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Not All Failure to Thrive Is Severe Acute Malnutrition: A Case of Suspected Bartter Syndrome with Nephrogenic Diabetes Insipidus and Severe Acute Malnutrition

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ABSTRACT

This case highlights a diagnostically challenging presentation of Bartter syndrome in a malnourished child with a background of tuberculosis and polyuria-polydipsia, emphasizing the diagnostic complexity and therapeutic management in resource-limited settings.

Keywords: *Bartter Syndrome, Malnutrition, Polyuria*

CASE REPORT

A 2-year-6-month-old girl, second child of a third-degree consanguineous marriage, developmentally normal and immunized according to the National Immunization Schedule, presented with a 6-day history of fever and rash. Past medical history of hospitalization was noted at 8 months with complaints of fever, vomiting, diarrhea, and hypokalemia, and at 18 months, she was diagnosed with failure to thrive secondary to clinical suspicion of tuberculosis (PPD positive, CBNAAT negative), treated with a six-month anti-tubercular regimen. A sibling had posterior urethral valves requiring surgery.

On detailed history, the mother reported that the water intake of the child was approximately 3 liters/day, and urination occurred within 5 minutes of drinking. Examination revealed lethargy, dehydration (sunken eyes, dry mucosa), lustreless hair, and severe acute malnutrition (weight and height <3rd percentile). Systemic examination was normal.

Suspecting Type 1 diabetes mellitus, random blood glucose and HbA1c levels were evaluated and were normal, and renal function and magnesium levels were normal. Other lab investigations revealed severe hypokalemia (1.6 mmol/L), mild hyponatremia, and metabolic alkalosis. ECG showed flat T-waves (Fig. 1). Urinary electrolytes demonstrated low potassium excretion, suggesting renal tubular dysfunction. A provisional diagnosis of Bartter syndrome was made, with differentials including Gitelman syndrome, nephrogenic diabetes insipidus, and chronic interstitial nephritis. Investigations for renin, aldosterone, and vasopressin response were planned but deferred due to financial constraints. The child was managed in the pediatric intensive care unit with intravenous and then oral potassium, indomethacin (3 mg/kg/day), and hydrochlorothiazide (2 mg/kg/day) to improve potassium reabsorption. Nutritional rehabilitation was initiated.

Electrolyte balance improved (serum potassium 3.4 mmol/L), and polyuria decreased. She was discharged in stable condition with follow-up planned for genetic testing, growth monitoring, and electrolyte surveillance.

Table 1:

Characteristic	Type 1	Type 2	Type 3	Type 4a	Type 4b	Type 5
Gene	*SLC12A1*	*KCNJ1*	*CLCNKB*	*BSND*	*CLCNKA* + *CLCNKB*	*MAGED2*
Protein	NKCC2	KCNJ1 (ROMK or Kir1.1)	CIC-Kb	Barttin	CIC-Ka + CIC-Kb	MAGE-D2
Inheritance	AR	AR	AR	AR	AR	XLR

Table 2:

Characteristic	Type 1	Type 2	Type 3	Type 4a	Type 4b	Type 5
Age at onset	Prenatally	Prenatally	0–5 years	Prenatally	Prenatally	Prenatally
Polyhydramnios	Severe	Severe	Absent or mild	Severe	Severe	Very severe
Gestational age at birth	32 (29–34)	33 (31–35)	37 (36–41)	31 (28–35)	31 (28–35)	29 (21–37)
Leading symptoms	Polyuria, hypochloremia, alkalosis, hypokalemia	Polyuria, hypochloremia, alkalosis, transient neonatal hyperkalemia	Hypokalemia, hypochloremia, alkalosis, failure to thrive	Polyuria, hypochloremia, alkalosis, hypokalemia	Polyuria, hypochloremia, alkalosis, hypokalemia	Polyuria, hypochloremia, alkalosis, hypokalemia
Calcium excretion	High	High	Variable	Variable	Variable	High
Nephrocalcinosis	Very frequent	Very frequent	Rare, mild	Rare, mild	Rare, mild	Rare, mild
Plasma Cl/Na ratio	Normal	Normal	Decreased	Decreased	Decreased	Increased
Other findings			Mild hypomagnesemia	Deafness, risk for CKD, ESRD	Deafness, risk for CKD, ESRD	Large for gestational age, transient disease

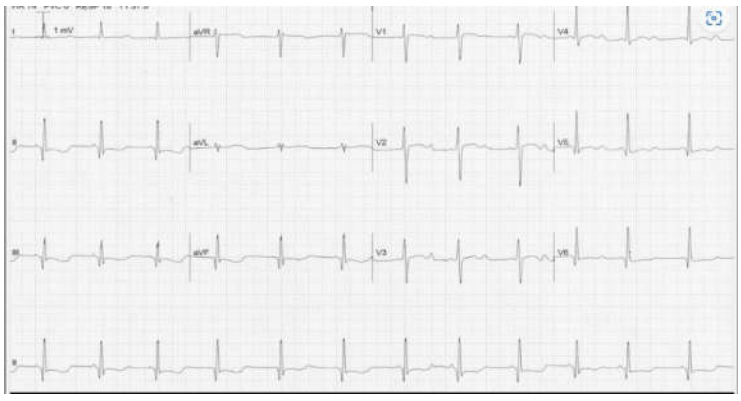

Figure 1: ECG of the child showing absent t waves

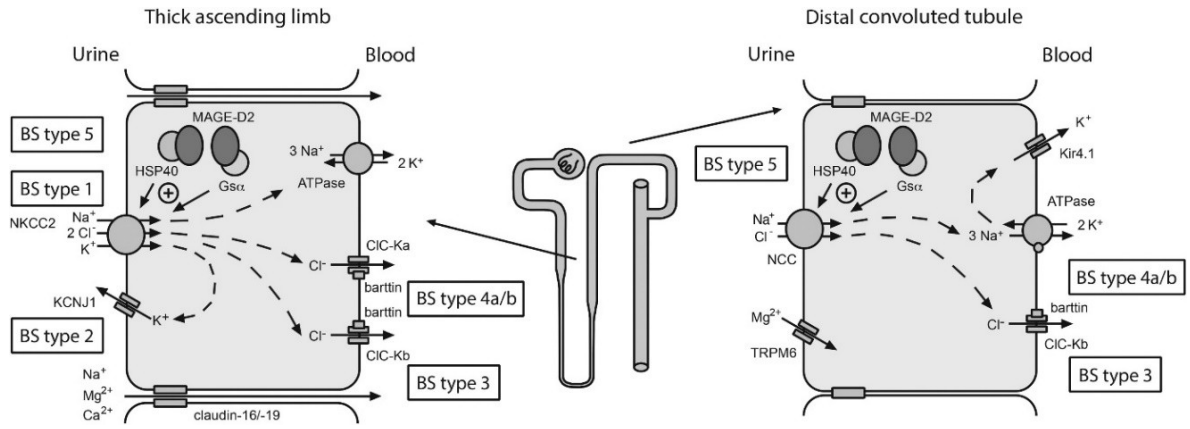

Figure 2: Pathophysiology of Bartter syndrome.

Schematic model of salt transport in the thick ascending limb and the distal convoluted tubule with associated defects in Bartter syndrome (BS) indicated. In the thick ascending limb, NaCl is reabsorbed by the NaK2Cl cotransporter NKCC2, which is mutated in BS type 1. Here, the potassium ion is recycled into the tubular lumen via the apical potassium channel KCNJ1 (ROMK), which is mutated in BS type 2. In the distal convoluted tubule, NaCl enters the tubular epithelium via the NaCl cotransporter NCC. In both tubular segments, chloride leaves the cell on the basolateral side through chloride-permeable ion channels ClC-Ka and ClC-Kb. A molecular defect of ClC-Kb causes BS type 3. Mutations in either the accessory subunit barttin or a combined defect of both chloride channels ClC-Ka and ClC-Kb result in BS types 4a and 4b. Finally, transient BS type 5 is caused by mutations of MAGE-D2. MAGE-D2 stimulates trafficking by protecting NKCC2 and NCC from intracellular degradation via HSP40 and promotes apical targeting of NKCC2 and NCC via Gs-alpha [4].

DISCUSSION

Bartter syndrome (BS) is a rare autosomal recessive renal tubular disorder that should be considered in the differential diagnosis of failure to thrive (FTT), especially in children presenting with polyuria, electrolyte abnormalities, and normal blood pressure. First described by Bartter et al. in 1962 [1], BS is due to defective salt reabsorption in the thick ascending limb of the loop of Henle due to mutations in specific ion transport genes [2].The pathophysiology of Bartter syndrome is as explained in Figure 2. The classic biochemical profile includes a normotensive child, hypokalemia, hypochloremia, metabolic alkalosis, and increased plasma renin and aldosterone levels.

Neonatal BS (types 1 and 2) presents with polyhydramnios, premature birth, severe polyuria, and nephrocalcinosis. Type 3 (classic BS) usually manifests in childhood with fatigue, polydipsia, growth failure, and salt craving, mostly without nephrocalcinosis. Type 4 includes sensorineural deafness, while type 5 is transient and X-linked [3, 4](table 1). Gitelman syndrome, a differential diagnosis, shares overlapping features but is distinguished by hypomagnesemia and hypocalciuria [5].

Nephrogenic diabetes insipidus may coexist with BS and worsen volume loss and electrolyte derangements [6]. Diagnosis depends on clinical features, biochemical testing, renal ultrasound, and, preferably, genetic confirmation. Treatment includes potassium supplementation and NSAIDs like indomethacin (1-5mg/kg/day) to reduce prostaglandin-mediated salt wasting and potassium-sparing diuretics for temporary relief for hypokalemia as used in this case. This case highlights the need to consider Bartter Syndrome as a potential cause in the evaluation of children with failure to thrive with a history of consanguinity and systemic involvement.

Follow-up in Bartter Syndrome includes clinical assessment for hydration, polyuria, growth, and muscle strength; biochemical monitoring of electrolytes, renal function, and parathyroid levels; urinary osmolality to track nephrogenic diabetes insipidus; imaging every 12-24 months for nephrocalcinosis and renal stones; and regular growth and hormonal evaluations to guide therapeutic adjustments and long-term management.

CONCLUSION

This case points out the need for a systematic evaluation of failure to thrive, emphasizing that all FTT should not be immediately attributed to nutritional deficiencies. Systemic and metabolic disorders such as Bartter Syndrome and nephrogenic diabetes insipidus must be considered, particularly in the presence of electrolyte disturbances and polyuria. Early diagnosis and targeted management can significantly improve growth outcomes and overall prognosis in these patients.

Consent

Written informed consent was obtained from the patient's legal guardian for publication of this case report.

Conflicts of Interest

The authors declare no conflicts of interest.

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