Gitelman syndrome in an infant

Sir,

A 5-month-old male infant, born of nonconsanguineous parents, was evaluated for failure to thrive and polyuria. The baby had been treated elsewhere with oral and intravenous fluids for several dehydration like episodes. He was exclusively breast fed and had no problem in sucking. Mother had a history of polyhydromnios. The vitals were stable with a blood pressure (BP) of 76/40 mmHg. Blood count was within normal limit. Serum sodium, potassium, chloride and ionized calcium were 120 meq/L, 2.1 meq/L, 70 meq/L and 0.94 mmol/L respectively. Serum creatinine was 0.6 mg/dl. Arterial blood gas analysis revealed hypokalemic metabolic alkalosis (pH 7.79, pCO_2 28 mm of Hg, pO_2 133 mm of Hg, bicarbonate excess of 21.8 meq/L). Urine report showed albumin ++, pus cells 10–15/hpf and red blood cell 1–2/hpf. Urine culture revealed the growth of *Staphylococcus aureus*. Ultrasonography of kidneys was normal. The urinary tract infection was treated with appropriate antibiotics and the cause for hypokalemic metabolic alkalosis was sought for. Urinary chloride level was 46 meq/L that ruled out the possibility of cystic fibrosis, diuretic overuse and excess vomiting, wherein this value is <25 meq/L. So, further, the possibilities of tubulopathies like Bartter syndrome (BS) or Gitelman syndrome (GS), primary mineralocorticoid excess, and exogenous alkali overload were considered wherein the urinary chloride is >25 meq/L. Normal BP and serum aldosterone level of 14.58 pg/ml (N-20–1100 pg/ml) ruled out primary mineralocorticoid excess. So now to differentiate between BS or GS, low urinary calcium: creatinine ratio (0.127) and hypomagnesemia (1.6 mg/dl) favored GS. Genetic testing could not be done due to lack of facilities.

With a diagnosis of GS, he was given oral potassium and magnesium supplementation. He responded to this with a good weight gain, improvement in polyuria and correction of hypokalemic metabolic alkalosis (pH 7.44, bicarbonate excess 0.2 meq/L, serum potassium 3.6 meq/L and serum magnesium of 2.2 mg/dl).

BS and GS are inherited renal tubulopathies that share biochemical features of hypokalemia and metabolic alkalosis but, differ in modes and ages of the presentation. The typical features of BS include characteristic triangular facies, protruding ears, large eyes and a history of polyhydromnios. The babies are symptomatic since early infancy with repeated episodes of dehydration and failure to thrive. GS, on the other hand, presents at a later age with features of recurrent muscle cramps and spasms. Differentiation is made on the basis of calcium excretion and serum magnesium levels. Urinary calcium: creatinine ratio <0.20 and hypomagnesaemia suggest GS. Also, serum renin, aldosterone and prostaglandin E levels are elevated in BS but not in GS.[1,2] The clinical features of the early age of presentation, polyhydromnios and recurrent episodes of dehydration were more in favor of BS in the index case. But, hypocalciuria and hypomagnesaemia and normal aldosterone level favored GS. The baby responded to oral potassium and magnesium supplementation. On searching the literature, we could find only one case report of GS in infancy[3] and a few in childhood.[4,5]

We report this case to sensitize our fellow clinicians to the possibility of GS even in infancy. It is important to differentiate it from BS by appropriate investigations as the management and prognosis of these two conditions differ.

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