Gitelman syndrome presenting as Hypokalaemic Periodic Paralysis

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Abstract

Several clinical syndromes may present as Hypokalaemic Periodic Paralysis (HPP). We present a young female who developed HPP due to Gitelman syndrome.

CASE REPORT

A 31-year old female patient was admitted to hospital as an medical emergency with sudden onset weakness of all four limbs. She had three similar episodes in the past. Each of these previous episodes of quadriparesis was of sudden onset ascending lower motor neuron symptoms without cranial nerve, sensory, bladder-bowel or respiratory muscle involvement. Each time there was documented hypokalaemia (2.0, 2.2 and 2.4 mmol/L respectively). During each episode, she was treated with potassium supplementation (including intravenous potassium chloride), spironolactone and acetazolamide and recovered within two days.

The patient was asymptomatic for three years but developed a similar attack of flaccid quadriparesis when the family physician asked her to stop potassium supplementation. Again there was symmetrical, flaccid quadriparesis with no cranial nerve, sensory or sphincter involvement. Blood pressure was 130/80 mm Hg, respiratory rate was 44/min and unlike in the previous episodes, the patient developed impending ventilatory failure with a single breath count of 10. She was immediately transferred to the Intensive Therapy Unit. Intravenous potassium chloride was started in the dose of 40 mmol/l in 3 liters of fluid/ day and continued for two days, followed by oral potassium chloride 80 mmol /day. Ventilatory support was not needed. Over the next two days, the paresis improved and single breath count improved to 32.

The ABG showed pH of 7.475, 7.36 to 7.44, PCO_2 of 37.5 mm Hg, PO_2 of 83.4 mm HG, HCO_3 of 27.7 mm Hg, SaO_2 of 94.2%, Hb-13 gm% and PCV-39%. There was no history of intake of drugs causing hypokalaemia, i.e. diuretics, insulin, $_2$-agonist or -antagonist, vitamin B12, folic acid, penicillin, aminoglycosides or amphoterecin-B. Thyroid function test was- T_3-120 ng/ml, T_4-8.2 g/dl, TSH-2.2 U/ml. 8 A.M cortisol after 1 mg overnight dexamethasone was 0.85 g/dl. Tensilon test was negative. Serum Na^+ was 141 mmol/l, K^+ was 4.2 mmol/L (after correction), calcium was 9.0 mg/dl and magnesium was 1.6 meq/L (N= 1.8-3 mg/dl). 24 hours urinary Na^+ was 198 meq(NR=100-260 meq), K^+ was 73.6 meq (N<15 meq/l), calcium was 96 mg (NR= 100-400 mg) and magnesium-50 mg was (N<31 mg). CSF and NCV studies were planned to exclude AIDP but were not needed as the patient improved rapidly. Kaluresis, hypocalciuria, hypermagnesuria and hypomagnesaemia suggested a diagnosis of Gitelman syndrome. She was discharged within four days with the advice to continue acetazolamide, spironolactone and potassium chloride long term.

DISCUSSION

Gitelman syndrome and Bartter syndrome are two rare autosomal recessive conditions with distinct chloride channelopathies and have three cardinal features- hypokalaemia, metabolic alkalosis and normal to low B.P. While the latter is characterized by hypercalcuria with normal serum and urinary magnesium levels, the former is associated with hypocalcuria, hypermagnesuria and hypomagnesaemia, as in our case.

The usual mode of presentation of Gitelman syndrome is with weakness, fatigue, muscle cramps and nocturia in adolescents and young adults. Gitleman syndrome can

present with carpopedal symptoms and there are reports of patients developing ventricular tachycardia induced by hypokalemia and hypomagnesemia

Gitelman syndrome, is due to loss-of-function mutation of the thiazide-sensitive Na-Cl co-transporter. Reduced Na^+ reabsorption results in volume depletion, eventually leading to kaluresis and hypokalaemia. Loss of activity of thiazide-sensitive Na-Cl co-transporter causes increased calcium reabsorption and produces hypocalciuria. Patients with Bartter syndrome and Gitelman syndrome have reduced vascular reactivity, normo-hypotension and decreased peripheral resistances in spite of biochemical abnormalities typical of hypertension.

Our patient had the typical biochemical features of Gitleman syndrome. [1,2] However our patients presentation was most unusual.

LEARNING POINTS

Gitelman syndrome, a rare chloride-channelopathy associated with hypokalaemia, hypomagnesaemia, hypermagnesuria and hypocalciuria, may present with periodic paralysis.

Ventilatory failure may necessitate intensive care.

Hypokalaemia may be present even in the asymptomatic period, unlike in the classical hypokalaemic periodic paralysis, where patients are mostly normokalaemic during the intervening, asymptomatic periods.

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