Bartter's Syndrome Masquerading as a Neurometabolic Disorder

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Introduction

Fredrick Bartter first described Bartter's syndrome in 1962 [1]. It is a rare autosomal recessive disorder of the renal tubules resulting in hypokalemic metabolic alkalosis, hyperreninism and hyperaldosteronism with normal blood pressure. This disorder has a varied spectrum of presentation [2]. We report a case of Bartters syndrome presenting as a neurometabolic disorder.

Case Report

A four month male infant with uneventful birth history presented with failure to thrive, recurrent vomiting, feeding difficulty and an episode of pneumonia since one month of age. There was no history of maternal polyhydramnios in the antenatal period. On clinical evaluation, the infant weighed 2.8 kilograms, had a protruding tongue, failure to thrive, severe generalized hypotonia, weakness of all four limbs, chest retractions, stridor and dehydration (Fig.1). He had an episode of cardio-respiratory arrest in hospital from which he was successfully resuscitated. On suspicion of a neurometabolic disorder, the child was appropriately investigated. Initial investigations revealed mild acidosis, raised serum ammonia of 156 mcg/dL (normal values 25- 94 mcg/dL), normal serum electrolytes and normal renal functions (Table 1). Subsequent neuromuscular evaluation revealed a normal

electromyography and nerve conduction velocity (motor and sensory). In view of raised ammonia and mild acidosis, an inborn error of metabolism (IEM) screening was done for defects in metabolism of aminoacids, fatty acids, urea cycle and deficiency of biotinidase by tandem mass spectrography,

Fig. 1 : Bartter's syndrome masquerading as a neurometabolic disorder

Table 1

Serum pH and Electrolytes

Investigations	On admission	After correction of dehydration	After treatment with indomethacin
Serum pH	7.20	7.6	7.3
Serum K ⁺	3.9 meq/L	2.9 meq/L	3.8 meq/L
Serum Na ⁺	135 meq/L	118 meq/L	136 meq/L
Serum Cl ⁻	96 meq/L	92 meq/L	101 meq/L
Urine chloride	122 meq/L	212 meq/L	25 meq/L

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all of which were negative.

At this point it was noted that during the episode of vomiting, the child maintained near normal urine output and the dehydration was out of proportion to vomiting. A repeat blood gas and electrolyte study after adequate hydration revealed hypokalemic metabolic alkalosis with hyponatremia, hypochloremia and hypocalcaemia (6.2mg/dL). Further investigations revealed a normal 24 hour urine calcium / creatinine ratio (0.17). However, plasma rennin activity was markedly raised (19.99ng/ml/hr) (normal values 1.31- 3.95 ng/ml/hr). Serum magnesium (1.8mEq/L) was within normal limits thereby differentiating it from Gitelman Syndrome. The child was thus diagnosed as a case of Bartter's syndrome. The child was treated with potassium supplements and indomethacin (5mg/kg) with improvement in muscle tone, hydration and electrolyte status. The baby is thriving well, achieving milestones normally and has normal muscle tone on follow up.

Discussion

A diagnosis of Bartter's syndrome was entertained based on failure to thrive, dehydration, polyuria, normotension with hypokalemia. There are four variants of Bartter's syndrome based on the age of presentation [3, 4]. Our patient was a type III Bartter's syndrome which usually presents in late infancy or childhood with polyuria, polydipsia, salt craving, tetany, failure to thrive and dehydration. However there have been reports of an early, atypical presentation and complications. During episodes of dehydration the typical features of Bartter's syndrome i.e. hypokalemic, metabolic alkalosis may not be obvious. Instead, child may have acidosis as reported in a five week old infant where alkalosis was first seen after two weeks of vigorous fluid and electrolyte replacement as in our case [5]. The baby manifested with an acute complication of severe hypokalemia after correction of dehydration, in the form of cardiorespiratory arrest as reported in literature [6 - 8]. The patient also had a protruding tongue without obvious dysmorphism suggestive of a neurometabolic disorder but a case series reported Bartter's syndrome with triangular facies, protruding tongue and large rounded eyes [9]. There was no evidence of hearing deficit suggestive of a type IV Bartter's syndrome. A kidney biopsy usually shows

juxtaglomerular hyperplasia. Management consists of potassium supplements and indomethacin. Spironolactone may be added if response is poor. However Bartters syndrome with sensorineural deafness (Type IV) responds poorly and it rapidly progresses to end stage renal disease (ESRD) [3].

On follow up at two months, the child was asymptomatic with satisfactory weight gain. However, the long-term prognosis in these cases is guarded. The problems anticipated in this child are related to indomethacin therapy either in the form of interstitial nephritis due to long term administration or due to poor compliance with frequent relapses and slow progression to chronic renal failure.

Conflicts of Interest

None identified

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