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Case Report

A rare presentation of adult Bartter syndrome: a case report

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ABSTRACT

Bartter syndrome (BS) is an inherited renal tubular disease. It is caused by a defective salt reabsorption in the thick ascending limb of loop of Henle. The term BS signify a group of renal disease which is the common divisor of hypokalemia and metabolic alkalosis. BS is categorized into 5 types based on the specific channel; type 1 is linked to gene SLC12A1, type 2 is linked to a gene KCNJ1, type 3 is linked to a gene called CICNKb while type 4 is linked to a gene BSND and type 5 is linked to CASR gene. This disorder is correlated with an increased antenatal and neonatal mortality. Here, a 61 years old female patient was presented with complaints of fever, myalgia, nausea, and cough for one day, decreased appetite and unable to do daily routine. Patient had conscious, oriented and afebrile. She had history of OAD, type 2 diabetes mellitus and hypertension. On hospital stay, she was started on IV calcium gluconate, potassium chloride, magnesium sulphate, antipyretics and other supportive measures. This case concludes the rarity of the BS. Correct diagnosis of BS is more expensive and not routinely available genetic testing. Therefore, the significance of diagnosing the case is more relevant.

Keywords: Bartter syndrome, Metabolic alkalosis, Dyselectrolemia, Acute infective exacerbation, Hyponatremic encephalopathy

INTRODUCTION

Bartter syndrome (BS) is an autosomal recessive inherent renal tubular disease caused by CLCNKB mutation resulting in salt wasting, hypokalemia and metabolic alkalosis. BS was first reported by Barter et al at 1962.[1] Characterized by extreme hypokalemia, metabolic alkalosis, hyperchloremia, hyper renin-induced angiotensinemia, hyper-aldosteronemia and normal blood pressure. CLCNKB gene encodes chloride channel Kb, which is expressed in the thick segment of the ascending branch of the loop of Henle, distal tubule and basolateral membrane of the epithelial cells of collective tubule.[2] It is classified into; based on phenotype, neonatal (antenatal) and genotype, classical BS.[3] The other five classification includes type 1 (antenatal BS; hyper prostaglandin E syndrome), type 2 (neonatal BS with transient hyperkalemia; hyper prostaglandin E syndrome), type 3 (classic BS), type 4 (antenatal BS; hyper prostaglandin E syndrome with sensorineural deafness, BART), type 5 (hypocalcemia with BS).[4] BS are usually present the pediatric population except type 3 and type 5. Type 4 is more severe than other types.

The treatment focus on correcting the dehydration, electrolyte disturbances and hypercalciuria. The other viable options for treating BS are NSAIDs and aldosterone agonist. Due to rarity of the disease, there is no curative management.[5,6] The durable clinical benefit was found in Cox II inhibitors.

CASE REPORT

A 61-year-old woman presented with complaints of fever, myalgia, nausea, cough since one day, decreased appetite and unable to do daily routine. She had medical history of

OAD, type 2 diabetes mellitus and hypertension and her medication history include T. spiranolactone 50 mg bd, inj. human actrapid 40-40-30 u S/C, syp. levodropropizine 5 ml TDS. On physical examination she was conscious, oriented and afebrile.

Laboratory investigation showed declined hemoglobin (8.6 gm/dl) potassium (1.9 mmol/l), sodium (122 mmol/l), phosphorous (2.2 mmol/l), magnesium (0.7 mg/dl), calcium (5.6 mg/dL), iron (29 ug/dL), TIBC (224 ug/dl), transferrin saturation (13%), eGFR (35 ml/min) and elevated ferritin (>1000 ng/ml), random blood sugar, CRP (80.4 mg/dl), urea (29 mg/dl), HbA1C (6.9%), lipid profile. Cardiology consultation was given, in view of elevated Trop I (28 pg/ml) and their order carried out. Echo taken, showed overall good LV systolic function (EF-50%). Calcium spot urine 11.5, calcium creatinine ratio 0.5. NCCT of abdomen with chest screening showed no significant abnormality. CT brain plain showed age related neuroparenchymal changes. Neuro medicine consultation was given. In view of irrelevant speech and weakness of lower limb and advised to continue same management. Blood C and S and C and S was sterile, in view of feature suggestive of BS.

Initially patient was started on IV antibiotics (inj. cefoperazone+sulbactam 1.5g IV BD and inj. doxycycline 100 mg IV BD), proton pump inhibitors (inj. pantoprazole 40 mg IV BD), corticosteroids (inj. methylprednisolone 4 mg IV BD and T. fludrocortisone 0.1 mg P/O 1-0-0), neuraminidase inhibitors (c. oseltamivir 15 mg P/O BD), syp. potassium chloride 15 ml P/O q8h), IV calcium gluconate, magnesium sulphate, sShe was hospitalized for 11 days and finally the patient got symptomatically improved and discharged.

DISCUSSION

BS is a genetic and hereditary disorder causing metabolic alkalosis and hypokalemia without hypertension. The pathogenic of BS is purely genetic based. The more common suspect is infants. The common features in BS are hypokalemia, hypovolemia, hypochloremia, hypercalciuria, hypermagnesuria, secondary hyperaldosteronism, metabolic alkalosis.[7] Main manifestation in this case was fever, myalgia, nausea, cough since one day, decreased appetite and unable to do daily routine. Hypokalemia is a common clinical presentation in BS and the present case showed the renal potassium loss.

There is no cure for BS and the treatment focus on correcting the symptoms like dehydration, electrolyte disturbances and hypercalciuria. The use of NSAIDs, inhibit then renin angiotensin system there by blocking prostaglandin synthesis, potassium retention diuretics are considered for the management of BS patients.[8-10]

CONCLUSION

This case emphasizes the rare case of adult-onset BS, which was a genetically inherited caused by mutations encoding ion channels or transporters. Diagnosis is also a challenging sequence in batter syndrome. The rarity leads the controversy in the treatment of BS, only the symptoms can be treated.

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