JMSCR Vol||05||Issue||10||Page 29030-29032||October 2017

www.jmscr.igmpublication.org
Impact Factor 5.84
Index Copernicus Value: 71.58
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v5i10.96

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

Gitelman's Syndrome Presenting with Features of Hypomagnesemia

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Abstract
Gitelman's syndrome is primary renal tubular hypokalemic metabolic alkalosis with hypocalciuria and magnesium deficiency. It commonly presents with muscular symptoms which reflect hypokalemia. We present a case of 65 year old female patient presenting with delirium which on evaluation revealed persistent hypokalemia. Hypokalemia work up showed hypomagnesemia, hypocalciuria and metabolic alkalosis. A diagnosis of Gitelman's syndrome was made. This case report aims at presenting the possibility of Gitelman's syndrome in patients presenting with features of hypomagnesemia.
Keywords: *Gitelman, hypomagnesemia.*

Introduction

Bartter and Gitelman's syndromes are the two major variants of familial hypokalemic alkalosis; Gitelman's syndrome is a much more common cause of hypokalemia than is Bartter syndrome (BS). Gitelman's syndrome is primary renal tubular hypokalemic metabolic alkalosis with hypocalciuria and magnesium deficiency, a benign disorder while Classic Bartter's syndrome is primary renal tubular hypokalemic metabolic alkalosis with normocalciuria or hypercalciuria, a severe disorder. The antenatal and classic Bartter's syndrome and Gitelman's syndrome represent distinct variants of primary renal tubular hypokalemic metabolic alkalosis and are easily distinguished on the basis of urinary calcium levels. The Gitelman's syndrome present during adolescence or adulthood, inherited as autosomal recessive traits. The dominant features are fatigue, weakness, hypocalciuria, hypomagnesemia with hypermagnesuria and normal prostaglandin production. Cases presenting primarily with features of hypomagnesemia are rare. We report here a case of Gitelman's syndrome who presented with features of hypomagnesemia.

Case Report

A 65 year old female was brought to the ER with complaints of altered sensorium for the past 4 days. Patient had no history of fever, trauma, weakness, seizure or loss of conciousness. Patient is a known diabetic for 10 years on irregular

treatment. No other comorbidities and no history of similar illness in the past. No history of parental consanguinity and no family history of similar illness.

On examination patient was delirious. Vitals: PR-90/mt, BP-130/70 mmHg, RR-18/mt, T-98.6^{o}, SpO2-96%. Systemic examination revealed no other focal neurlogical deficit.ECG revealed U waves. CT Brain showed normal brain parenchyma. A provisional diagnosis of metabolic encephalopathy was made. Initial laboratory workup showed serum sodium-136 mEq/l, serum potassium-2.6 mEq/l, urea-43 mg/dl, creatinine-1.3 mg/dl. Intravenous Potassium correction was given at 20 mEq over 4 hours. Since renal and liver parameters were normal and repeat serum potassium was 2.4 mEq/l complete lab workup for refractory hypokalemia was done

Table 1: Hypokalemia work up

serum magnesium	1.3 mg/dl (1.7-2.5)
urine potassium	18.7 mmol/day
urine chloride	60 mmol/l
urine calcium	60 (100-300) mg/day
urine osmolality	340 mOsm/kg
TTKG	4.2
urine calcium/ creatinine ratio	0.12
Blood pH	7.55(HCO_3-32/CO_2-37.3)

Thyroid profile was normal. As the invstigations revealed hypokalemia, hypomagnesemia, hypocalciuria and metabolic alkalosis a diagnosis of Gitelman's syndrome was made. Patient was started on IV magnesium correction along with potassium correction. Patient condition improved over one day and oral supplementation of potassium and magnesium was continued.serum potassium and magnesium normalized over 2 days. Patient was discharged with oral potassium and magnesium supplementation and advised a high sodium high potassium diet. Our patient has been normokalemic without any symptoms for one year and is on follow up once in 6 months.

Discussion

Bartter and Gitelman syndrome are inherited disorders of salt reabsorption, they lead to ECF volume depletion and normal to low blood pressure[2]. The clinical phenotypes can be classified according to the predominant site of deficient salt transport.

Gitelman's syndrome typically presents with hypokalemic alkalosis. The arterial pressure is typically low normal and the serum magnesium is reduced. The urinary calcium excretion is suppressed. The syndrome results from mutation of the gene (SLC12A2) that encodes the thiazide-sensitive NaCl cotransporter (NCC) in the distal convoluted tubule[5]. Gitelman's syndrome affects males and females in equal numbers. The disorder occurs in approximately 1 in 40,000 Caucasian individuals[4]. It typically presents after puberty with hypokalemic alkalosis. Most individuals present with muscular symptoms and fatigue. Hypocalciuria and hypomagnesemia are characte-ristic features of Gitelman's syndrome. Paralysis and prolonged QT interval with malignant arrhythmias have been observed and ascribed to electrolyte abnormalities. Most asymptomatic patients are untreated and progression to renal insufficiency is very rare[1] .

Our patient presented with delirium which is a manifestation of hypomagnesemia. Diagnosis of Gitelman's syndrome was made with the help of diagnostic algorithm[5] as patient had hypokalemia, hypomagnesemia, hypocalciuria and metabolic alkalosis.

The hypocalciuria in Gitelman’s syndrome is helpful in distinguishing it from classic Bartter’s syndrome[3]. The greater urinary calcium excretion in patients with classic Bartter’s syndrome is consistent with impaired reabsorption in the ascending limb of loop of Henle. Alternatively the hypocalciuria of Gitelman’s syndrome suggests the involvement of the distal convoluted tubule, where reduced chloride absorption is associated with augmented calcium absorption. Hypocalciuria in Gitelman's syndrome is not accompanied by changes in serum calcium,

phosphate, vitamin D or PTH suggesting a direct effect on renal calcium transport. Our current understanding of tubular function does not easily explain the dissociation between calcium and magnesium excretion in these disorder.

There is no cure for Gitelman's syndrome. The mainstay of treatment for affected individuals is oral potassium and magnesium supplements. Potassium sparing diuretics may be used.

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