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# Bartter Syndrome

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## Abstract

We report a case of Neonatal Bartter syndrome in a baby girl born at 25 weeks+ 5 days of gestational age to a Primi mother with antenatal polyhydramnios. Baby was admitted as a case of respiratory distress syndrome in NICU and she developed severe dehydration leading to pre-renal renal impairment along with typical biochemical changes of hypokalaemic, hypochloraemic metabolic alkalosis.

**Keywords:** Neonatal; Bartter syndrome

## Introduction

Bartter syndrome is a rare condition due to defective absorption of sodium and chloride in the Thick ascending loop of Henle resulting in excessive loss of urinary electrolytes and fluid. High index of suspicion is necessary to diagnose the case as biochemical changes which are characteristic of the condition such as hypokalaemic hypochloraemic metabolic alkalosis may be masked early in neonatal period especially with polyuria and dehydration.

## Case Report

This extreme preterm female baby born at 25 weeks and 5 days gestational age to a Primi mother (birth weight of 700 grams) was admitted to our Neonatal unit as a case of respiratory distress syndrome. Mother had severe polyhydramnios. Amnio reduction was done in the antenatal period and amniotic fluid was sent for analysis which was unremarkable. Baby was managed with surfactant administration and respiratory support with nasal CPAP and she remained stable till 2nd week of life when Fio2 requirement gradually started to increase. Echocardiography revealed large PDA which was ligated on 21 st day of life and again baby was stable on nasal CPAP.

In the second week of life baby had raised urea and creatinine levels highest urea being 35.5mmol/L and highest value of creatinine 294 mmol/L on 9th day of life. Since then her urea levels have decreased upto 10 mmol/l at the end of 2 weeks and creatinine upto 94 mmol/l by 3rd week of life. At around 3 weeks of age she developed significant hyponatremia (Na125), Hypokalemia (K 2.58) and hypochloremia (Cl 71.8). She was also noted to have compensated metabolic alkalosis and was failing to thrive. Blood gas (pH 7.53, pCO2 82.2, BE 37.9, HCO3 68.2).Low serum potassium and chloride levels were persistent even after large amounts of supplements.

Neonatal Bartter's syndrome was suspected. Her urine output has always been more >6 ml/kg/hour. Her urine chloride levels were found to be high. (103 mmol/l), Urine calcium creatinine ratio 1.15 (mg/dl), random urine sodium (105 mmol/l), urine potassium (13mmol/l). Her electrolyte imbalance was corrected by Intravenous supplements and TPN and later commenced on oral supplements of sodium and potassium chloride. She required large amounts of oral potassium chloride and sodium chloride to maintain the electrolyte levels within normal range.

She continued to have significant metabolic alkalosis and most of the time it was compensated. Her renal function tests improved gradually with decline of creatinine and urea. Her electrolyte disturbances were managed with adjustment of electrolyte supplements. She also developed hypocalcemia (serum ionized calcium varying from 0.8 -1.10 mmol/l) and put on oral calcium supplements. Her Magnesium levels have always been in normal range. Baby developed chronic lung disease and was oxygen dependent even at the time of discharge. Repeat ECHO done at

three and half months of age revealed severe Hypertrophic Obstructive Cardiomyopathy (HOCM) along with cleft anterior mitral leaflet and baby was put on propranolol as advised by pediatric cardiologist.

USG abdomen revealed bilateral enlarged kidneys with increased cortical echoes and partially maintained corticomedullary differentiation. No evidence of nephrocalcinosis.

Baby was discharged at around 6 months of age with weight of 3.52 Kg (0.4-2nd centile). Head circumference 34 cm (below 0.4 centile) on 28% oxygen by nasal cannula to long term care on oral supplements of sodium chloride, potassium chloride, multivitamins, ferrous sulfate, oral calcium and propranolol.

## Discussion

Bartter syndrome also called salt-losing tubulopathy with secondary hyperaldosteronism, originally described by Bartter and colleagues in 1962 represents a set of closely related, autosomal recessive renal tubular disorders characterized by hypokalemia, hypochloremia, metabolic alkalosis, and hyperreninemia with normal blood pressure. Long term activation of the renin-angiotensin-aldosterone system (RAAS) and subsequent secondary hyperaldosteronism causes hyperplasia of the juxtaglomerular apparatus and hence increased renin levels.

The defective sodium chloride transport in the loop of Henle associated with Bartter syndrome leads to the impaired electrochemical gradient, which is necessary for calcium and magnesium reabsorption, leading to increased urinary loss of calcium and magnesium. Nephrocalcinosis is commonly seen in patients with Bartter syndrome.

Traditionally it had been classified into three main clinical variants:

- Neonatal (or antenatal) Bartter syndrome
- Classic Bartter syndrome
- Gitelman syndrome

With the advances in molecular diagnostics it had been found that Bartter syndrome results from mutations in genes that affect the function of ion channels and transporters that mediate salt reabsorption in the distal nephron segments. This revelation created a new classification system for Bartter syndrome on the basis of the underlying genetics.

Neonatal Bartter syndrome type 1 results from mutations of the gene located on chromosome 15q15-q21 which codes for Na-K-2Cl co-transporter (NKCC2) of the Thick Ascending Loop of Henle. Mutations of ROMK gene situated upon chromosome 11q24-25 which codes for K+ channels responsible for recycling and reabsorbing K+ back into tubular lumen results in Neonatal Bartter type 2.

Neonatal Bartter syndrome may present as maternal polyhydramnios, secondary to fetal polyuria evident by 24-30 weeks' gestation. Delivery often occurs before term. The subsequent course is characterized by life-threatening episodes of fluid loss, clinical volume depletion, and failure to thrive. Some patients with neonatal Bartter syndrome (types IV and V) develop sensorineural deafness.

Bartter syndrome is diagnosed on the basis of clinical presentation and laboratory findings, including severe hypokalemia and metabolic alkalosis in almost all cases. Other abnormalities include increased serum renin and aldosterone levels along with hypercalciuria. Urine electrolytes show elevated chloride, sodium, potassium, and PGE2 excretion. Differential diagnoses may include Gitelman's syndrome, Primary hyperaldosteronism and Pseudo-Bartter syndrome which were excluded in our case.

While managing the babies with Bartter syndrome the target is to prevent dehydration and normalize potassium levels in serum which can be achieved with high doses of oral potassium supplementation. Sodium supplementation may also be required either by saline infusion in neonatal period or oral sodium chloride. Indomethacin, a prostaglandin inhibitor has also been found to be effective in many cases. Bartter syndrome is not curable, the degree of disability depends on the severity of the receptor dysfunction. With close monitoring of electrolyte balance, volume status and nutrition long term prognosis is good.

Finding of hypertrophic obstructive cardiomyopathy in our case posed a diagnostic dilemma whether it is a separate entity from Bartter or same genetic mutation is the underlying cause of both the conditions.

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