A Case of Adult Onset Barrter Syndrome - A Rare Entity

Dear sir,

In 1962, Bartter *et al*.[1] described a clinical condition with a rare autosomal recessive renal tubulopathy characterized by decreased salt reabsorption in thick ascending limb of the loop of Henle, leading to renal salt wasting, hypokalemia, metabolic alkalosis, low-to-normal blood pressure, hyper-reninemic hyperaldosteronism, resistance to pressor effect of angiotensin, and hyperplasia of juxtaglomerular apparatus. Most cases present antenatally or in early childhood, and presentation in adults is very rare.[2] A 56-year-old gentleman, a known case of coronary artery disease with normal cardiac function post stenting, presented to us with weakness of all four limbs for a few months. There was no history of fever, weight loss, vomiting, diarrhea, polyuria or nocturia, and passage of stone. The patient denied history of use of laxatives, diuretics, and over-the-counter or herbal medicines. There was no history of exacerbation of symptoms with exercise or excessive carbohydrate intake. There was no family history of similar complaints. On examination, his pulse was 90/min, blood pressure was 90/60 mmHg, and there was no edema with normal power and reflexes. Other systemic examination was normal. His previous records showed persistent hypokalemia (2.5–3.2 mEq/L). His Blood urea was 21 mg/dl, creatinine 0.9 mg/dl, sodium 131 mmol/l, potassium 3.1 mmol/l, chloride 91 mEq/l, calcium 9.4 mg/dl, magnesium 2.1 mg/dl, phosphorus 2.4 mg/dl, and uric acid was 5.6 md/dl. Arterial blood gas showed a pH of 7.56, pCO_2 42, pO_2 83, and HCO3-33, with a urine pH of 5.

There was no proteinuria, hematuria, or active urinary sediment. Twenty-four-hour urinary excretion of sodium was 294 mEq (40–220 mEq), potassium 152 mEq (25–125 mEq), chloride 293 mEq (110–250 mEq), and calcium 120 mg (100–300 mg), with urine osmolality of 464 mOsm/kg. His fractional excretion of sodium (FE_{Na}) was 2.1%, fractional excretion of potassium (FE_K) was 22%, and his fractional excretion of chloride (FE_{Cl}) was 2.3%, all of which were more than the reference range. His transtubular potassium gradient (TTKG) was 12, which was high indicating renal potassium wasting. In view of chronic hypokalemia, further investigations showed that serum renin was 432.20 mIU/ml (range, 4.40–46 mIU/ml), and serum aldactone of 49.10 ng/dl (range, 2.52–39 ng/dl), which were both high. Ultrasound showed normal-sized kidneys, and echocardiography was normal. Our patient had chronic hypokalemia, hypotension, metabolic alkalosis with acidic urine, normal serum magnesium, high urinary sodium, potassium and chloride loss, normal calcium loss, and hyper-reninemic hyperaldosteronism in the absence of diuretic or laxative intake. The diagnosis of Bartter syndrome was made, and the patient was started on a potassium rich diet, potassium replacement, tab. aldactone 50 mg twice a day, and tab. indomethacin once a day. His repeat investigations done a month later showed serum K + 4.2 mEq/l, his 24-hour urinary potassium was 70.3 mEq, calcium 108 mg, sodium 233 mEq, and chloride was 235 mEq. Repeat ABG done was normal.

Bartter syndrome is a group of autosomal recessive genetic disorder whose primary pathogenic mechanism is defective transepithelial chloride reabsorption in the thick ascending limb of the loop of Henle, and is characterized by hypokalemia, metabolic alkalosis, hyper-reninemia, hyperaldosteronism, and hyperplasia of the juxtaglomerular apparatus. Five types of genetic mutations are associated with the five different forms of the disease (3). Types I, II, and IV are classified as antenatal Bartter syndrome and are usually present in the neonatal period. Type III is known as the classical Bartter syndrome, usually presents in the first two years of life, although can be present in childhood and adolescence.[4] Another related tubulopathy is Gitelman syndrome characterized by hypokalemic metabolic alkalosis with hypomagnesemia and low urinary calcium excretion, which is more common in adults. It is an autosomal recessive disorder of thiazide-sensitive sodium chloride cotransporter.[5]

Our patient was a middle-aged man with generalized weakness, hypotension, and persistent hypokalemia. His blood gas analysis showed metabolic alkalosis with acidic urinary pH and high potassium loss in urine. Despite having clinical hypotension, our patient had increased sodium and chloride excretion with hypokalemia and metabolic alkalosis. Although we expected low urinary potassium loss and alkaline urine, the patient had high potassium loss and acidic urine with a TTKG of 12, which was consistent with hyperaldosteronism, either primary or secondary. The patient had high renin and aldosterone levels, normal cortisol, and Adrenocorticotrophic hormone. All these were suggestive of a secondary cause for aldosteronism, either gastrointestinal loss or renal loss. Surreptitious vomiting and diuretic use are the two other major causes of unexplained hypokalemia and metabolic alkalosis. Our patient lacked any history of vomiting and diuretic abuse, and investigations revealed high renal loss of sodium, potassium, and chloride, normal serum magnesium, and normocalciuria, all of which were suggestive of a tubulopathy. Bartter and Gitelman syndrome are two types of renal salt wasting diseases. Although Gitelman disease is more common in adults, but our patient had normal serum magnesium. Bartter type phenotype has also been reported with diuretic and laxative abuse, bulimia, gentamycin nephrotoxicity,[6] colistin toxicity,[7] chronic sialedinitis,[8]

Sjogren syndrome, and tuberculosis.[9] In our patient, there was no history or finding suggestive of any underlying condition, and hence, we reported it as idiopathic Bartter syndrome. Though Bartter syndrome is diagnosed at a young age, an adulthood presentation is possible due to phenotypic variation. The patient was started on treatment and there was reversal of metabolic abnormality.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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