Case Report

DOI: 10.7860/JCDR/2020/43817.13654

Gitelman Syndrome: A Cause for Recurrent Hypokalaemia with Tetany

Internal Medicine Section

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ABSTRACT

An important genetic cause of recurrent hypokalaemia is defect in the distal tubules. This case report is about a 35-year-old male patient that presented to the casualty with acute onset of flaccid weakness of all the four limbs with muscle pain, cramps and tetanic spasm. Patient also had similar weakness in the past, 2 years back. On examination, he was normotensive with hypotonia noted in all four limbs with retained reflex. Evaluation showed hypokalaemia (1.4 mEq/L), hypomagnesaemia (1.1 mg/dL), metabolic alkalosis (pH 7.535, bicarbonate 33 mEq/L), increased urinary sodium (80 mEq/L) and potassium (60 mEq/L) and reduced urinary calcium excretion (0.73 mmols/L). Renal functions were normal and renal ultrasound showed normal size kidneys. Based on this clinical and biochemical background, a diagnosis of Gitelman syndrome was made patient was managed with intravenous and oral potassium chloride, injectable magnesium sulphate and normal saline. The aim this report was to emphasise that Gitelman syndrome though rare, is an important differential diagnosis for recurrent hypokalaemia presenting with tetany.

Keywords: Hypomagnesaemia, Metabolic alkalosis, Tetanic spasm

CASE REPORT

A 35-year-old male, farmer by occupation, presented with acute onset difficulty in using all four limbs with muscle pain, cramps and tetanic spasm, for one day. Patient also had similar compliant 2 years back. There was no history of loss of consciousness, seizure or trauma.

On examination, patient had quadriparesis with power of 3/5 and hypotonia. Deep tendon reflexes were present with flexor plantar response. Chovstek and Trousseau's sign were positive.

Investigation showed normal renal function, serum sodium value (140 mEq/L) with potassium (1.4 mEq/L) and magnesium (1.1 mg/dL) with serum calcium (9 mg/dL). Arterial blood gas analysis showed metabolic alkalosis with pH of 7.53 and serum bicarbonate 33 mEq/L. ECG showed QTc prolongation. Urine sodium value (80 mEq/L) and urine spot potassium (60 mEq/L) were found to be high; later the urine calcium was checked which was also found to be (0.73 mmols/L). Patient underwent ultrasound abdomen which showed normal size kidney. Patient was suspected to have hypokalaemic paralysis.

Patient was treated with potassium supplementation and serum electrolytes were monitored every 6th hourly. Magnesium correction was done. After two days, serum potassium increased to 3.3 mEq; limb power improved to 4/5. He was started on spironolactone, a potassium sparing diuretic, which prevents further episode of hypokalaemia.

The details of the therapy are: Intravenous potassium chloride 40 mEq in 500 mL normal saline over 5 hours × 3 doses for 2 days and oral potassium chloride 20 mL three times daily for 2 days from second day Injection magnesium sulphate 2 gram in 500 mL normal saline for 3 days.

The clinical background depicts recurrent flaccid paralysis, muscle cramps, spasm, positive Chovstek and Trousseau's sign with retained reflex and without sensory involvement. As there was improvement in weakness with potassium supplementation along with hypomagnesaemia and hypocalciuria in a normotensive individual, an empirical diagnosis of Gitelman syndrome was made. Genetic study for Gitelman syndrome was not done due to financial limitations.

DISCUSSION

Gitelman syndrome is an autosomal recessive condition leading to defect in the distal nephrons causing renal potassium loss, magnesium loss, metabolic alkalosis and hyper-reninemic hyperaldosteronism [1]. Important genetic defect noted in the gene encoding sodium chloride and magnesium transporter which is located in the thiazide-sensitive segment of distal nephron [2]. Mutations are usually seen in SLC12A3 gene in thiazide sensitive sodium chloride co-transporter, TRPM6 gene encoding for tubular magnesium transport [2].

Common clinical presentation includes muscle weakness spasms and cramps. Other features like episodes of fatigability, dizziness and fainting (due to reduced blood pressure) are also observed. Some patients also show positive Chovstek and Trousseau's sign. Most of the patients usually manifest seizure secondary to hypomagnesenemia [3]. Some patients have joint involvement in the form of swelling, tenderness and redness secondary to calcium deposition called as chondrocalcinosis as a consequence of hypomagnesaemia [4].

Laboratory investigation usually shows hypokalaemia, hypomagnesaemia, metabolic alkalosis and normal sodium levels. Urine analysis shows increased urine sodium and potassium with low calcium. Management usually is based upon the presenting symptoms. Usually asymptomatic patients need no intervention. If patient has symptomatic hypokalaemia, potassium replacement is needed [1,5-7]. Magnesium supplementation may be necessary in most of the patients. Some articles also showed the beneficial effect of potassium sparing diuretic like spironolactone and amiloride, if patient tolerates because usually they have blood pressure on the lower side [5,8,9].

CONCLUSION(S)

Though the common cause of recurrent hypokalaemia is familial hypokalaemic paralysis. Other causes like renal tubular acidosis, potassium losing nephropathy and enteropathy should also be ruled out. Tetany with normal blood ionised calcium is mostly due to hypomagnesaemia. Hypokalaemia, hypomagnesaemia and hypocalcuria in a normotensive is the common presentation of Gitelman syndrome. Long term follow-up is important in these genetic disorders to prevent recurrences.

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PLAGIARISM CHECKING METHODS: [Jain H et al.]

- Plagiarism X-checker: Feb 04, 2020
- Manual Googling: Mar 19, 2020
- iThenticate Software: Mar 30, 2020 (9%)

ETYMOLOGY: Author Origin

AUTHOR DECLARATION:

- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. NA

Date of Submission: **Feb 01, 2020**
Date of Peer Review: **Feb 27, 2020**
Date of Acceptance: **Mar 19, 2020**
Date of Publishing: **Apr 01, 2020**