

Gitelman Syndrome in 3 Months ANC

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Abstract: Our patient, 26 year female with 3 months ANC presented with chief complaints of bilateral upper and lower limb weakness since 8-10 days with inability to walk and get up from sitting since 8-10 days. Clinical examination reveals power is diminished in all four limbs and deep tendon reflex absent. There was no history of any drugs taken (Thiazide and Potassium supplementation)

Laboratory evaluation revealed hypokalemia, hypomagnesemia, metabolic alkalosis, Increased urinary chloride excretion, and decreased urinary calcium excretion. Renal function test was within normal limits. Renal ultrasound was normal. A diagnosis of Gitelman syndrome established after excluding other probable diagnosis.

Keywords: Hypokalemia, Metabolic alkalosis, Hypocalciuria, Hypomagnesemia, Gitelman syndrome

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I. Introduction

Gitelman syndrome (GS) is a rare salt losing tubulopathy characterized by hypokalemia, metabolic alkalosis with hypomagnesemia and hypocalciuria^{1, 2}. GS is considered as the most commonly inherited tubulopathy. The disease is autosomal recessive caused by biallelic inactivation mutation SLC12A3 gene encoding the diuretic sensitive sodium chloride cotransporter expressed in optical membrane cells lining the distal convoluted tubule. Presence of hypomagnesemia and hypocalciuria are highly predictive of the diagnosis. GS as a syndrome is considered benign and usually presents in adolescents and young adults. The disease does have the potential for severe manifestations like rhabdomyolysis, seizure, arrhythmias etc.

II. Case Report

A 26 yrs old female with 3 months ANC brought up by relatives in casualty with complaints of weakness in B/L upper and lower limb since 8-10 days. Patient had inability to walk and get up from sitting since 8-10 days. She also complains of tingling sensation in bilateral upper and lower limb in the same time period. Patient also had lower abdominal pain since 2 days and 2 episodes of vomiting per day for 4 days (8-10 days back). The weakness had started gradually and progressed to the current status where the patient is bed ridden.

Patient had no complaints of facial weakness, convulsions, loss of consciousness. She has no bowel or bladder complaints. She gives no history of fever before the onset of above mentioned complaints.

No previous history of any medical illness. No history of any drugs taken (Diuretics, Potassium supplementation). No family history of any kidney disease. Obstetric History reveals G3 P2 L2 A0 with previous two normal full term vaginal delivery.

On General Examination Patient is afebrile with pulse rate 80/min, irregular and blood pressure of 130/70 mm hg. Cardiovascular examination reveals irregular heart rate. Respiratory examination shows air entry equal on both sides. Per Abdominal examination reveals just palpable gravid uterus. On CNS Examination power is diminished in all four limbs. Proximal muscle power loss is more than distal muscle. All deep tendon reflexes are absent and B/L plantar reflexes are flexors.

Obstetric examination reveals uterus of size 12 to 14 weeks, external ballotment +, relaxed, cervix and vagina are healthy.

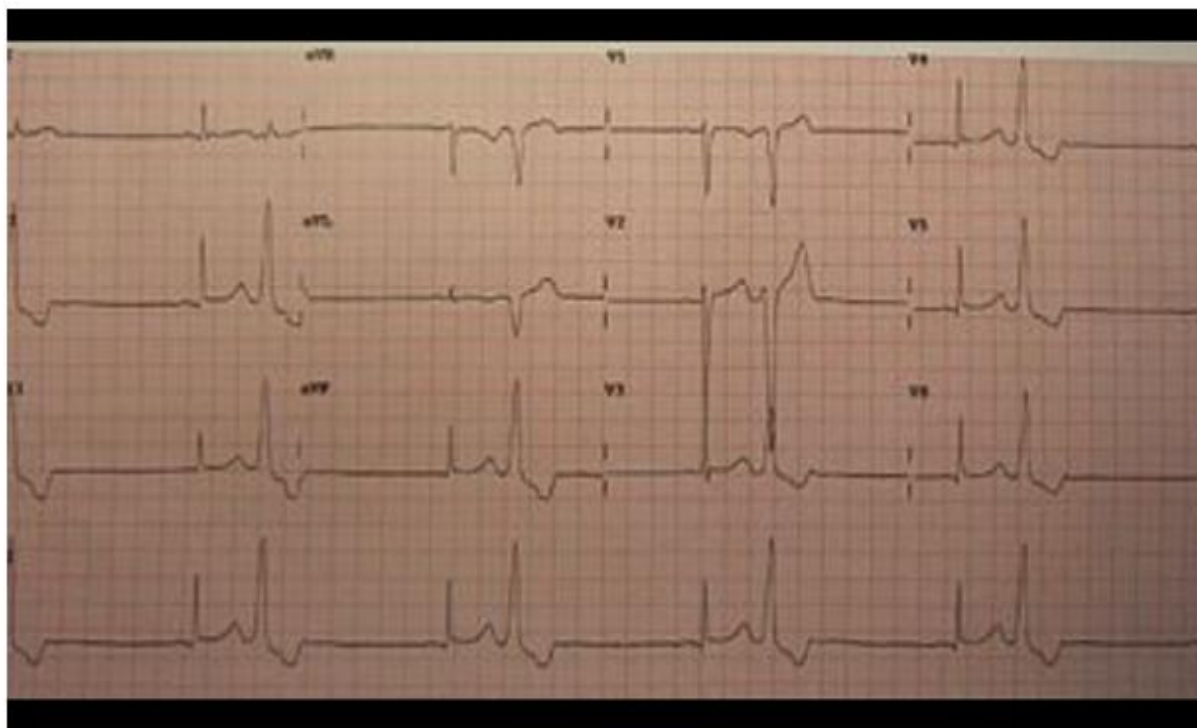
Lab Reports:-

- On ABG, she had metabolic alkalosis (pH - 7.51, HCO₃ - 36.2)
- Hypo-kalemia (1.3 mEq/L), hypo-magnesium (1.5 mg/dl)
- Increased spot urinary chloride excretion - 116.7 mEq/L
- Increased spot urinary sodium excretion - 70 mmol/L
- Decreased urinary calcium excretion - 7.9 mg/dL
- Haemogram :- Hb 10.1, TLC 10600, PLT 277000, MCV 72, HCT 31.
- RFT :- Blood Urea 25 mg/dl, Creatine 1.1mg/dl

- Serum Calcium 8.9 mg/dL
- CPK Total 41631 (Normal range 30-135)
- Thyroid Profile :- TSH 2.05, T3 - 2.7, T4 – 1.87
- Urine Myoglobin 2459mg/dL (Normal range 0-1000)

	26/5	27/5	28/5	29/5	30/5	31/5	2/6	3/6
Na	136	141	138	134	135	134	134	135
K	1.3	1.5	2.3	2.5	2.9	3.2	3.2	4.3

- Urine Examination :- Urine Albumin – Present, Urine Sugar – Nil, M/E – Occasional pus cells.
- ECG shows ventricular bigeminy with HR-94/min



Electrocardiography was suggestive of ventricular bigemini. Abdominal ultrasound revealed normal kidneys, with obstetrics USG suggesting single live intrauterine pregnancy of 14 weeks.

III. Discussion

Gitelman syndrome is characterized by Hypokalemia with metabolic alkalosis with Hypocalcemia and Hypomagnesemia. Clinical symptoms are similar to prolonged administration of thiazide diuretics. Vomiting and diuretic use are excluded by measuring high urinary chloride excretion and no history of diuretic taken before.

Other differential diagnosis of Hypokalemia are renal tubular acidosis TYPE 1 and TYPE 2 excluded as patient having metabolic alkalosis and the differential diagnosis were the genetic disorders of Gitelman and Bartter syndromes. Bartter syndrome excluded as it has earlier onset and urinary calcium excretion is often increased.

The diagnosis of Gitelman Syndrome is based on the clinical symptoms and biochemical abnormalities which include hypomagnesemia, hypokalemia, metabolic alkalosis, hypocalciuria. Treatment consist of life long potassium supplementation and symptomatic hypokalemia if not corrected, it can be associated drugs that antagonize aldosterone activity to block sodium channels ENaC in the collecting duct. An option is combination of amiloride, spironolactone or eperenone with potassium chloride.

Treatment

The patient began iv kesol supplement with Syrup Kesol 10 ml TDS with Tab.Maguion 400mg ½ BD

Outcome And Follow Up

month later, she was asymptomatic with power improved in all four limbs and deep tendon reflexes are improving **power**

With improved reflexes.

5/5	5/5
5/5	5/5

References

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