CASE REPORT

Long-term survival with congenital Bartter syndrome: A case report

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Abstract

Bartter syndrome is a rare congenital salt-wasting renal tubular disorder, caused by defective salt reabsorption in the thick ascending limb of the loop of Henle. It is characterized by hypokalaemia, hyponatraemia, hypochloraemia, metabolic alkalosis, hyper-reninaemia, and hyperaldosteronism.

The case of a 26 years old male, resident of Karachi, Pakistan, survivor of congenital Bartter Syndrome, with regular quarterly follow up is presented. At the age of 3 years, he got severe vomiting and diarrhoea and developed hypovolaemic shock. He was investigated and clinically diagnosed with Bartter Syndrome based on serum electrolytes imbalance (hypokalaemia/ hyponatraemia/ hypochloraemia), arterial blood gas analysis (metabolic alkalosis) and raised serum renin and aldosterone levels. Urine analysis also showed elevated sodium, potassium, chloride, calcium, and prostaglandin E2 excretion. After diagnosis, the patient was managed with oral indomethacin and potassium supplements during follow up visits. This is the first reported case of a long-term survivor of congenital Bartter syndrome from Pakistan.

Keywords: Bartter Syndrome, polyuria, hypokalaemia, hypochloraemia, metabolic alkalosis.

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Introduction

Bartter's syndrome (BS) is a rare congenital salt-wasting renal tubular disease that affects approximately 1 in 1,000,000 population.[1] It is characterized by polyuria, polydipsia, hypokalaemia, hyponatraemia, hypochloraemia, metabolic alkalosis, hyperreninaemia, and hyperaldosteronism.[1] It is caused by defective salt reabsorption due to mutations in genes encoding sodium, potassium, or chloride transporters in the thick ascending limb of the loop of Henle.[2] It is classified into two types based on the phenotype, neonatal (antenatal) and adolescent or adult-onset (classical). Bartter syndrome is classified into five subtypes based on genotype.[3] It usually presents in neonates, with vomiting, dehydration, and failure to thrive.[3] Bartter syndrome is associated with increased antenatal and neonatal morbidity and mortality, because many patients fail to thrive. The diagnosis of Bartter syndrome is established either clinically or through genetic sequencing. Clinical diagnosis based on serum electrolyte imbalance (hypokalaemia, hyponatraemia, hypochloraemia), arterial blood gas analysis (metabolic alkalosis), and on raised serum renin and aldosterone levels. Administration of potassium chloride, a prostaglandin inhibitor, and a potassium-sparing diuretic are effective treatment modalities.[3]

Currently, the presenting features and initial management of Bartter syndrome (BS) are well described; however, long-term treatment and survival outcomes remain inadequately studied.[4,5] We report a case of long-term survival in a patient with Bartter Syndrome who was first seen on 19th March 2009 in the Paediatric OPD of Kharadar General Hospital, Karachi and has remained under follow up for the past 16 years.

Case report

A 26 years old male, resident of Karachi, Pakistan, survived congenital Bartter Syndrome, with regular quarterly follow up. The patient's father was a factory worker who died of hepatitis B in 2020 at the age of 54 years. The patient has four living siblings (two sisters and two brothers), while two siblings (twin sisters) died at the age of seven years with the similar sign and symptoms. Currently, the patient has a body weight of 62 Kg and a height of 178 cm, with a normal BMI (19.6 kg/m2). He lived a normal life with no limitation of physical activity and is self-employed, providing financial support for his family.

Initially, at the age of one month, he clinically presented with recurrent vomiting, diarrhoea, and failure to thrive. He was treated by a local physician at a private healthcare facility with water and electrolytes replacement as a case of gastroenteritis. At the age of 3 years, he had stunted

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Figure: Survived Congenital Bartter Syndrome Patient.

growth, weak muscles, and inability to sit without support. He got severe vomiting and diarrhoea and developed hypovolaemic shock for which he was admitted to Aga Khan University Hospital (AKUH), Karachi. At AKUH, he was investigated and clinically diagnosed as a case of Bartter Syndrome, based on serum electrolytes imbalance (hypokalaemia/ hyponatraemia/ hypochloraemia), arterial blood gas analysis (metabolic alkalosis), and raised serum renin and aldosterone levels. After diagnosis, the patient was managed with oral indomethacin and potassium supplements.

From March 2009 to date, the patient made quarterly follow-up visits at the paediatric OPD of Kharadar General Hospital, Karachi. On each quarterly follow-up visit, serum potassium level is monitored and on each annual visit, serum renin levels and renal ultrasound monitored. Adjustments of potassium and Indomethacin doses were made according to the body weight and serum potassium and renin levels.

In 2020, at the age of 22 years, the patient again experienced symptoms of muscle weakness and cramps, prompting adjustments of the indomethacin dose. As symptoms persisted, treatment was changed to oral Ibuprofen, resulting in symptomatic improvement and stabilization of serum renin and potassium levels. His renal ultrasound revealed grade 1 renal parenchymal disease without evidence of Nephrocalcinosis.

In 2023, during a follow-up visit, the patient complained of chest pain and the event of losing consciousness three days earlier. Additionally, the attendant reported significant weight loss over an unspecified duration. Clinical assessment and the attendant's history revealed suboptimal compliance with the prescribed treatment plan, which contributed to the patient's current symptoms. Laboratory investigations confirmed that the patient's potassium level was below the normal limit, likely due to inconsistent adherence to the treatment regimen.

To address these concerns, the patient continued on (Ibuprofen) for symptomatic relief, and Neo-K (Potassium Chloride) was prescribed to correct the hypokalaemia associated with Bartter syndrome. In addition, the healthcare team provided counselling to the patient and their family, emphasizing the importance of compliance with the treatment plan to prevent complications effectively and improve the patient's overall condition.

In 2024, the patient's treatment plan was carefully reviewed, and previously prescribed medications were continued. The overall therapeutic approach remained focussed on symptom management and improving the patient's quality of life.

Discussion

Bartter syndrome (BS) is a rare salt-wasting tubulopathy caused by gene mutations in sodium, potassium, or chloride transporters or channels responsible for sodium chloride reabsorption in the thick ascending limb of the loop of Henle.[2]

Bartter syndrome was first described by Dr. Fredric Bartter in 1962 in two African Americans who presented with metabolic alkalosis, hyperplasia of the juxtaglomerular apparatus, and normotensive hyperaldosteronism.[6] These patients were different from the typical patients with hyperaldosteronism because they were younger, had normal blood pressure, and had growth retardation[6]. In this report, we describe a case of long term follow-up and survival of a patient with Bartter syndrome from Pakistan. Only a limited number of studies on BS have been reported from Pakistan.[7,8] Among these, mostly reported were Neonatal Bartter syndrome cases[7] except one study that reported the adult case of BS8. We present a unique case that was clinically diagnosed during early childhood at the age of 3 years, and then continuously remained under follow-up until reported.

Bartter Syndrome can lead to long-term complications like chronic kidney disease (CKD) due to chronic hypokalaemia, Nephrocalcinosis, developmental delays and growth impairments.[2,9] The present case did not

exhibit any evidence of CKD; however, signs of developmental delay and growth impairment were observed during childhood, which gradually improved (in terms of body weight and height) over time with long-term follow-up. Currently, there is no definitive cure for BS, treatment mainly focus on correcting hypokalaemia, metabolic alkalosis, supplementing potassium therapy, and reducing potassium loss.

Gitelman syndrome and cystic fibrosis may present as a pseudo-Bartter syndrome, with overlapping clinical features. Gitelman syndrome is also a rare, recessive, salt-wasting tubulopathy, but with impaired salt reabsorption in the distal convoluted tubule (DCT). Previously, Bartter syndrome has been differentiated from Gitelman syndrome by some clinical features, such as early onset, severity, presence of hypercalciuria, polyhydramnios, or growth retardation. However, recent research has revealed that Bartter syndrome patients might experience delayed presentation, and some characteristics, including hypercalciuria, are not always present.[10] Cystic fibrosis and Bartter syndrome are rare hereditary diseases with overlapping clinical manifestations such as dehydration, failure to thrive, and similar electrolyte abnormalities.[11]

The current case of BS was diagnosed on clinical grounds; genetic sequencing was not performed due to the unavailability of a specialized laboratory in the country. Antenatal and early postnatal data were unavailable. Bartter's Syndrome is a much-neglected disease in Pakistan, as physicians often consider it extremely rare and may not prioritize its diagnosis.[12] All the children and young adults presenting with failure to thrive, polyuria, polydipsia and hypokalaemia must be investigated for BS. Failure to do so may result in a significant number of cases being missed, leading to delays in diagnosis and management. Early diagnosis and treatment are essential to improve the prognosis and prevent various complications.

Conclusion

This is the first case of long-term survivor of congenital Bartter syndrome from Pakistan. It was diagnosed clinically on the bases of clinical presentation (recurrent vomiting, diarrhoea, and failure to thrive), serum electrolytes imbalance (hypokalaemia/ hyponatraemia/ hypochloraemia), arterial blood gas analysis (metabolic alkalosis), and raised serum renin and aldosterone levels. Case managed by oral indomethacin and potassium supplements. Bartter Syndrome is a neglected disease; it should be suspected in any child with history of failure to thrive and metabolic alkalosis. Early diagnosis, long term regular follow up and treatment adherence can improve the prognosis and prevents from complications.

Consent to Publish: The document signed by the patient was submitted for publishing the case report.

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AUTHORS' CONTRIBUTIONS:
GB: Concept, design, identify uniqueness and final approval.
MA: Literature search and writing.
KI: Evaluate past and present physical statute & well-being and final approval.
RH: Revision, validated the history and physical examination.
AM: Responsible for consultant order and clinic follow-up.
SW: Patient care during stay in hospital and after discharge, coordinated with Bartter syndrome patient and hospital and administration for hospital visits.