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RESEARCH ARTICLE

AN UNCOMMON PRESENTATION OF BARTTER SYNDROME IN AN ADULT: A CASE REPORT

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Abstract

Bartter syndrome is an uncommon autosomal recessive renal tubular condition that is usually diagnosed in childhood; there are few reports of adult-onset cases and even fewer that are correctly diagnosed. We present a 48-year-old woman with no co-morbidities who was seen with generalized weakness, fatigue, carpopedal spasms and tingling in both hands, muscle cramps, and frequent urination. Clinical assessment showed a positive Trousseau's sign, and laboratory tests established hypokalemia, metabolic alkalosis, hyper reninism, hypocalcemia and hypercalciuria with renal biopsy showing juxtaglomerular hyperplasia. The patient was treated with intravenous potassium and calcium supplementation, ACE inhibitors, NSAIDS, followed by oral therapy, dietary counseling. She stayed asymptomatic on follow-up. Adult-onset Bartter syndrome is a rare clinical phenomenon that must be kept in mind in the differential diagnosis of unexplained metabolic alkalosis and hypokalemia, particularly in normotensive individuals. Timely diagnosis and proper treatment may result in outstanding outcomes. This case highlights the significance of recognizing late-onset variants and distinguishing them from other tubulopathies like Gitelman syndrome.

Introduction:-

Bartter syndrome is a collection of rare autosomal recessive hereditary renal diseases with the characteristics of hypokalemia, metabolic alkalosis, normal or low blood pressure and hyperreninemic hyperaldosteronism. It is caused by abnormal salt reabsorption in the thick ascending limb of the loop (TAL) of Henle, causing urinary loss of excessive amounts of sodium, potassium, and chloride. There are five distinct sub types that have been reported due to mutations in various ion channel or transporter genes like SLC12A1, KCNJ1, BSND, CLCNKB, and CASR.

•**Type I:** Resulting from mutations in the SLC12A1 gene encoding the sodium-chloride-potassium cotransporter in the kidney.
•**Type II:** Resulting from mutations in the KCNJ1 gene encoding the ROMK potassium channel in the kidney.
•**Type III**: Due to mutations in the CLCNKB gene, which provides instructions for making the chloride channel Kb in the kidney.
•**Type IV**: Due to mutations in the BSND gene, which gives instructions for making barttin, an accessory subunit of the chloride channels. Type IV is also linked to sensorineural deafness.

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•**Type V:** Due to mutations in the CASR gene, which provides instructions for making the calcium-sensing receptor. Certain variants of Bartter syndrome occur antenatally with severe hypokalemia, metabolic alkalosis, and severe systemic presentation.Bartter syndrome III and V typically present later in life and are mildly symptomatic.Bartter syndrome occurs in 1 in 1,000,000 people and is far less frequent than Gitelman syndrome. And there is an adult form of it as well, which is even less common to encounter.

Adult-onset Bartter syndrome is uncommon and can be caused by milder or partially protective mutations that slow the onset of symptoms. Such patients usually have nonspecific presentations in the form of generalized weakness, muscle cramp, and electrolyte disturbances, which can be mistakenly referred to other more prevalent diseases.

Where resources are limited, such as in India, a clinical diagnosis by exclusion of secondary causes of electrolyte disturbances based on typical laboratory results may be necessary due to a lack of access to high-tech genetic testing.

Symptoms can be dramatically improved and long-term consequences such as nephrocalcinosis or chronic kidney disease avoided with early diagnosis and treatment involving correction of the electrolyte deficiency, inhibition of prostaglandin synthesis, and potassium-sparing diuretics. This case report describes a rare case of adult-onset Bartter syndrome in a 48-year-old female, highlighting the importance of awareness of this entity by clinicians despite the absence in the classical pediatric age group.

Case Presentation:

•A 48 year oldfemale presented to Dhiraj hospital with the chief complaint of generalised weakness and fatigue since 15 days which was gradual in onset.She also presented a history of intermittent recurrent carpopedal spasms bilaterally since 10 days. It was accompanied by tingling in both hand, muscular cramp and frequent micturition. She was not a known case of diabetes, hypertension or thyroid disorder. She had no history of diarrhoea, vomiting, nausea, fever, starvation, similar complains in the past, no history of intake of any drug, no history of paralysis/paresis of limbs, recurrent urinary tract infections, fractures, disturbances in hearing, or loss of vision. There was no family history of renal diseases. She was admitted in medicine ward and examined and investigated. Patient was conscious and oriented, moderately built and well nourished. The heart rate was 78/min, BP was 110/70 mmhg and respiratory rate was 18/min. Trousseau's sign came positive. Lab tests showed hypokalemic alkalosis with hypocalcemia, hyper reninism and hypercalciuria, with serum potassium-2.8mEq/L (3.5-5), arterial blood gas analysis- pH- 7.54, paO2- 78 mmHg, HCO3- 30mEq/L, pCo2- 24 mmHg, calcium- 5.9mEq/L (9-11),plasma renin activity 3.95 ng/ml/hr (1.9-3.7 ng/ml/hr). Sodium-134mEq/L (135-145), chloride-94mEq/L (96-106), magnesium-1.6mEq/L (1.7-2.2), phosphorous- 4mg/dl (3.5-4.5), uric acid 13.4 mg/dl, serum creatinine- 1.6mg/dl, urea-10mg/dL, parathyroid hormone- 30 (10-60), Vit D levels within normal range. 24 hours urinary potassium was 130 mEq/gmcreat (normal range 13-116), urinary calcium- 284 mg/gm, creat(normal range 12-244), urinary calcium:creatinine ratio- 0.28 (normal <0.14) indicated of hypercalciuria, urine specific gravity- 1.030. S.Aldosterone was 3.20 pg/ml/hr (2.5-3.1 pg/ml/hr). ECG showed inverted T waves with prolongation of QT. Relevant investigations of blood and serum showed normal hemoglobin, hematocrit, white blood cell count, total protein, albumin, alkaline phosphatase and transaminase.

Her Immunological profile (ANA, dsDNA, Anti Sm, and Anti Ro/La) and viral markers (HIV, HbsAg, and HCV) were negative. Ultrasound abdomen suggestive bilateral renal concreations with normally sized kidneys.

Percutaneous kidney biopsy was done from the lower pole of the left kidney having 5 glomeruli with juxtaglomerular hyperplasia, renal tubules with peri tubular fibrosis andinterstitial inflammation.

Figure 1:- Of Trousseau's sign positive-carpopedal spasm.

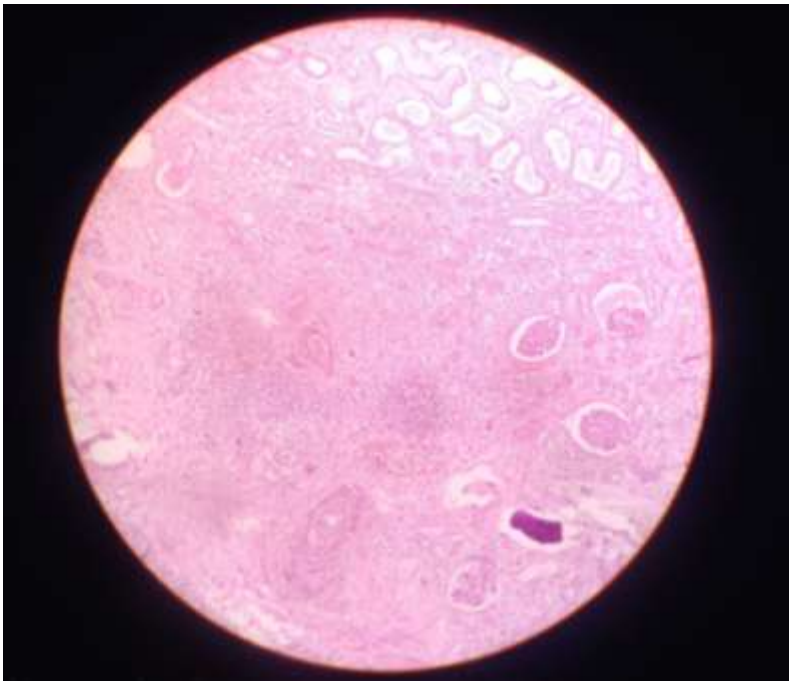

Figure 2:- Demonstrating renal biopsy with H&E stain in Low power field with findings penned above.

Results were in keeping with hypokalaemic metabolic alkalosis, renal potassium wasting, hyperreninemia, hyperaldosteronism, and salt-wasting — all characteristic features of Bartter syndrome. Adult-onset is attested by similar reported cases in which biopsy showed juxtaglomerular hyperplasia.

The patient was then given intravenous potassium (40 mEq twice daily) and calcium supplementation and supportive care till patient was symptomatically improved. Once her electrolytes were normalized, she was discharged on oral potassium chloride (750 mg thrice daily), calcium carbonate (500mg thrice daily), enalapril, 2.5

mg daily and indomethacin, 25 mg thrice a day along with instructions for adequate hydration and sodium and potassium-rich foods.

The patent was called on follow up after one month of discharge and was seen to have improvement of symptoms and no episodes of carpopedal spasms.

Table 1:- Clinical manifestations before and after treatment .

Clinical Manifestation	Before treatment	After treatment
Weakness	+	-
Fatigue	+	-
Carpopedal spasms	+	-
Muscle cramps	+	-
Polyuria	+	-
Growth retardation	-	-
Short stature	-	-
Failure to thrive	-	-
Salt craving	+	-
Tingling numbness	+	-
Sensorineural deafness	+	-
Constipation	-	-

Discussion:-

Bartter syndrome is a collection of rare autosomal recessive tubular renal disorders that are associated with defective sodium, potassium, and chloride reabsorption within the thick ascending limb (TAL) of the loop of Henle.

The underlying defect causes chronic renal salt loss, activation of the RAAS, and resulting hypokalemia, metabolic alkalosis, hyperreninemia, and secondaryhyperaldosteronism, usually in the context of normal or low blood pressure. The condition can also be accompanied by hypercalciuria and increased urinary prostaglandins. Bartter syndrome classically presents in infancy or early childhood with typical features of polyuria, dehydration, growth retardation, failure to thrive, vomiting, and in neonatal presentation forms that are more severe (Types I and II), polyhydramnios and prematurity.

Adult-onset Bartter syndrome, however rare, has a more subtle presentation. They present with nonspecific symptoms like paraesthesia, cramps, fatigue, nocturia, salt craving, or orthostatic hypotension, and usually remain misdiagnosed or underdiagnosed for years. The adult phenotype is most commonly due to less severe genetic mutations or partially compensatory transporter function that postpones the onset of symptoms. In our experience, a 48-year-old female patient came to us with generalised weakness, fatigue, muscle cramps, carpopedal spasm and polyuria, and also with biochemical evidence of hypokalemia, metabolic alkalosis, increased renin and aldosterone levels, and loss of potassium and calcium in the urine. The renal biopsy, which showed juxtaglomerular hyperplasia, helped to further establish the diagnosis. Notably, she had no diuretic abuse, gastrointestinal fluid loss or systemic disease, thereby ruling out secondary causes of electrolyte disturbance.

One of the critical challenges in the Indian healthcare environment is restricted access to cutting-edge diagnostic equipment, such as genetic testing. Under these conditions, physicians have to depend on a synthesis of clinical presentation, elaborate history-taking, and focused biochemical investigations to reach a diagnosis. This case highlights the worth of clinical judgment under low-resource conditions, wherein stepwise diagnosis is still essential.

Treatment involves reversing electrolyte deficiencies, downregulating RAAS activity, and preventing renal prostaglandin-mediated salt loss. Intravenous and oral potassium and calcium supplementation, dietary therapy, ACE inhibitors and NSAIDS helped our patient.

This case contributes to the sparse literature on adult-onset Bartter syndrome in India and underscores the need to look for inherited tubulopathies in adults with chronic hypokalemia and metabolic alkalosis.

Conclusion:-

Adult-onset Bartter syndrome, while uncommon, must be part of the differential diagnosis in unexplained hypokalemia, normotension, and metabolic alkalosis presenting patients.

Early identification and tailored therapy are important in avoiding complications like nephrocalcinosis and chronic kidney disease. In economically disadvantaged countries such as India, clinical diagnosis using simple biochemical studies is both effective and possible. This case underlines the importance of clinical suspicion and the need for the identification and management of uncommon tubular illnesses in adult patients.

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