A Gitelman Syndrome with Hydroureteronephrosis – A Case Report

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Abstract

BACKGROUND:
A 45 years old male presented with complaints of abdominal pain and vomiting for 1 week. On evaluation, we found a rare presentation of Gitelman Syndrome with obstructive uropathy - mild HUN

INVESTIGATIONS:
Complete Hemogram with urinary and serum electrolytes revealed persistent Hypokalemia and renal loss of potassium with urinary k+ 30 mmol/L serum potassium < 2.5 mg/dl. S.calcium 7.3mg, serum mg. 1.7mg/dl and serum po4 3.3 along with urinary loss of calcium 7.5 mg/dl , urine osmolality 308mOsm. CT- KUB revealed Bilateral mild HUN and ABG analysis revealed Metabolic alkalosis with compensatory respiratory acidosis. Impression was Gitelman syndrome with hypokalemia, Hypocalciuria, hypomagnesemia and hypophosphaturia.

CONCLUSION:
Gitelman syndrome is a kidney disorder that causes an imbalance of charged atoms (ions) in the body, including ions of potassium, magnesium and calcium. It presented with a rare involvement of hydroureteronephrosis in our case and treated accordingly with electrolytes correction and relieving obstruction.

Keywords: Gitelman, Serum potassium, disorder of charged ions

INTRODUCTION

Gitelman syndrome is a kidney disorder that causes an imbalance of charged ions in the body, including ions of potassium, magnesium and calcium. Here we discuss about one rare presentation of Gitelman syndrome with hydroureteronephrosis.

CASE HISTORY AND PRESENTATION

A 45-year-old male presented with complaints of abdominal pain - 10 days insidious onset, progressing and no aggravating factors and vomiting – 1 week, not blood, bile stained and h/o polyuria seen. no H/o loss of appetite. He is k/c/o Type 2 Diabetes mellitus. He is a known alcoholic and smoker. On examination he was conscious oriented no pallor, no pedal edema. vitals were stable. CVS RS and Per abdomen, CNS examination were normal. During the course of stay he developed tetany and recovered. Motor and sensory system were normal. He was diagnosed as cystitis for evaluation. Routine investigations were taken and serum potassium was persistently low below the range of 2.5 and calcium -7.3, s.po4 -3.3, s.uric acid-5.0 and urinary Na was low and k-30 mmoL. Urine protein- 43 creatinine- 25 and urinary calcium was 7.8 mmol/L. ESR was 60 in1st hr and 90 in 2nd hour. Ultrasound and CT-KUB was shows normal kidney size with Bilateral mild Hydroureteronephrosis and bladder wall thickening seen. Echocardiography was normal. Arterial Blood gas analysis revealed metabolic alkalosis with compensatory respiratory acidosis. Finally, patient was diagnosed as GITELMAN syndrome with obstructive uropathy with transtubular kidney gradient was 5 and BP was low or normal with
urinary calcium/creatinine ratio was less than 0.10 and use of diuretics was excluded. Patient was managed with nsaid, inj.magnesium and potassium chloride infusion and inj.calcium gluconate titrated according to the serial measurements of electrolytes and bladder catherization done and serial input output monitoring and managed conservatively.
DISCUSSION

DEFINITION:

Gitelman syndrome, also referred to as familial hypokalemic hypomagnesemia is characterized by hypokalemic metabolic alkalosis in combination with significant hypomagnesemia with low urinary calcium excretion. Mutations in solute carrier family 12, member 3 gene, SLC12A3, encodes NaCl cotransporter are found in majority of GS patients.

INCIDENCE:

The prevalence is estimated as 1:40,000 and in heterozygotes is approximately 10% in Caucasian populations making it one of the most frequent inherited renal tubular disorders. In the majority of cases symptoms do not appear before age of 6 years and the disease is usually diagnosed during adolescence or adulthood.

CLINICAL FEATURES:

- Transient periods of muscle weakness and tetany sometimes accompanied by abdominal pain, vomiting and fever are often seen in GS Patients.
- Paresthesia especially in the face, frequently occur.
- Appearance of adult age of chondrocalcinosis that causes swelling, local heart and tenderness over affected joints may occur.
- Blood pressure is lower than that in general population.
- Sudden cardiac arrest has been reported occasionally.

DIAGNOSTIC CRITERIA:

1. Chronic hypokalemia (<3.5 mmol/L) with inappropriate renal potassium wasting (spot PCR > 2.0 mmol/L)
2. Metabolic alkalosis
3. Hypomagnesemia (< 0.7 mmol/L) (<1.70 mg/dl) with inappropriate renal magnesium wasting (FeMg > 4%)
4. Hypocalcemia (spot calcium-creatinine ratio < 0.2 mmol)
5. High plasma renin activity or levels and low or normal blood pressure levels
6. Fractional excretion of chloride > 0.5%
7. Normal renal ultrasound

TREATMENT:

- We recommend to encourage their propensity for salt consumption
- Lifelong oral potassium or magnesium supplementation or both is mainstay of treatment.
- A reasonable target for potassium may be 3.0 mmol/L and magnesium 0.6 mmol/L (1.46mg/dl)
- The dose will be titrated individually based on the monitoring of the values
- Oral NSAID and low dose oral colchicine are effective against acute Chondrocalcinosis
- At least, annual follow up in a nephrology clinic to monitor potential complications and evolution is advocated

CONCLUSION:

Despite a solid understanding of the underlying renal mechanisms, the wide spectrum of clinical severity, ranging from incident diagnosis in essentially asymptomatic patients to severe diastolic in others despite similar biochemical abnormalities remains an enigma. A better understanding of the factors involved in this variability is critical to provide better treatments.

REFERENCES: