Case Report

Status epilepticus as the only presentation of the neonatal Bartter syndrome

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ABSTRACT

Bartter syndrome is a rare hereditary (autosomal recessive) salt-losing tubulopathy characterized by hypokalemia, hypochloremia, metabolic alkalosis, and normal blood pressure with hyperreninemia, The underlying renal abnormality results in excessive urinary losses of sodium, chloride, and potassium. We report a case of a four-month-old infant with neonatal Bartter syndrome, who presented only with status epilepticus. To the best of our present knowledge, there is no reported case of Bartter syndrome who presented with status epilepticus.

Key words: Bartter syndrome, neonate, status epilepticus

Introduction

The hallmarks of the Bartter syndrome (BS) are renal salt wasting and hypokalemic metabolic alkalosis accompanied by normal or low blood pressure despite secondary hyperaldosteronism.[1] The BS is clinically categorized into antenatal BS and classical BS.[2] BS is caused by genetic disruptions of ion transporters or channels, in the thick ascending limb of the loop of Henle. Laboratory tests typically reveal increased excretion of urinary potassium, chloride, increased serum bicarbonate, metabolic alkalosis, and increased serum renin and aldosterone levels.[3] Here, we report a case with BS that initially presented with features of sepsis followed by status epilepticus.

Case Report

A four-month-old male baby, born to a non-consanguineous parent, with an uneventful antenatal and perinatal period, was delivered by Cesarean section, with a birth weight of 2.3 kg. The baby was presented at our pediatric emergency with lethargy, poor feeding, low-grade fever, and abnormal staring for two to three days prior to admission. There was no history of vomiting or convulsion, and urine output was normal. The mother was primi, with no history of abortion and there was no significant family history.

He was on exclusive breast feeding, and was apparently normal before this illness. On general physical examination, the child was properly nourished with a normal weight of 5 kg, length of 62 cm and head circumference of 40 cm, without bulging anterior fontanels. He was well-hydrated with no facial dysmorphism, normotensive, and had no respiratory distress. Central nervous system examination did not reveal any localizing signs. Abdominal and cardio-respiratory examinations did not reveal any abnormalities. Therefore, the baby was investigated as a case of septicemia, with suspected meningitis. Management was started with intravenous fluids, antibiotics (inj. Ceftriaxone) and other supportive measures. Investigations including complete blood count, blood culture, and a Cerebrospinal Fluid (CSF) study were found to be within normal range except for the mild anemia (Hb% - 9.8 gm%) and raised C-reactive protein (CRP-10mg/L). The random blood sugar was 72 mg%. The Computed Tomography (CT)

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scan of the brain revealed no abnormality. As the baby improved on the second day of antibiotic treatment, this was assumed to be a case of septicemia. However, on the sixth day of admission, the baby again became lethargic and developed generalized tonic seizures, which were not controlled by injection lorazepam, phenobarbitone and phenytoin. Later, injection Midazolam infusion was started to control the status epilepticus. He rapidly went into shock. After initial resuscitation with fluid boluses, dopamine and dobutamine infusion was begun and serum electrolytes were sent. Arterial blood gas (ABG) analysis was sent, which showed severe hypokalemia (2 meq/L), hyponatremia (112 meq/L), hypochloremia, and metabolic alkalosis (pH - 7.56). Serum levels of creatinine (0.8 mg/dl), ionized calcium (4.2 mg/dl), and magnesium (2.1 mg/dl) were normal. The urinary examination revealed hyposthenuria (specific gravity 1.007) with increased urinary loss of sodium, chloride, potassium, and calcium (sodium 56 mEq/L; chloride 45 mEq/L; potassium 40 mEq/L; calcium to creatinine ratio –0.38). Urine output was 2 – 3 mL/kg/hour. Ultra sonogram (USG) of the abdomen revealed nephrocalcinosis. Elevated plasma level of renin of 40 U/L (normal 4 – 8 U/L) was also found. As this patient was fulfilling all criteria, diagnosis of Bartter syndrome was made. A genetic study could not be performed in our case. The child was treated with intravenous bolus of both hypertonic saline and potassium chloride as he had severe hyponatremia and hypokalemia. He was maintained on supportive care, potassium supplements, and intravenous indomethacin. There was improvement in the clinical and biochemical parameters, with below normal range of serum electrolytes. Although the seizures were controlled, he developed aspiration pneumonia and was put on a ventilator on day 10, with a rapid and progressive downhill course. Subsequently the baby died on the thirteenth day of admission.

Discussion

Bartter syndrome (BS) is an autosomal recessive heterogeneous renal tubular disorder in which one of the key transport proteins involved in transcellular Na-K-CL transport in the thick ascending limb of the Henle's loop or distal convoluted tubule is impaired.[3,4] The antenatal hypercalciuric variant of BS, also termed as the 'hyperprostaglandin E syndrome' is a more severe condition of the polyuric loop dysfunction.[4] It shares common features (hypokalemia, metabolic alkalosis, and hyperreninemic hyperaldosteronism with normal blood pressure) with the other types of BS.[5] The hallmark of antenatal BS is polyhydramnios and prematurity, followed by postnatal polyuria and hypercalciuria.[6] Postnatally affected infants present with the typical pattern of impaired tubular reabsorption in the thick ascending limb of Henle's loop including salt-wasting, isosthenuric or hyposthenuric polyuria, and hypercalciuria.[7] In addition to iso- or hyposthenuric polyuria, newborns lose large amounts of sodium and chloride through the urine. Increased sodium delivery to the distal tubule, particularly when combined with the activation of the renin-angiotensin-aldosterone system, enhances excretion of potassium and hydrogen ions, causing hypokalemic alkalosis.[8,9] Within the first months of life, nearly all patients develop medullary nephrocalcinosis in parallel with persistent hypercalciuria.[8] In our case, the diagnosis of BS was confirmed by a biochemical analysis suggestive of BS. Hyponatremia is not very common in BS, which is seen only in severe cases.[8] In our case, the patient had severe hyponatremia, which presented as seizures. However, the patient did not have any typical clinical features like facial dysmorphism, failure to thrive or any history of polyuria, vomiting or other suggestive features. In a case where antibiotics like amino glycosides were used, it might have precipitated the electrolyte abnormality in an apparently healthy recovering baby.[4] However, in our case, only ceftriaxone was used in the beginning. Thus, the possibility of secondary BS was ruled out. As we could not perform genetic tests in our case, the disease presenting with seizures and renal tubulopathy, like the EAST (epilepsy, ataxia, sensorineural deafness, and tubulopathy) syndrome[10] should also be kept in the differential diagnosis. In this patient, BS was clinically suspected first on the basis of typical laboratory findings including hypokalemic metabolic alkalosis, hyperreninemia, hypercalciuria, increased urinary sodium and chloride concentration, and nephrocalcinosis in USG.

Therefore, in any case that presents with status epilepticus, serum electrolytes need to be sent and if any abnormalities like hyponatremia or hypokalemia are present, Bartter syndrome must be kept in mind.

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