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Case Report

Hypotonia an unusual childhood presentation of Bartter syndrome

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ABSTRACT

Bartter syndrome is a congenital functional renal anomaly, characterized by hypokalemic metabolic alkalosis with renal salt wasting with normal blood pressure. It presents in infancy and early childhood age group with failure to thrive and episodes of polyuria and dehydration.

Keywords: Bartter syndrome, Childhood presentation of Bartter syndrome, Hypotonia

INTRODUCTION

Bartter syndrome is a rare autosomal recessive renal tubular disorder characterized by hypokalemic, hypochloraemic metabolic alkalosis, normotension, hyperreninaemic hyperaldosteronism, hypercalciuria, prostaglandinuria and increased urinary loss of sodium, potassium and chloride.[1] The exact incidence of Bartter syndrome is not known. In Costa Rica, incidence of neonatal Bartter syndrome from live births is estimated as one per 1.2 million.[2]

Most cases of Bartter syndrome present in neonates. Usual clinical manifestations are failure to thrive, polyuria and episodes of dehydration. We report an unusual case of Bartter syndrome presented with hypotonia.

CASE REPORT

2-year-6-months old boy brought to us with loss of tone (muscle weakness) in the form of not able to lift head and limbs from bed for 2 days. Child had acute gastroenteritis 8 days back and was treated elsewhere. He was born of non-consanguineous marriage, at term, without history of poly-hydramnios, antenatal ultrasound scan was normal. Achieved all developmental milestones appropriate for age, was not on any medications, and was vaccinated to date. On examination child was conscious, vitals stable, normal blood pressure, no facial dysmorphism, generalized hypotonia with power 2/5 at neck and all limbs with normal reflexes. On pull to sit child had head lag but no localizing signs on neurological examination. He had growth retardation, weight was 9.0 kg (z score -3), height was 91 cm (z score 0).

Preliminary investigations revealed severe hypokalemia (potassium 2.0 mmol/l) with normal other blood investigations. Emergent management of hypokalemia was instituted with intravenous extra potassium, serum potassium level improved but not satisfactory and he had persistent hypokalemia. So, on further investigation arterial blood gas analysis suggested metabolic alkalosis (PH - 7.54, HCO_3 - 35 mEq/L) and he had hypomagnesemia (magnesium 1.34 mg/dl). Urinary investigation revealed raised urinary chloride 332 mEq/L, hypercalciuria (calcium 22.39 mg/dl), high urinary sodium and potassium and raised urinary calcium:creatinine ratio 1.69. Ultrasound abdomen was normal (no nephrocalcinosis). Plasma renin activity was markedly raised 12.6 ng/ml/hr. Molecular study revealed CLCBRK gene mutation, the child was thus diagnosed as a case of classic Bartter syndrome (type III Bartter syndrome). The child was treated with potassium and

magnesium supplements with improvement in muscle tone and electrolyte status. Child was discharged on supportive care, potassium supplements and dietary advice, with clinical and biochemical improvement. The baby is thriving well and has normal muscle tone on follow up.

DISCUSSION

Bartter Syndrome is an inherited renal tubular disorder, first described in 1960 but, over the years several phenotypic and genotypic variants of original description of Bartter syndrome have been identified.[3,4] It was traditionally classified as neonatal, classical and Gitelman syndrome. It is caused by mutations of genes encoding proteins that transport ions across renal cells in the thick ascending limb of loop of Henle (TALH).[5] Abnormalities of Bartter syndrome are all suggestive of a defect related to Cl - transport in the medullary thick ascending loop of Henle and distal convoluted tubule (DCT). Failure to reabsorb chloride results in a failure to reabsorb sodium and leads to excessive sodium and chloride (salt) delivery to the distal tubules, leading to excessive salt and water loss from the body and activation of renin-angiotensin aldosterone system (RAAS).

Bartter syndrome is characterized by hypokalemia, hypochloremic metabolic alkalosis, hyperreninemia, hyper-prostaglandinism, normal blood pressure, with increased urinary loss of sodium, chloride, potassium, calcium and prostaglandins.[3] The onset of Bartter syndrome may be during the neonatal period, infancy or childhood. Antenatal Bartter syndrome (type I, II and IV) typically manifests in infancy and has a more severe clinical presentation than classic Bartter syndrome (type III) which presents during childhood.

Antenatal features of Bartter syndrome consist of polyhydramnios and premature delivery. Other clinical features include polyuria, failure to thrive and distinctive appearance with thin, triangular face, prominent forehead, large eyes, protruding ears, drooping mouth, strabismus, sensorineural deafness (type IV Bartter), convulsions and increased susceptibility to infections.[6] Children with classic Bartter syndrome commonly present during the first 2 years of life as polyuria, polydipsia, vomiting, salt craving, tendency to dehydration, lethargy, developmental delay and failure to thrive. The neonatal period usually passes without major problems.

Treatment of Bartter syndrome includes prevention of dehydration, maintain good nutrition, correct electrolyte imbalance. Apart from potassium supplementation, administration of indomethacin after 6-12 weeks of life is beneficial. Indomethacin (dose 1-5 mg/kg/day) is most frequently used.[7] Other drugs used are acetylsalicylic acid (100 mg/kg/day), Ibuprofen (30 mg/kg/day). Addition of potassium sparing diuretics may be initially effective in the control of hypokalemia but their effect is transient. However, Type IV Bartter syndrome responds poorly and it rapidly progresses to end stage renal disease (ESRD).[8]

CONCLUSION

With close attention to electrolyte balance, volume status and growth, the long-term prognosis is generally good. In a minority of patients, chronic hypokalaemia, nephrocalcinosis, and chronic indomethacin therapy can lead to chronic interstitial nephritis and chronic renal failure. Poorly managed patients may develop a progressive tubulointerstitial nephropathy that can lead to terminal chronic renal failure. As Bartter syndrome is an autosomal recessive inherited disorder, genetic counselling should be offered to the families.[9]

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