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Case Report

Neonatal Bartter Syndrome: A Case Report

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Abstract

Bartter syndrome is a genetic rare disorder usually present with set of renal tubular disorders characterized by hypokalaemia, hypochloraemia, metabolic alkalosis, and hyper reninemia and normal blood pressure. Renal tubular abnormality of sodium, potassium and Chloride resorption results in excessive urinary losses of sodium, chloride, and potassium. We report a case of Neonatal Bartter syndrome, who presented from 48 hrs of birth.

Keywords: Bartter syndrome; Neonate; Hyponatremia; Hypokalaemia; Alkalosis

Introduction

Bartter syndrome is an autosomal recessive disorder and is a rare condition .The main problem in Barter syndrome is failure in the absorption of sodium and chloride from the loop of Henle as result there is excessive urinary electrolyte losses, polyurea and dehydration. The volume depletion causing hyperaldosteronism which leads to hypokalaemia and metabolic alkalosis.

Case Presentation

A 10 minutes old boy of a first degree consanguineous parent was admitted in the Neonatal Intensive Care Unit (NICU) of Bangabandhu Sheikh Mujib Medical University (BSMMU), Dhaka, Bangladesh, for prematurity (31 weeks of gestation), low birth weight (1100 gram) and delayed cry after birth requiring bag and mask ventilation for one minute. There was history of poly hydramnios (amniotic fluid index 38 cm) of the mother, requiring amniocentesis. Mother got 2 doses of antenatal corticosteroid 24 hrs prior to delivery. There was history of sib death in the first week of life due to severe dehydration but cause of death was not confirmed. On arrival, baby was pink in the air, reflex & activity good, SPO2 96% without supplemental 0_2, normothermic, capillary refill time <3 second, euglycemic, and vital signs were within normal limit. Anthropometric measurements shows weight below 10th centile, length 45 cm and OFC, 29 cm above 10th centile and systemic examination revealed only cachexia and mild dehydration, blood pressure 68/42 mmHg (mean-52 mmHg) otherwise normal. After admission full blood count and electrolytes were normal (Na: 134 mmol/L K: 4.6 mmol/L Cl: 99 mmol/L, TCO2 : 19.9 mmol/L) and he was on nothing per oral with 10% dextrose in aqua and later on 0.225% NaCl and orogastric feeding. At 48 hrs of age there were hyponatremia, hypokalaemia with metabolic alkalosis (Na: 126 mmol/L, K: 2.3 mmol/L, Cl: 89 mmol/L, TCO2: 28.1 mmol/L, S Creatinine 1.46 mg/dl) intravenous fluid and electrolytes correction was given with 0.9% NaCl and intravenous potassium. After 12 hrs of correction there were till dyselectrolytemia and high urinary excretion of sodium potassium and chloride with normal calcium and magnesium (Serum Na: 122 mmol/L, K: 2.1 mmol/L, Cl: 78 mmol/L, TCO2: 28.2 mmol/L, S creatinine- 1.16 mg/dl, Serum osmolality- 281 m osmole/kg) there was high urinary sodium, potassium and chloride and calcium with high calcium creatinine ratio (Urinary Na- 102 mmol/L, K- 38.7 mmol/L, Cl- 121 mmol/L, P^H 6.5, Specific gravity-1.010, Urinary calcium- 7.8 mg/dl, Urinary creatinine- 2 mg/dl, Urinary calcium/creatinine excretion ratio was elevated, 3.8) and high urine output, 5.6 ml/kg/hr, arterial blood gas showed metabolic alkalosis (pH- 7.66, PCO_2 - 28 mmHg, PaO_2 – 110 mmHg, HCO3 – 31.5 mmol/L). Plasma rennin and aldosterone was elevated (plasma rennin, 47.20 pg/ml, aldosterone 360.00 pg/ml. normal, 20-180 pg/ml). Subsequently an electrolyte was with normal limit with supplementation of 3% NaCl and potassium supplementation. Serum creatinine and urinary output become normal at 7th day of post natal age (Figure 2 and 3). At discharged Sodium supplement was given orally by 3% NaCl with breast milk along oral potassium supplementation. Follow up after 7 days of discharge weight gaining was adequate 25 gm/day, no dehydration (Figure 1) electrolytes, and creatinine was normal and we scheduled for a long term follow up.

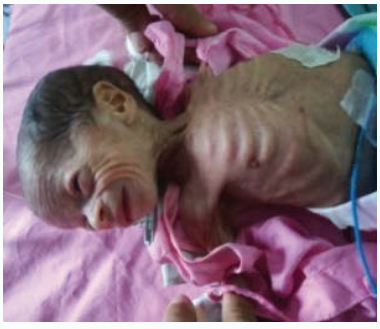

Figure 1: Inflammatory changes and bowel wall thickening centred.

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Figure 1B: After rehydration and electrolyte correction.

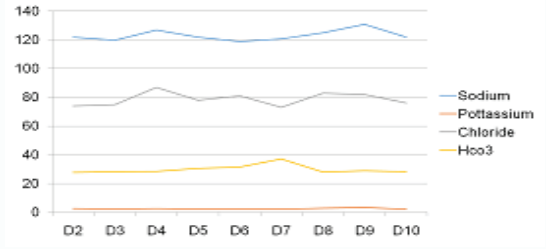

Figure 2: Electrolytes trends.

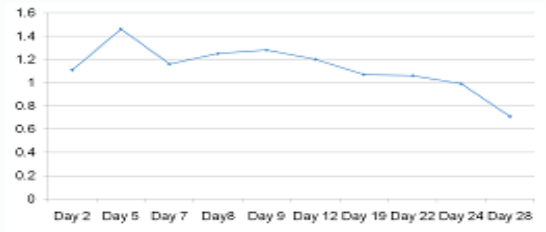

Figure 3: Creatinine trends.

His full blood count, blood urea, magnesium and creatinine were normal but despite serum sodium

Being normal (135 meq/l), potassium (2.5 meq/l) and chloride (92 meq/l) were low. His arterial blood gas revealed metabolic alkalosis with a pH of 7.56 and serum and urinary osmolalities were 270 mosm/kg water and 159 mosm/kg water respectively. His urine electrolytes revealed increased excretion of sodium, potassium and chloride. Although normocalcaemic, his urinary calcium/creatinine excretion ratio was elevated (0.68), indicating hypercalciuria. His subsequent ultrasound scan of abdomen was normal.

Discussion

Bartter Syndrome (BS) is a rare autosomal recessive tubulopathies with a prevalence of 1 in 100 000 [1]. Federic Bartter first describes this phenomenon in 1962 as a combination of hyperplasia of juxtaglomerular complex, hyperaldosteronism and hypokalaemic metabolic alkalosis. Genetic analysis proved that this kind of electrolytes abnormalities were due to the mutations of genes encoding proteins that transport ions across renal cells in the thick ascending limb of loop of Henle [2]. Bartter syndrome is usually classified in three categories neonatal, classical and Gitelman syndrome. The current classification includes type I and II (neonatal or antenatal Bartter syndrome) due to defective NKCC2 and ROMK genes respectively, affecting the Na - K - 2 Cl symporter predominantly [3]. In our case clinical features and lab findings suggestive of Bartter syndrome type I, though genetic analysis was not done for confirmation of the disease. The 'classic' Bartter syndrome, Type III, is due to CLCNKB genetic abnormalities causing defect in the basal chloride channel. Type IV is the most severe form and rare and there is combined loop and distal tubule dysfunction and usually associated sensorineural deafness. In case sodium resorption mechanism by the renal tubular system, 70% is reabsorbed in the proximal tubule and 30% is reabsorbed from the water impermeable thick ascending limb via a Na - K - Cl co transporter driven by low intracellular concentration of sodium. If this mechanism of sodium resorption is defective there result in large volume of urine output with an increased amounts of Sodium and potassium loss through the renal tubular system leading to hypokalaemia, hypochloraemia and dehydration. In our patient there was maternal polyhydramnios secondary to fetal polyuria and there was neonatal dehydration but there was no dysmorphic feature or any sensory neural hearing defect. In a classic Bartter there is linear growth retardation in the later life and there may be episodic dehydration, polydipsia, polyurea and recurrent carpopedal spasm. Bartter syndrome may confuse with Gitelman syndrome in which magnesium levels are significantly low. The differentiating points are hypercalciuria is prominent in Bartter syndrome but it is negligible in Gitelman syndrome, in which magnesium excretion is greatly elevated and there is mild hypomagnesemia [3]. In Bartter syndrome there is elevated serum renin, aldosterone and prostaglandin E2, in our case renin and aldosterone was also elevated. A definitive diagnosis can be reached by genetic mutation analysis [4]. These cases should be treated with proper hydration, potassium and sodium supplementation. One study showed that potassium sparing diuretic; spironolactone may benefit transiently [5]. In neonatal Bartter syndrome there is possibilities of nephrocalcinosis, chronic renal failure and short stature, so long duration of follow plan should be made and complication should be address in time [6].

References

1. Ji W, Foo JN, O'Roak BJ, Zhao H, Larson MG, Simon DB, et al. Rare independent mutations in renal salt handling genes contribute to blood pressure variation. Nat Genet. 2008;40(5):592-9.
2. Rodriguez-Soriano J. Bartter and related syndromes: the puzzle is almost solved. Pediatr Nephrol. 1998;12(4):315-27.
3. Naesens M, Steels P, Verberckmoes R, Vanrenterghem Y, Kuypers D. Bartter's and Gitelman's syndromes: from gene to clinic. Nephron Physiol. 2004;96(3):65-78.
4. Patrick WR, Tse Y, Ashton E, Daniela Iancu D, Jenkins L, Bienias M. et al. Clinical and diagnostic features of Bartter and Gitelman syndromes. Clinical Kidney J. 2018;11(3):302-9.
5. Zaffanello M, Taranta A, Palma A, Bettinelli A, Marseglia GL, Emma F. Type IV Bartter syndrome: report of two new cases. Pediatr Nephrol. 2006;21(6):766-70.
6. Vaisbich MH, Fujimura MD, Koch VH. Bartter syndrome: benefits and side effects of long-term treatment. Pediatr Nephrol. 2004;19(8):858-63.