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CASE REPORT

Severe Metabolic Alkalosis in an Infant: Bartter Syndrome

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ABSTRACT

Metabolic alkalosis is an uncommon acid/base disorder in children in which serum bicarbonate concentration is increased. Two most important causes of metabolic alkalosis are emesis and diuretic use. However, in the absence of these two etiologies, a thorough investigative workup is of paramount importance to reach a definitive diagnosis. A case having severe metabolic alkalosis diagnosed as a case of Bartter syndrome is being reported.

Keywords: Bartter syndrome, Hypokalemia, Metabolic alkalosis.

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INTRODUCTION

Metabolic alkalosis is an uncommon acid/base disorder that occurs in critically ill children. Without treatment, severe metabolic alkalosis may result in significant adverse consequences, including impaired perfusion, diminished respiratory drive, cardiac arrhythmias, seizures, and death. Identifying the underlying pathophysiology is essential to the management of this disorder.[1] Metabolic alkalosis occurs when a primary pathophysiologic process leads to the net accumulation of base within or the net loss of acid from the extracellular fluid; typically, the intracellular compartment becomes more acidic in potassium-depletion alkalosis. Unopposed by other primary acid/base disorders, metabolic alkalosis is recognized by increases in both arterial blood pH – alkalemia – and plasma bicarbonate concentration.[2]

A case in pediatric intensive care unit, with severe metabolic alkalosis, was eventually diagnosed as a case of Bartter syndrome (BS).

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CASE REPORT

A four-and-a-half-month-old male child was brought to the Pediatrics outpatient department with complaints of refusal to feed since 3 days and labored breathing and vomiting since 1 day. The child vomited twice after feed which was nonbilious, nonblood stained, and contained milk. Simultaneously, he started having a little labored breathing, which was persistent and associated with restlessness.

Child had attained developmental milestones appropriate for the age. On examination, child was afebrile, dehydrated, and tachypneic. Oxygen saturation on nonrebreathing mask was normal. Pulses were well felt. Urine output monitoring was done, which showed polyuria. On head to toe examination, the child had triangular face with low set and protruding ears. On anthropometry, his weight, height, and head circumference were below –3 standard deviation. Systemic examination was normal. Patient was started on maintenance intravenous fluids, along with symptomatic treatment, and lab investigations revealed hyponatremia (119 mEq/L), hypokalemia (2.5 mEq/L), and hypochloremia (53 mEq/L).

In view of tachypnea, arterial blood gas was ordered. It showed: pH 7.64, pCO_2 47, pO_2 70, HCO_3 50.6, tCO_2 52. Double potassium correction was started in view of hypokalemia. Common causes of metabolic alkalosis like excessive vomiting, use of bicarbonate (baking soda), and use of diuretics were ruled out. Urinary electrolyte levels were sent, which were as follows: Urinary Na – 16, K – 37.3, Cl – 20.

In view of high chloride levels in urine in the background of metabolic alkalosis (chloride-resistant metabolic alkalosis), blood pressure monitoring was done. It was within normal limits. Ultrasonography (abdomen and pelvis) was normal. Renal Doppler did not show any evidence of renal artery stenosis. So causes like adrenal adenoma, renovascular disease, renin-secreting tumor, Cushing syndrome, 17β-hydroxylase deficiency, 11β-hydroxylase deficiency, and Liddle syndrome (all associated with high blood pressure) were ruled out. In the setting of normal blood pressure, metabolic alkalosis, hypokalemia, high urinary chloride levels, hypochloremia, and young age of the patient along with the presence of dysmorphic features mentioned above, diagnosis of BS was established. Child was started on oral potassium.

With continued treatment, child became better and started accepting feeds well. Serum electrolytes normalized soon.

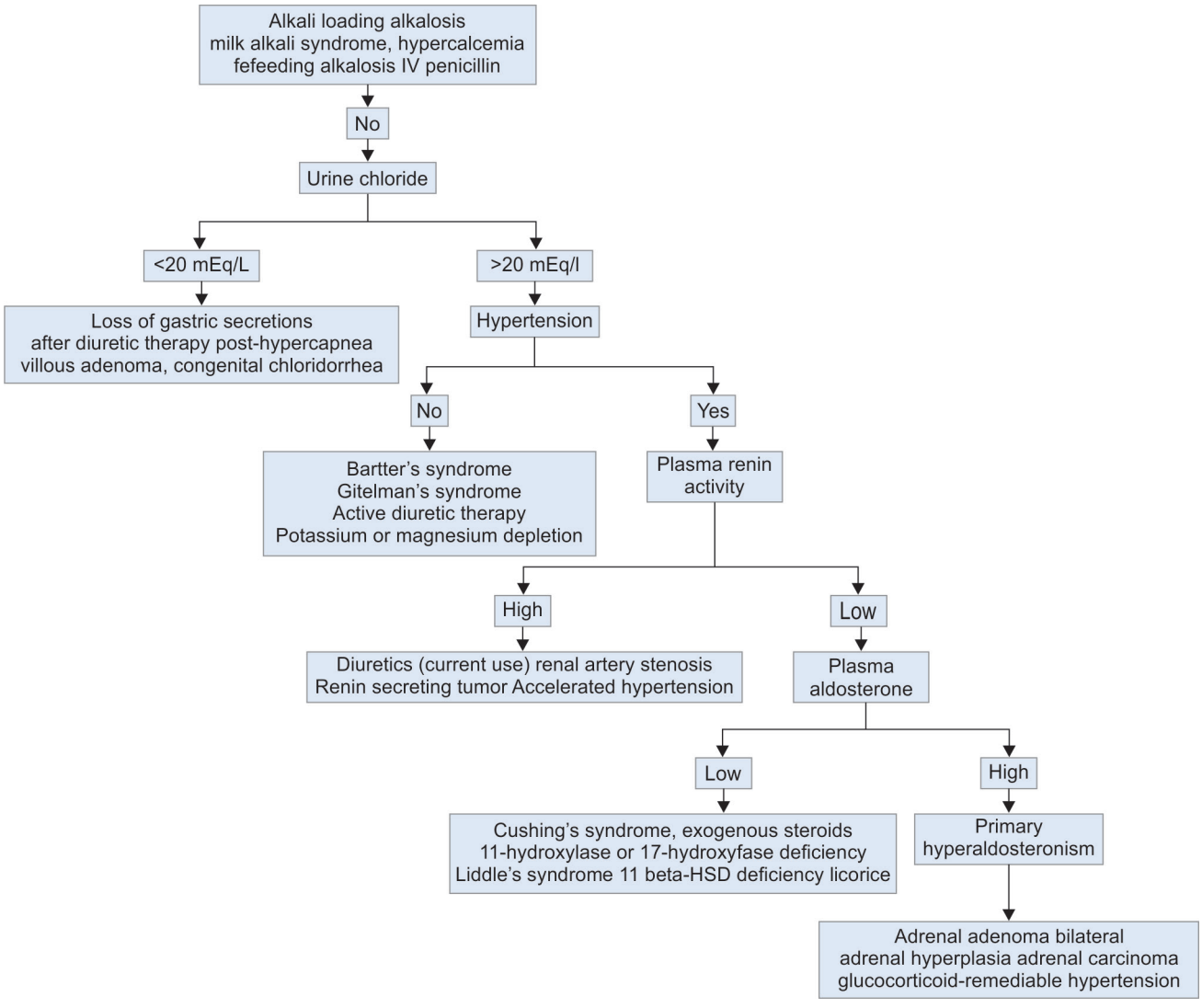
DISCUSSION

Metabolic alkalosis is classified as chloride responsive or chloride resistant. Etiologies of chloride-responsive metabolic alkalosis are chloride-depleting diuretic therapy (e.g., furosemide and chlorothiazide) and gastrointestinal loss (e.g., vomiting and nasogastric suctioning). Chloride-resistant metabolic alkalosis commonly occurs secondary to excessive mineralocorticoid activity or severe hypokalemia.[1] Alkalosis in BS and Gitelman syndrome (GS) and their variants are associated with both potassium and chloride depletion.[2]

In an approach to look for cause of metabolic alkalosis in a child, common causes like emesis, diuretic use, or excess base administration should be asked for. This is followed by estimation of urine chloride levels. If urine chloride levels are less than 20 mEq/L, the cause can be emesis, repeated nasogastric suctioning, diuretic use, chloride losing diarrhea, chloride-deficient formula, cystic fibrosis, or posthypercapnia state. If urine chloride level is more than 20 mEq/L, it is suggestive of chloride-resistant metabolic alkalosis. Measurement of blood pressure at this point is crucial in clinching the diagnosis. In the presence of hypertension, the cause can be adrenal adenoma, renovascular disease, renin-secreting tumor, Cushing syndrome, Liddle syndrome, licorice ingestion, 17β-hydroxylase deficiency, or 11β-hydroxylase deficiency. If blood pressure is normal, the cause can be GS, BS, autosomal dominant hypoparathyroidism, epilepsy, ataxia, sensorineural hearing loss, tubulopathy syndrome, or base administration.[3] Algorithm for approach to metabolic alkalosis is depicted in Flow Chart 1.[4]

In 1962, Bartter et al[5] described a new disease entity in two African Americans who presented with metabolic alkalosis, hyperplasia of juxtaglomerular apparatus, and normotensive hyperaldosteronism. Over the years, several phenotypic and genotypic variants of the original descriptions of BS have been identified. It is an uncommon

Flow Chart 1: Algorithm for metabolic alkalosis



inherited renal tubular disorder with hyponatremia, hypokalemia, hypochloremic metabolic alkalosis, hyperreninemia with normal blood pressure associated with increased urinary loss of sodium, potassium, calcium, and chloride. The primary defect in BS is an impairment in one of the transporters involved in sodium chloride reabsorption in the thick ascending limb of loop of Henle, viz., Na-K-2Cl cotransporter (NKCC2) or apical K channel (ROMK) or basolateral chloride channel (ClCNKB). Bartter syndrome is transmitted as an autosomal recessive disorder. The estimated prevalence is approximately 1 per million in the Western population.[6]

The current classification includes type I and II (neonatal or antenatal BS) due to defective NKCC2 and ROMK genes respectively, affecting the Na–K–2Cl symporter predominantly. Type III, or "classic" BS, is due to CLCNKB genetic defect causing abnormal basal chloride channel. Type IV is rare and the most severe combined loop and distal tubule dysfunction with associated sensorineural deafness and is due to CLCNKA impairment or their beta subunit BSND genetic mutation.[7]

Dysmorphic features include triangular facies, protruding ears, and large eyes. Strabismus and drooping mouth may be present on examination. Older children can have history of recurrent episodes of polyurea with dehydration, failure to thrive, nonspecific fatigue, dizziness, and chronic constipation. Blood pressure is usually normal. Renal function is typically normal. Urinary calcium levels are typically elevated, as are urinary potassium and sodium levels. Establishment of hypokalemia and hypochloremia with metabolic alkalosis is vital for diagnosis. Although renal biopsy is not essential for diagnostic purposes, the cardinal feature will be hyperplasia of juxtaglomerular apparatus in most specimens. A definitive diagnosis can be reached by genetic mutation analysis.[7] The treatment consists of proper hydration, potassium supplementation, and indomethacin therapy, which will blunt the prostaglandin overproduction and correction of hypokalemia. Potassium-sparing diuretic spironolactone may benefit transiently. Nephrocalcinosis, chronic renal failure, and short stature are a few known complications and should be kept in mind and, if encountered, addressed accordingly.[7]

CONCLUSION

Bartter syndrome should be suspected in any child with history of failure to thrive and metabolic alkalosis. Early diagnosis and treatment with potassium supplementation are lifesaving.

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