASSIUM	20.4mmol/day		

A clinical approach to know the cause of hypokalaemia was done.

The Trans Tubular Potassium Gradient (TTKG) here is 9.8 and ABG analysis shows metabolic alkalosis and USG abdomen shows normal kidney size and shape with peritoneal cavity showing accumulation of fluid.

DISCUSSION

On evaluating for persistence of low potassium levels even with intravenous and oral potassium correction, our patient had increased loss of urinary potassium 20.4 mmol/day i.e, >15 mmol/day and TTKG > 9.8 indicating that the loss is through renal tubules and ABG analysis showing metabolic alkalosis. The differential diagnosis constituting these parameters includes - 1. Surreptitious vomiting, 2. Diuretic abuse, 3. Bartter's syndrome and 4. Gitelman's syndrome.

The first two are ruled out as no history suggestive of the same, Gitelman's syndrome can be ruled out based on magnesium levels which are within normal levels and are not low as in Gitelman's syndrome[2], hence Barter's syndrome is the probable diagnosis.

Acquired Bartter's syndrome and Gitelman's like syndrome are associated with autoimmune disorders like sarcoidosis and various drugs[3, 4].

CONCLUSION

We speculate that this patient represents Bartter's syndrome, and the same should be considered as a provisional diagnosis in patients with electrolyte disturbances as mentioned above irrespective of clinical presentation and age of presentation, Bartter's syndrome is also documented in elderly too[5].

This patient may have a new acquired mutation in renal tubular cells, and this mutation couldn't be confirmed in our patient as the genetic testing for Bartter's syndrome is not available in India[6].

REFERENCES

1. Davison AM, Cameron JS, Grünfeld JP, et al, editors. Oxford textbook of clinical nephrology. 3rd ed. New York: Oxford University Press; 2005. p. 995-1000.
2. Salant DJ, Patel PS. Polycystic kidney disease and other inherited tubular disorders. In: Fauci AS, Braunwald E, Kasper DL, Hauser SL, Longo DL, Jameson JL, et al, editors. Harrison's principles of internal medicine. 17th ed. New York: McGraw Hill Medical; 2008. p. 1797-1806.
3. Tung-Miny, Shih-Hua L, Chuang Y, et al. A syndrome resembling Bartter›s syndrome in sarcoidosis. Nephrol Dial Transplant 2009;24:667-9.
4. Schwarz C, Barisani T, Bauer E, Druml W. A woman with red eyes and hypokalemia: A case of acquired Gitelman syndrome. Wien Klin Wochenschr 2006; 118:239-42.
5. Tamagawa E, Inaba H, Ota T, Ariyasu H, et al. Bartter syndrome type 3 in an elderly complicated with adrenocorticotropin-deficiency. Endocr J. 2014;61(9):855-60. Epub 2014 Jun 24.
6. Sachin B Punatar, Divyashree S, Vishal M Jogi. A novel variant of bartter's syndrome. Journal of The Association of Physicians of India 2015; 63:58-61.