



A Gitelman Syndrome with Hydroureteronephrosis –A Case Report

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Type of Publication: Case Report

Conflicts of Interest: Nil

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/L. Urine protein- 43 creatinine- 25 and urinary calcium was 7.8 mmol/L. ESR was 60 in 1st 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 catheterization done and serial input output monitoring and managed conservatively.

PIONEER SCAN CENTRE
NO.21/10, KAVERI STREET, RAM NAGAR,
AMBATTUR, CHENNAI-600 053 PH: 9176669432

PNDT R.NO.16/09

Patient name	Mr. KUMAR PANDURANGAN	Age/Sex	45 Years / Male
Patient ID	A34888	Visit No	1
Referred by	Dr. (GOV HEALTH POST)	Visit Date	05/07/2021

Abdomen and KUB Scan Report

Real time B-mode Ultrasonography of Abdomen and KUB done


Abdomen
Liver normal in size and echotexture. No diffuse or focal lesion seen in the liver. IHBR not dilated. Portal vein and Hepatic veins appeared normal.
Gall bladder distended and shows normal wall thickness. No calculi or sludge seen within the gall bladder.
Common duct appeared normal. No calculi or sludge seen in the common duct.
Pancreas appeared normal in size and echotexture. No ductal dilatation seen.
Spleen appeared normal.
Aorta appeared normal. No para aortic nodes seen.
Peritoneal cavity appeared normal. No Ascites.

KUB
Right kidney measured 12.3 X 5.6 cms.
Left kidney measured 11.4 X 6.4 cms.
Both kidney shows dilated pelvicalyceal system. Ureter dilated. Cortical echoes increased in both kidneys. no calculi seen.
Bladder grossly distended and shows irregular Wall thickening. Post void residual urine measured 310 cc.
Prostate measured 2.5 X 3.0 X 2.4 cms. (Weight = 9.36 gms.)
Prostate appeared normal. No intra vesical enlargement of prostate gland seen.

Impression

BILATERAL HYDROURETERONEPHROSIS (GRADE IV)

CHRONIC URINARY RETENTION


**DR. HEMAMALINI V. MBBS., DNB.,
SONOLOGIST**



RADIUM HOSPITAL
KUMARAN
PATIENT RESULTS

Analysis time: 21-Jul-21 9:53:45 AM
Sample type: Arterial

Patient ID: mtr.kumar
Patient name: mr.kumar

MEASURED VALUES

Blood Gas (37°C)
pH ↑ 7.49
pCO₂ ↑ 53.2 mmHg
pO₂ ↓ 82 mmHg

Hematocrit ↑ 66 %
Hct

Electrolytes/Metabolites
cNa ↓ 134 mmol/L
cK⁺ ↓ 2.37 mmol/L
cCa²⁺ ↓ 0.65 mmol/L
cCl⁻ ↓ 85 mmol/L
cLac ↑ 2.6 mmol/L

DERIVED VALUES

cHb 21.7 g/dL
cHCO₃ (P) 39.6 mmol/L
cHCO₃ (P.st) 36.6 mmol/L
cBase(B) 12.7 mmol/L
cBase(Ecl) 15.0 mmol/L
cBase(B,ex) 12.6 mmol/L
cAnionGap(K⁺) 12.7 mmol/L
cCO₂ (B) 30.4 mmol/L
cCO₂ (P) 41.3 mmol/L
cO₂ (A-a) 0.68 mmol/L

Anion 11.8 mmol/L
Gap(K⁺) 9.5 mmol/L
Anion Gap N/D mmHg
pO₂ (A) N/D mmHg
pO₂ (A-a) N/D %
pO₂ (a/A) 96.6 %
sO₂ 29.2 Vol%
tO₂ N/D %
Rf

MESSAGES

N/D not derived

PATIENT INFORMATION

Gender: Male

User: ANONYMOUS
Analyzer: s71 312066
SC lot: 317412
SC sm: 3006831
Sequence: 110471
Sample#: 9914
Software version: 3.23 (BASIC)
Date: 21-Jul-21 9:53:47 AM

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 heat 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.

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