ORIGINAL RESEARCH PAPER

Internal Medicine

UNRAVELING GITELMAN SYNDROME - CASE REPORT- ELECTROLYTE IMBALANCE MIMICS CARDIAC DISEASE

KEY WORDS: Hypokalemia, Gitelman Syndrome

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ABSTRACT

Gitelman syndrome is a rare autosomal recessive renal tubular disorder caused by mutations in the SLC12A3 gene encoding the thiazide-sensitive sodium-chloride cotransporter in the distal convoluted tubule. It is characterized by hypokalemia, hypomagnesemia, metabolic alkalosis, and hypocalciuria. The clinical spectrum ranges from asymptomatic biochemical abnormalities to severe neuromuscular and cardiac manifestations. We report the case of a 21-year-old male presenting with recurrent episodes of palpitations, generalized weakness, and tachy-brady syndrome associated with persistent severe hypokalemia. Despite multiple prior admissions for electrolyte disturbances, the underlying etiology remained undiagnosed. Detailed biochemical evaluation revealed persistent hypokalemia, hypomagnesemia, metabolic alkalosis, and low urinary calcium excretion. Genetic testing confirmed a homozygous mutation in the SLC12A3 gene, establishing the diagnosis of Gitelman syndrome. The patient improved with aggressive potassium and magnesium replacement along with potassium-sparing therapy. This case highlights the importance of considering inherited renal tubular disorders in young patients presenting with refractory hypokalemia and unexplained cardiac arrhythmias.

INTRODUCTION

Gitelman syndrome is an inherited salt-losing tubulopathy transmitted in an autosomal recessive manner. It results from mutations in the SLC12A3 gene located on chromosome 16, which encodes the thiazide-sensitive sodium-chloride cotransporter in the distal convoluted tubule. Dysfunction of this transporter leads to impaired sodium and chloride reabsorption, causing secondary hyperaldosteronism and excessive renal potassium and magnesium wasting.

The syndrome is classically characterized by:

- Hypokalemia.
- Hypomagnesemia
- Metabolic alkalosis.
- Hypocalciuria

Patients commonly present during adolescence or early adulthood with fatigue, muscle weakness, cramps, salt craving, paresthesias, or tetany. Cardiac manifestations such as arrhythmias may occur secondary to severe electrolyte imbalance and can occasionally mimic primary cardiac disease. Because symptoms are often nonspecific, diagnosis may be delayed for years.

Early recognition is important as timely electrolyte correction and long-term monitoring significantly reduce morbidity and improve quality of life.

Case Report

A 21-year-old male presented to the department of medicine with complaints of recurrent palpitations and generalized weakness for the past six months. He had multiple previous hospital admissions for severe hypokalemia but had not undergone evaluation for the underlying etiology. There was no history suggestive of vomiting, diarrhea, diuretic abuse, hypertension, or endocrine disorder.

On Examination

On examination, the patient was conscious and oriented. Vital examination revealed fluctuating pulse rates with episodes of tachycardia alternating with sinus bradycardia. Blood pressure was within normal limits.

Cardiovascular examination showed irregular heart rhythm during symptomatic episodes.

Investigations

Laboratory Investigations

The patient showed persistent electrolyte abnormalities:

- Serum potassium: 1.4–2.5 mmol/L
- Serum magnesium: 1.6 mg/dL
- Arterial blood gas analysis was suggestive of metabolic alkalosis.
- Urinary Electrolyte Analysis
- Urinary investigations revealed:
- Low urinary calcium excretion (hypocalciuria)
- Genetic Analysis
- Genetic testing identified a homozygous mutation in the SLC12A3 gene, confirming the diagnosis of Gitelman syndrome.

MANAGEMENT

The patient was managed with:
Intravenous and oral potassium supplementation
Magnesium replacement therapy
Spironolactone therapy.

The patient reported marked improvement in fatigue, palpitations, and muscular weakness. Serum potassium and magnesium levels improved significantly with treatment.

The patient was discharged on long-term oral electrolyte supplementation and advised regular follow-up with periodic cardiac and biochemical monitoring.

DISCUSSION

Gitelman syndrome is often under-recognized because of its variable and nonspecific clinical presentation. Although many patients present with mild symptoms, severe hypokalemia and hypomagnesemia can predispose to potentially life-threatening cardiac arrhythmias.

Persistent hypokalemia can alter cardiac membrane excitability and conduction, leading to arrhythmias such as premature ventricular contractions, prolonged QT interval, ventricular tachycardia, and sinus node dysfunction.

Differential diagnoses considered in patients with chronic hypokalemia include:

- Bartter syndrome
- Diuretic abuse
- Hyperaldosteronism
- Gastrointestinal potassium losses
- Renal tubular disorders

The presence of hypomagnesemia and hypocalciuria strongly favored Gitelman syndrome over other causes. Definitive diagnosis was established through identification of the SLC12A3 mutation.

Management primarily involves lifelong electrolyte replacement with potassium and magnesium supplementation. Potassium-sparing agents such as spironolactone or amiloride may help reduce renal potassium wasting. Regular monitoring is essential to prevent recurrent arrhythmias and long-term complications.

This case emphasizes the importance of maintaining a high index of suspicion for inherited renal tubular disorders in young individuals presenting with refractory hypokalemia and unexplained cardiac manifestations. Early diagnosis and appropriate management can prevent recurrent hospitalizations and improve patient outcomes.

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