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A Case Report on Gitelman Syndrome: Rare Cause of Hypokalemia

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Abstract

Gitelman syndrome is an autosomal recessive renal tubular disorder characterized by hypokalemic metabolic alkalosis which is a common clinical problem for practicing endocrinologists and nephrologists. A 30 year old female was admitted into the hospital with chief complaints of spasm of both upper limbs and lower limbs since one day, fever and cough since 2 days. Lab investigations revealed decrease in serum electrolytes and serum creatinine. CT scan revealed chondrocalcinosis. Treatment included calcium and magnesium supplements.

Keywords: Calcium Supplements, Chondrocalcinosis, Hypokalemia, magnesium.

Introduction

Gitelman syndrome, is an autosomal recessive renal tubular disorder characterised by hypokalemic metabolic alkalosis which is a common clinical problem for practicing endocrinologists and nephrologists discovered by Gitelman, Graham and Welt, in 1966. It is milder than other sub- types of Bartter's syndrome. Patients with GS usually present during childhood or adolescence.[1]

A relative and overlooked cause of hypokalemia is Gitelman syndrome[2,] which is a salt losing tubulopathy caused by mutation of SLC12A3 gene. This gene encodes for Thiazide sensitive transporter NCCTI(sodium – chloride co transporter). This NCCT is located in distal tubular cells of responsible for 7-10% electrolyte reabsorption.[3]Prevalance is estimated at 25 per million and accordingly, the prevalence in heterozygotic twins is 1%.[4]It affects males and females equally. In many cases of Gitelman's syndrome the disorder may go undiagnosed or misdiagnosed making it very difficult to estimate the actual frequency of Gitelman's syndrome in Normal population.

Gitelman patients are mostly thought to be asymptomatic. Often presented with asymptomatic hypokalemia, but on closer questioning 80% of patients complain of dizziness and fatigue; 70% complain of muscle weakness and

cramps[5].Signs and Symptoms vary between children and adults, but may include: salt craving ,weakness of muscle, fatigue, limited sports performance , fainting episodes, cramps, muscle spasms. Diagnosis include a number of blood and urine tests such as Blood gases ,Blood tests to measure magnesium, aldosterone, and renin. Urine analysis for sodium, potassium, and calcium. The mainstay treatment involves careful monitoring, high-sodium and potassium diet, and oral potassium and magnesium supplements. Potassium and magnesium supplements are used along with potassium sparing diuretics[6].

Case Report

A 30 year old female was admitted into the hospital with chief complaints of spasm of both upper limbs and lower limbs since one day, fever and cough since 2 days. Fever was intermittent type and was not associated with chills and rigors. Patient also complained about muscle cramps. Pain and burning sensation in the stomach was also noted in the patient.

A. Laboratory investigations:

B. Routine tests

Parameter	Value	Reference range(unit)
COMPLETE BLOOD COUNT		
Haemoglobin	10.7	12-16 (g/dL)
RBC	5.4	4-5 (millions/cumm)
WBC	7200	4000-10000(cells/cumm)
Platelet count	3lakhs	1.5-3 lakhs/micro liter
Erythrocyte sedimentation rate	45	(< 10mm/hr).
LIVER FUNCTION TESTS		
Total Bilirubin	1.1	<1.9mg/dl
Direct Bilirubin	0.1	<0.4mg/dl

C. Definitive tests

Parameter	Value	Reference range(unit)
SERUM CHEMISTRY/ SERUM ELECTROLYTES		
Sodium	128	(135-153 mEq/L)
Potassium	2.3	(3.5-5.0 mEq/L)
Chloride	103	(95-105 mEq/L)
Magnesium	1.2	(1.7-2.2mg/dL)
Calcium	7.8	(9-10.5mg/dL).
RENAL FUNCTION TESTS		
Serum Creatinine	0.5	(0.7-1.4 mg/dL)
Blood Urea- mg/dL	15	(20-40 mg/dL)

CT scan revealed calcific masses within joint capsule which shows chondrocalcinosis. See arrow in below figure 1.

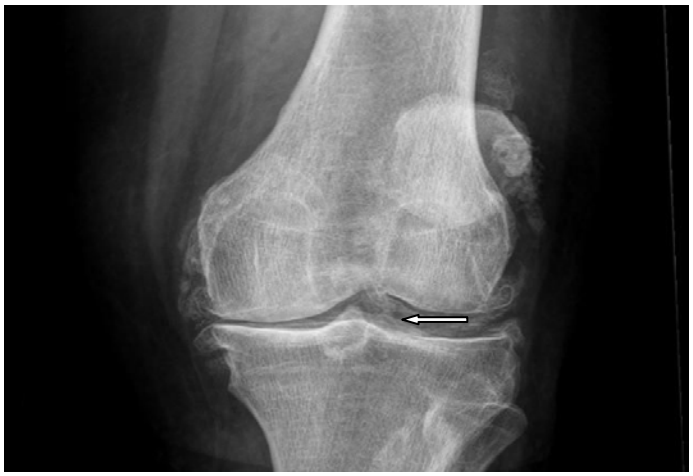

Figure 1.CT Scan of Left Leg

Interpretation: After performing definitive tests patient was diagnosed as Gitelman's Syndrome.

D. Treatment

The appropriate treatment included Tab. Shelcal (500 mg-OD), Tab. Flexon (BD), Syp. POTKLOR (10ml-TID), Inj.MgSO4(1amp in 1pint NS)- to reduce severe muscle cramps, Inj. Calcium Gluconate (20CC-OD),Cap. Renerve (500mg-OD).

E. Discharge medications:

The patient was discharged after 5 days after symptomatic relief. The patient was advised to continue Tab.Shelcal (500 mg-OD), Tab. Flexon (BD), Cap. Renerve(500mg-OD).

Discussion

Antenatal Bartter's syndrome, classical Bartter's syndrome and Gitelman's syndrome are the three recognized phenotypes of Bartter's syndrome. Mutations in several renal tubule transport protein have been shown to be responsible for this syndromes. In Gitelman's syndrome, mutations have been found in the thiazide sensitive Nacltransporter[7].

Disease is rarely diagnosed in asymptomatic adults with Hypokalemia and transient episode of weakness, abdominal pain, fever, nausea and vomiting[8, 9].Severe manifestations include chondrocalcinosis, rhabdomylosis and seizures. Progression to ESRD(End Stage Renal Disease)is quite rare.[10] In case study patient has symptoms of fever, hypokalemia and chondrocalcinosis.

The diagnosis of GS is generally based on clinical and biochemical findings. Disease-free intervals may be prolonged which results in delay of diagnosis until adulthood. Biochemically, disease is characterized by hypokalemia, hypochloremic metabolic alkalosis and hypocalciuria. [11]

In our case Potassium and calcium supplements are given to patient to minimize the symptoms and Flexon is given to treat chondrocalcinosis. Adequate supplementation of potassium and magnesium remain the main cornerstone in the treatment of GS. Oral therapy is difficult as large quantity of potassium chloride is required and oral magnesium often causes diarrhea. Hence it is sometimes not possible to recover the normal levels of electrolytes in the body of patient which ameliorates the symptoms furthermore.[11]

Conclusion

One of the rare causes of hypokalemia is Gitleman's Syndrome which is a challenge for physicians. Most asymptomatic patients with Gitleman's syndrome remain untreated. Lifelong supplementation with magnesium and potassium is needed. Cardiac work-up is needed to minimize cardiac diseases. Suitable treatment prevents patients from severe complications.

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