Case Report: A Rare Case of Refractory Hypokalemia - Gitelman Syndrome

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***Abstract*—Gitelman syndrome (GS) is a rare autosomal recessive renal tubular disorder characterized by hypokalemia, hypomagnesemia, metabolic alkalosis, and hypocalciuria. It results from mutations in the SLC12A3 gene, which encodes the thiazide-sensitive sodium-chloride cotransporter (NCC) in the distal convoluted tubule. Here, we report a case of a young adult presenting with recurrent muscle cramps and fatigue, ultimately diagnosed with Gitelman syndrome.**

I. INTRODUCTION

Gitelman syndrome is a salt-wasting tubulopathy that typically presents in late childhood or early adulthood with nonspecific symptoms such as fatigue, muscle weakness, tetany, and cramps. It is often diagnosed incidentally during routine electrolyte testing. Unlike Bartter syndrome, which affects the thick ascending limb of the loop of Henle, GS involves dysfunction of the distal convoluted tubule, leading to characteristic biochemical abnormalities.

II. CASE PRESENTATION

A 26-year-old female presented with complaints of recurrent muscle cramps, fatigue, and intermittent tingling sensations in the hands and feet over the past two years. She reported occasional dizziness but denied significant polyuria, polydipsia, or hypotension. There was no history of nephrolithiasis or diuretic use. Her past medical history was unremarkable, and there was no family history of renal disorders.

On physical examination, she appeared well-hydrated, with a blood pressure of 110/70 mmHg and no postural drop. No signs of edema or neuromuscular abnormalities were observed. Laboratory investigations revealed persistent hypokalaemia (2.8 mmol/L), hypomagnesemia (1.2 mg/dL), metabolic alkalosis (serum bicarbonate: 30 mmol/L), and hypocalciuria (urinary calcium-to-creatinine ratio: 0.02). Her plasma renin activity was elevated, while aldosterone levels were within normal limits. A 24-hour urine analysis showed increased potassium and sodium excretion, consistent with renal potassium wasting.

Given these findings, a clinical diagnosis of Gitelman syndrome was made, which was later confirmed by genetic testing showing a homozygous SLC12A3 gene mutation. The patient was managed with oral potassium and magnesium supplements, along with dietary modifications. She was advised to maintain adequate hydration and consume a diet rich in potassium and magnesium. During follow-up at six months, she showed symptomatic improvement with stable serum potassium and magnesium levels.

III. DISCUSSION

Gitelman syndrome results from a loss-of-function mutation in the SLC12A3 gene, leading to dysfunction of the sodium-chloride cotransporter (NCC) in the distal convoluted tubule. The defect results in increased sodium, potassium, and magnesium loss in the urine, leading to electrolyte imbalances. Patients often present with fatigue, muscle cramps, tetany, and dizziness due to persistent hypokalemia and hypomagnesemia.

A key distinguishing feature of Gitelman syndrome versus Bartter syndrome is hypocalciuria, as opposed to the hypercalciuria seen in Bartter syndrome. Other differential diagnoses include chronic diuretic use, surreptitious vomiting, and other renal tubular disorders.

IV. MANAGEMENT

1.) Electrolyte supplementation: Lifelong oral potassium and magnesium replacement is often required.
2.) Dietary advice: High-potassium, high-magnesium diet.
3.) Monitoring: Regular electrolyte and renal function monitoring to prevent complications such as arrhythmias or severe muscle weakness.
4.) Spironolactone or Amiloride (in severe cases): These potassium-sparing diuretics may be beneficial in patients with persistent hypokalemia.

V. CONCLUSION

Gitelman syndrome is a rare inherited renal tubulopathy that leads to significant electrolyte imbalances, requiring lifelong management. While it has a benign prognosis, proper electrolyte correction and dietary modifications are necessary to prevent complications. Increased awareness among clinicians is essential to ensure early diagnosis and appropriate management.

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