J MEDICINE 2012; 13 : 118-119

Gitelman Syndrome

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Abstract:

Gitleman syndrome is a rare entity. It is a benign disorder that can present with many metabolic derangements. Here we report a patient presenting with recurrent episodes of muscle weakness, muscle cramps that was diagnosed as a cas of Gitlemans syndrome.

Keyword: *Gitelman's syndrome, Hypocalciuria, hypokalemia, Hypomagnesemia.*

Introduction:

Gitelman's syndrome is an autosomal recessive disorders with a characteristic set of metabolic abnormalities.[1, 2] It is a benign disorder presents as primary renal tubular hypokalemic metabolic alkalosis with hypocalciuria and magnesium deficiency.[3] In Gitelman's syndromes the patients are not hypertensive; and the plasma renin activity is increased, not suppressed by aldosterone-induced volume expansion. The estimated prevalence is approximately 1 per 40,000 for Gitelman's syndrome.[4] However; the prevalence of heterozygote may be as high as 1 percent. 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.[5] The dominant features are fatigue, weakness, hypocalciuria, hypomagnesaemia with hypermagnesuria and normal prostaglandin production.[6] We report here a patient who presented with features of Gitelman's syndrome.

Case report:

A 60 year old man, hailing from Narayangonj was suffering from recurrent attacks of weakness in both lower limbs for last 4 years. He also complained of pain in the thigh and calf muscle during the attack and became bed ridden during that time.In between each attack he remained asymptomatic. In each episode, he improved without any residual weakness. He has no h/o bowel and bladder involvement and it was not associated with diarrhea, vomiting or heavy meal. And he did not have any sensory impairment. He was never put on diuretics, steroid, laxative, insulin or beta agonist.

He took diclofenac sodium, ketorolac and tramadol. And he took potassium chloride on various occassions. His other family members were in good health. He had history of smoking for ten pack years. He came from a family with lower socio economic condition. He worked as a cleaner in a market and used to drink tube-well water. He was anxious, co-operative, his body build was below average, pulse was 80/m, regular, blood pressure was 80/60mm of Hg, temperature 99º F, there was corneal opacity in the rt. eye due to trauma. His systemic examination revealed no abnormality. His hemoglobin was 13.2 gm/ dl, ESR 50 mm in first hour, Total count was 9,000/ cmm, Differential count was Neutrophil 66%, Lymphocyte 20%, Monocyte 04%, Eosinophil 10%. His urine routine examination was normal. His potassium level was below normal on repeated occasion, on April 01, 2010- serum potassium was 2.0 mmol, seven days after on April 8,2010 it was 1.9, on April17, 2010 potassium level was 2.4, after about one month on May3,201was 2.9mmol, his sodium and chloride & bi carbonate level was within normal value, urinary potassium level was as follows, on 17- 4-2010 was 35.1mmol/day, on 26- 4-2010 was 26.1 mmol/day,(normal value 25-125 mmol/day), serum magnesium label was 1.2 mg/dl on01-04-2010 (normal value 1.9-2.5mg/dl), on 17-4-2010 it was 1.2mg/dl, serum calcium 10mg/dl, serum albumin: 39gm/l (35-50gm/l), urinary calcium: 66mg/day(normal value100-300mg/day), serum creatinine was 1.8mg/dl on 14-12-2009, 1.4mg/dl on 1-4-2010 & again 1.4mg/dl on 17-4-2010, creatinine clearance rate: 8-4-2010 - 19ml/min, 17-4-2010 - 37ml/min and 70 ml/min. Fasting blood glucose was 4.6mmol/l, Serum TSH was 3.59 MIU/L (normal). Serum FT4: 10.51pmol/l (normal) Blood gas analysis: PH: 7.44(7.37-7.44), Po_2:102mmg (80-90), Pco_2:45mmg(34-35), Hco_3:31mmol/l (23-29), Tco_2: 4mmol/l (24-30), USG of KUB: rt. renal para pelvic cyst with normal size of kidney. So, the patient's serum potassium was low on several occasions, hypocalceuria with higher serum calcium and lower serum magnesium level with alkalosis meets the criteria for Gitelman syndrome. The low blood pressure is also associated with Gitelman syndrome.

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Discussion:

In Gitelman's syndrome, mutations have been found in the thiazide sensitive Nacl transporter. The reduced sodium reabsorption in the distal convoluted tubule leads to volume depletion and Hypokalemia Loss of activity of the thiazide sensitive transport increases tubule calcium reabsorption, leading to the classic finding of hypocalciuria in Gitelman's syndrome.[6] In this patient, diagnosis of Gitelman's syndrome was based on clinical findings and laboratory investigation, findings like hypokalemia, hypocalciuria, hypermagnesuria and alkalosis[7]. Rodriguez-Soriano et al were the first to suggest that hypocalciuria may be useful in distinguishing the Gitelman's syndrome from classic Bartter's syndrome.[8] It is less certain whether changes in calcium excretion provide insight into the renal tubular pathophysiology of these syndromes. 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.[5] The usual mode of presentation of Gitelman syndrome is with weakness, fatigue, muscle cramps and nocturia in adolescents and young adults. Gitleman syndrome can present with carpopedal symptoms and there are reports of patients developing ventricular tachycardia induced by hypokalemia and hypomagnesemia. In this case the patient presented with only muscle weakness mimicking familial periodic hypokalemic paralysis. In familial periodic paralysis, age of onset is below 25 years and usually attack occurs following a large carbohydrate meal. It didn't fit with our patient, our patients thyroid function was normal and all biochemical parameters were consistent with Gitelman syndrome.

Conclusion:

Evaluation of Hypokalemia in Bangladeshi settings limits within the common causes. There are other rare causes which are not usually looked for. Only supplementation by potassium does not resolve the problem. Gitelman & Bartter's syndromes are rare. The case report is a breakthrough to the evaluation of hypokalemia. The literature supports the prevalence of Gitelman syndrome is one in fourty thousand. But case report in Bangladesh is minimal. This case report will encourage others to identifying Gitelman syndrome in approach to a patient with parapresis/quadriparesis.

Conflict of Interest: None

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