ADULT VARIANT BARTTER'S SYNDROME- A CASE REPORT

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ABSTRACT

BACKGROUND

Bartter syndrome is a group of channelopathies with different genetic origins and molecular pathophysiologies, but sharing common feature of decreased tubular transport of sodium chloride in thick ascending loop of Henle (TAL),[1] although more common in antenatal group. Classic adult variant of Bartter syndrome is a rare entity. We hereby present a rare adult variant of classic Bartter syndrome.

KEYWORDS

Bartter.

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BACKGROUND

Bartter syndrome was described by Bartter in 1962 as a disorder affecting Thick Ascending Limb (TAL) of Henle's loop with classic clinical characteristics such as salt wasting, hypokalaemic, hypochloraemic metabolic alkalosis, hyperaldosteronism with hyperreninaemia and normal blood pressure. Bartter syndrome has been classified into five types based on mutations in different genes in various parts of the renal tubular cells. Type I, II, IV and V Batter's syndrome are common in neonates, while type III or the classic Bartter syndrome can present both in neonates (antenatal type) and adolescents. Type III Batter syndrome is due to mutations in CLCNKB (chloride) channel located in basolateral membrane of renal tubular cells and shares similar clinical characteristics with other Bartter types. In contrary, type III Batter has less severe disease progression rate.

CASE REPORT

A 32-year-old female presented with history of low-grade fever for 8 days followed by weakness in all four extremities for 2 days. She did not have diurnal variation in fever, chills or rigor. Weakness of the extremities was sudden and simultaneous in onset without any complaints pertaining to sensory system. She was referred as a case of acute inflammatory demyelinating polyneuropathy. On examination, respiratory rate was 10 cycles/min., blood pressure was 110/70 mmHg, power in all four limbs was 2/5, generalised areflexia and normal sensory system. Other system examination was found to be normal.

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Provisional Diagnosis

Quadriparesis under evaluation.

Baseline Laboratory Investigation

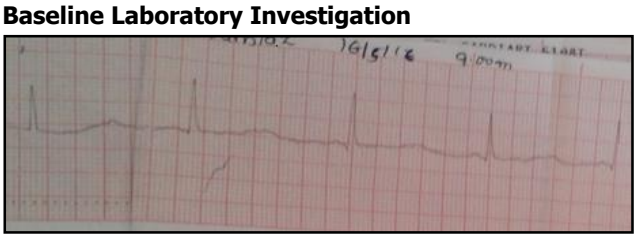

Figure 1. ECG Showing U Waves

Her serum potassium level was 1.8 mEq/L. ECG showed classic U waves of hypokalaemia (Figure 1). Arterial blood gas analysis showed metabolic alkalosis. Urine potassium was 30 mmol/day and transtubular potassium gradient was 6.14 indicating increased distal K+ secretion. Urine chloride level and urinary Ca2+/Cr ratio was 157 mmol/L and 0.51, respectively. Ultransonography of abdomen was found to be normal.

Final Diagnosis

Barter syndrome (as per algorithmic approach (Figure 2)).

Clinical Management and Follow-Up

Patient was given potassium through IV (20 mEq/hrs.) and oral (Potchlor syrup 40 mEq/day) supplementations, which was tapered slowly based on patient's response. She developed an episode of carpopedal spasm during her hospitalisation period for which calcium gluconate supplementation was provided. Her hypomagnesaemia was also corrected. Acid base status showed improvement. Her power improved from 1/5 to 5/5 over a course of 10 days

and she walked on her own to home happily. She was put on potassium sparing diuretics and discharged.

Serum Biochemistry	Normal Range	Day 1	Day 4	Day 8	Day 10
Calcium (mg/dL)	8.1-10.6	7.4	9.0	9.3	9.2
Magnesium (mg/dL)	1.8-2.6	1.0	1.9	4.3	4.1
Sodium (mmol/L)	135-145	145	141	136	132
Potassium (mmol/L)	3.5-5	1.8	1.7	2.5	4.7
Chloride (mmol/L)	90-110	96	98	100	104

Table 1. Laboratory Parameters of the Patient During the Period of Hospitalisation

Other parameters-
24 hrs. urine potassium- 20 mmol/L.
Urine osmolality- 813.2 mOsm/kg (50-1400 mOsm/kg).
Plasma osmolality- 292 mOsm/kg (282-295 mOsm/kg).
24 hrs. urine calcium- 205 mg/dL (100-240 mg/dL).
24 hrs. urine creatinine- 396 mg/dL (500-2000 mg/dL).
Serum aldosterone- 288 pmol/L (60-260 pmol/L).
ABG analysis- pH 7.5, pO2- 100 mmHg, pCO2- 42 mmHg, HCO3- 30 mmol/L.

Interpretation

Primary metabolic alkalosis with secondary metabolic acidosis.

We followed the algorithm below to find out the cause of hypokalaemia.

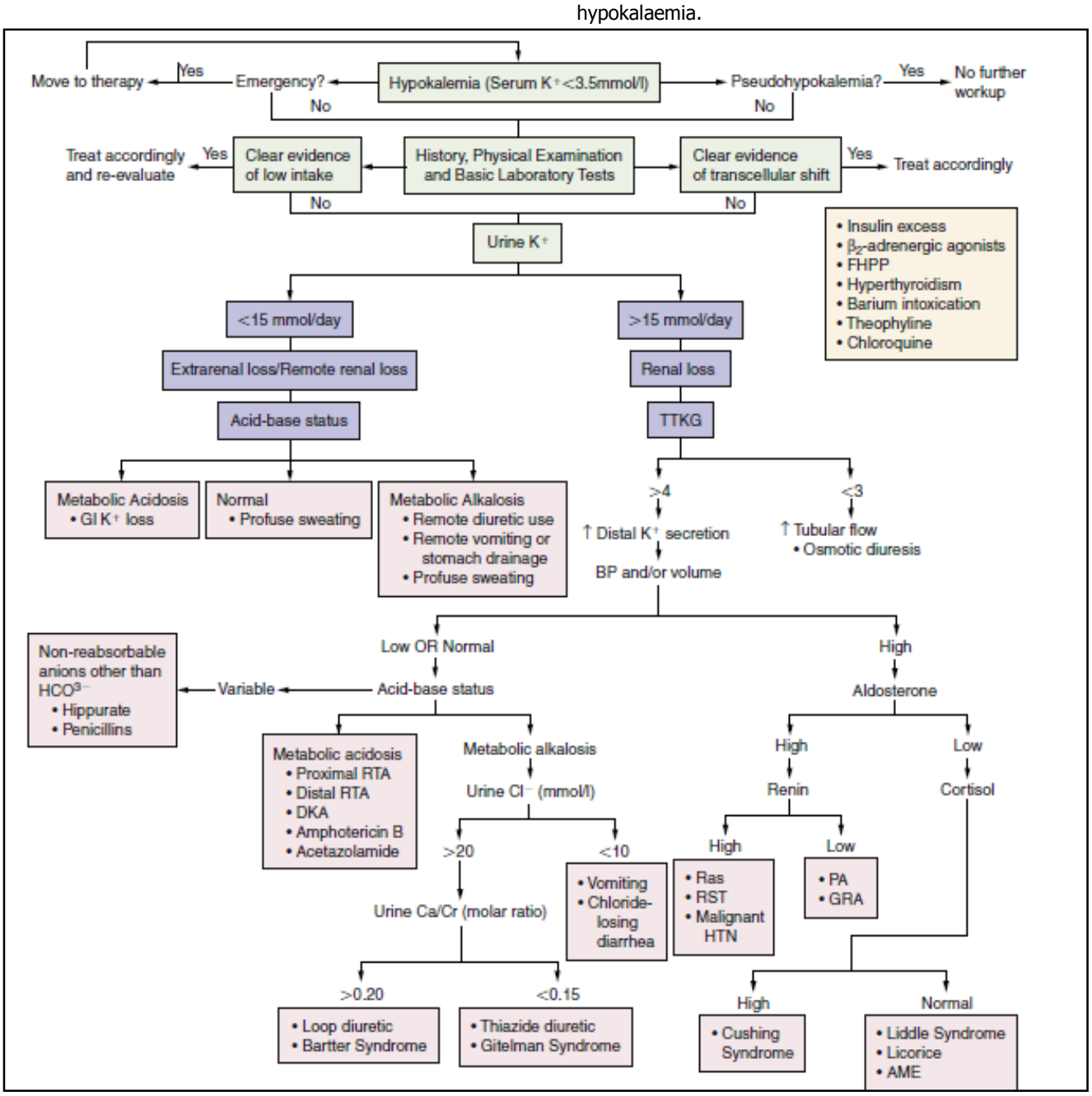

Figure 2. Algorithmic Approach to Hypokalaemia

Clinical approach to hypokalaemia; AME- Apparent mineralocorticoid excess; BP- Blood pressure; CCD- Cortical collecting duct; DKA- Diabetic ketoacidosis; FHPP- Familial hypokalaemic periodic paralysis; GI- Gastrointestinal; GRA-

Glucocorticoid-remediable aldosteronism; HTN- Hypertension; PA- Primary hyperaldosteronism; RAS- Renal artery stenosis; RST- Renin-secreting tumour; RTA- Renal tubular acidosis; TTKG- Transtubular potassium concentration gradient.

DISCUSSION

Patients with classic Batter syndrome typically suffer from polyuria and polydipsia due to the reduction in renal concentrating ability of the renal tubular cells with marked activation of the renin-angiotensin-aldosterone axis.[2,3] There may be an increase in urinary calcium excretion in these patients and around 20% of them will be hypomagnesaemic.

Patients with antenatal Batter syndrome suffer from a severe systemic disorder characterised by marked electrolyte wasting, polyhydramnios, hypercalciuria, nephrocalcinosis with increased renal prostaglandin synthesis and excretion accounting for much of the systemic symptoms. All the five types of Batter's syndrome is due to mutation in either of the genes regulating the channels, which transports Na+, K+ and Cl- ions at TALH leading to hypokalaemia and salt wasting. In contrast, Gitelman Syndrome (GS) is genetically homogeneous caused almost exclusively by loss-of-function mutations in the thiazide-sensitive Na^+-Cl^- cotransporter of the distal convoluted tubules. Patients with GS are uniformly hypomagnesaemic and exhibit marked hypocalciuria rather than the hypercalciuria typically seen in Batter syndrome. Thus, urinary calcium excretion is a critical diagnostic criterion to differentiate these two salt wasting syndromes.

Our patient presented with acute onset quadriparesis with hypokalaemic metabolic alkalosis. There was no history of diuretic abuse or vomiting. Hypomagnesaemia, a feature of Gitelman can be seen in 20% Bartter syndrome, which was seen in our case. Hypomagnesaemia could well be important in the pathogenesis of chondrocalcinosis through two simultaneous mechanisms by reducing the activity of pyrophosphatases and by facilitating the crystallisation of pyrophosphates. These mechanisms could explain the association of Bartter's syndrome and chondrocalcinosis.[4]

After following the algorithmic approach, the diagnosis of classic variant of Bartter syndrome was diagnosed. The patient improved with potassium supplementation and power improved from 1/5 to 5/5 at discharge.

CONCLUSION

In a patient with quadriparesis, always first rule out treatable causes like in this case, which was referred as acute inflammatory demyelinating polyneuropathy subjecting patient to unnecessary imaging. Bartter syndrome usually presents in antenatal group and very few cases have been reported in adults in India.[5] Prompt recognition of these cases is necessary since these patients need lifelong supplements and timely treatment help in reducing the mortality and morbidity associated with the syndrome.

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