

Case Report: A Hidden Cause for Electrolyte Derangement in the ED: Gitelman Syndrome

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Abstract

We present the case of a 32-year-old patient with a known history of Gitelman syndrome who presented with hypokalemia and hypomagnesemia. The patient was treated with supplemental potassium, magnesium and intravenous fluids in the ED. The patient required hospital admission for further electrolyte correction. An awareness of Gitelman syndrome is important for the emergency physician. Some patients have a known history of Gitelman Syndrome. In such cases, it is imperative to be aware of what electrolyte abnormalities to expect. Some new diagnoses of GS can be made in patients presenting with hypotension, or myalgias along with hypokalemia and hypomagnesemia.

Key words: Gitelman syndrome; Gitelman syndrome in emergency medicine

Introduction

Electrolyte derangements are a common finding in the emergency department, whether incidental or the cause for presenting symptoms. Gitelman syndrome (GS) can be the cause for recurrent hypokalemia and hypomagnesemia. [1] While often diagnosed when the patient is young, a clinician should keep this on the differential when seeing repeated visits with electrolyte deficiencies and treating them. [2] Here we discuss a case of how Gitelman syndrome has presented in the ED and what to learn from it.

Case Presentation:

A 32-year-old male presented to the ED with a complaint of pain and cramping to his right hand. He had a known history of Gitelman Syndrome and was concerned about his potassium levels, despite taking prescribed potassium supplements. The patient was also prescribed amiloride to take at home. Other past medical history was positive for a stroke. He denied any surgeries. Social history was positive for tobacco, alcohol, and marijuana use. Vitals signs were blood pressure 117/71 mm Hg, heart rate of 51 beats per minute (bpm), respiratory rate of 19 breaths per minute, temperature 96.9 degrees Fahrenheit orally, and a pulse oximetry of 100% on room air. His Body Mass Index was 30.65 kg/M².

Physical examination revealed a non-toxic, well appearing male. Pertinent positives include tenderness to the right-hand dorsal metacarpals. Pertinent

negatives include no heart murmurs, no adventitious lung sounds, no decreased range of motion of the hand, no joint pain in the right hand, no skin changes over the right hand, no decreased sensation of the right hand, and no increased capillary refill time. Otherwise, exam was unremarkable.

An x-ray was performed of the right hand showing no acute fracture and normal soft tissue. Labs ordered in the ED included CBC, CMP, magnesium, phosphorus, creatinine kinase, and urinalysis. Pertinent results included CK of 286 U/L, magnesium of 1.1 mmol/L, potassium of 2.2 mmol/L, creatinine of 1.04 mg/dL, phosphorus of 1.8 mg/dL, and an unremarkable urinalysis. The patient was treated with supplemental potassium, magnesium and intravenous fluids in the ED. The patient went on to be admitted for further electrolyte correction.

Discussion:

GS, also known as familial hypokalemia-hypomagnesemia, is an autosomal recessive disorder that causes salt wasting in the distal convoluted tubule (DCT). Specifically, the mutations in the apical membrane of the DCT are SLC12A3, coding for thiazide-sensitive sodium chloride cotransporter, and TRPM6, which codes for DCT magnesium transport [1] Symptoms of these mutations do not typically occur before age six and often is diagnosed in adolescence or adulthood. The prevalence is 1:40,000, with prevalence of heterozygotes being 1% in Caucasian populations.[2] This is one of the most inherited renal tubule disorders, however Bartter syndrome is the most

important other genetic tubule disorder to consider in differential diagnosis. Early genetic testing can be useful in suspected cases in childhood, including possible screening of family members. [2]

Due to the mutation causing blockade of salt absorption, patients typically present with hypokalemia and hypomagnesemia. However, the frequently acknowledged clinical symptom is malaise. GS is often diagnosed via laboratory findings [3]. Other common clinical findings include muscle cramps, weakness, polydipsia, paresthesias, palpitations, orthostatic hypotension, and salt craving. [4]Electrolyte derangements in the blood include hypokalemia, hypomagnesemia, metabolic alkalosis, hypocalcemia. Urinary excretion of chloride and sodium are increased. Renal function and anatomy of the kidney on ultrasound are typically normal [5]. Of note, a patient with use of thiazide diuretics could present with similar findings. Treatment consists of highly encouraged sodium intake, oral potassium and magnesium supplements. If the case of GS is still causing symptoms of hypokalemia with the supplements, oral potassium-sparing diuretics or renin angiotensin system blockers may be added to the treatment plan. There is also research on potential use of NSAIDs as treatment [6]. GS patients are often managed by Nephrology for several reasons. The electrolyte balance can be difficult to achieve. GS patients blood pressure regulation can also be difficult to manage given the mechanism of the mutation [7].

Conclusion:

An awareness of Gitelman syndrome is important for the emergency physician. Some patients have a known history of Gitelman Syndrome. In such cases, it is imperative to be aware of what electrolyte abnormalities to expect. Some new diagnoses of GS can be made in patients presenting with hypotension, or myalgias along with hypokalemia and hypomagnesemia.

Conflict of Interest: There was no funding related to this case report. The authors declare that they have no conflicts of interest.

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