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A Rare Case Report of Gitelman's Syndrome Present, Mimic as Acute Paraparesis due to Persistant Hypokalemia

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ABSTRACT

Gitelman syndrome (GS), also referred to as familial hypokalemia-hypomagnesemia , is an salt losing renal tubulopathy that is characterized by hypomagnesemia, hypocalciuria, and secondary aldosteronism, which is responsible for hypokalemia and metabolic alkalosis . It is beningn disorder inherited as autosomal recessive traits.

Keywords: *Hypocalciuria, Hypokalemia, Hypomagnesemia, Gitelman's syndrome.*

INTRODUCTION

1% in Caucasian populations, making it one of the most frequent inherited renal tubular disorders. This renal syndrome is caused by mutations in the solute carrier family 12, member 3, SLC12A3 gene, which encodes the renal thiazide-sensitive sodiumchloride co-transporter (NCCT) that is expressed in the cells in the distal convoluted tubule (1). Gitelman syndrome (GS), also referred to as familial hypokalemia-hypomagnesemia, is an salt losing renal tubulopathy that is characterized by hypomagnesemia, hypocalciuria, and secondary aldosteronism, which is responsible for hypokalemia and metabolic alkalosis (2). It is beningn disorder inherited as autosomal recessive traits.

- The prevalence is estimated at ~25 per million and accordingly, the prevalence of heterozygotes is approximately is expressed in the cells in the distal convoluted tubule (1).
- Gitelman syndrome present during adolescence and adulthood. Dominant features are fatigue, weakness, hypocalciurea, hypomagnesemia. We report here a patient who presented with features of gitelman syndrome.
- Symptoms of Gitelman's syndrome repoted in literature range from asymptomatic to mild symptoms of cramps and fatigue to severe manifestation such as tetany, paralysis and rhabdomyolysis (3).

CASE REPORT

- 18 year male patient was admitted in our hospital with complaints of recurrent weakness in lower limbs since 2weeks .he had similler illness 9 month back of weakness in lower limb n taken medication n relived for some days. On examination having pseudohypertrophy of calf muscle on both side.
- No history of drug taken in past like diuretics, laxatives, antibiotics…etc.
- Not having trauma/any surgery/fracture/local tenderness in lower limbs.
- No history of simmilar illness since birth.
- No history of simmilar illness in any other family members and no h/o of parental consanguinity.
- Not having any sensorineural deafness.
- History of one episode of vomitting containing food particle, nonbillous, projectile.
- No h/o diabetic melliitus
- No h/o hypo/hyper thyroids'
- Bowel /bladder /sleep /appetite –normal. Afebrile, conscious, oriented to time, place and person.
- No -pallor/icterus /cyanosis/clubbing /lympha denopathy
- Blood pressere-100/60 mm hg, pulse rate-80/min

- There is not any neurological deficiency .thyroid function test (T3,T4,TSH)were normal. Ultrasound of abdomen did not reveal any abnormality.
- No sensorineural deafness on ENT examination
- Complete blood count/renal function test/liver function test/CSF fluid examination/lipid profile/randome blood sugar –all within normal limit.
- Investigation shows:serum sodium-134.47(135-145),serum pottasium-2.66 (normal range 3.5-4.5), serum magnesi um-1.76 mg/dl(1.8-2.8 mg),serum calcium-8.75 mg/dl(8.4-10.2),urine sodium-17.0 mmol /l,urine calcium-0.86mmol/l(2.5-7.5 mol/l)
- Urine pottsium-50.3(25-120),urine chloride-73.5meq/l(110-250)
- TSH-0.56uIU/ml,T3-12.9 ug/dl,T4-12.9ug/dl
- PH-7.545(7.35-7.45),bicarbonate-27.1mmol/l (22-26 mmol/l)
- Metabolic alkalosis with hypocalciurea with hypomagnesemia
- Patient was treated with oral magnesium and potassium supplement.
- At the time of discharge patient was stable and advice to continue oral potassium and magnesium supplement and continue in follow up.

DISCUSSION

- Gitelman et al. first described a familial salt-losing tubulopathy that was associated with hypokalemic metabolic alkalosis and hypomagnesemia. Bettinelli et al. (4) in 1992 found that in addition to hypomagnesemia, this syndrome was also associated with hypocalciuria. Four years later, the molecular defect in GS was identified when it was demonstrated that GS was the result of loss of function mutations in the SLC12A3 gene located on the long arm of chromosome 16 (5). Epidemiologic studies have demonstrated that there is no ethnic predilection for GS, and both sexes are equally affected.
- Rodriguez-soriano et al were the first to suggest that hypocalciurea may be useful in distinguishing the gitelmansyndome from bartters syndrome.
- Prostaglandin synthetase inhibitors are of no benefit in gittelman`s syndrome.
- Conclusion: The purpose of our study is to remind Gitelma's syndrome mimic as acute paraparesis due to persistanat hypokalemia.

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