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Endocrinology

PRESENTATION OF GITELMAN SYNDROME WITH HYPOCALCAEMIC TETANY: A RARE ENTITY

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ABSTRACT This a rare case report of a 49 year old lady diagnosed with "Gitelman Syndrome with Hypocalcaemic tetany". The lady, a known hypertensive, presented with complaints of bilateral jaw pain for four days; associated with tingling and numbness of skin around the mouth accompanied with generalised weakness. There was a history of recurrence of similar episodes in the past and a strange habit of excessive salt craving. On examination, Chvostek's sign and Trousseau's signs were positive. Blood tests showed low potassium and magnesium, and calcium which were corrected conservatively. Urine analysis of electrolytes showed low calcium with high potassium and magnesium. Subsequently, a diagnosis of Gitelman Syndrome with Hypokalaemic periodic paralysis was made.

KEYWORDS : Gitelman syndrome, Familial hypokalemia-hypomagnesemia syndrome, Hypocalcaemic tetany

INTRODUCTION:

Gitelman Syndrome or familial hypokalaemia-hypomagnesemia is characterised by hypokalaemic metabolic acidosis with significant hypomagnesemia and low urinary calcium excretion. It is an autosomal recessive disorder. Mutation in the solute carrier family 12, member 3 gene SCL12A3 – which encodes thiazide sensitive NaCl co-transporter – is found in the majority of patients. It has a prevalence of around 1 in 40000. Symptoms may include weakness, muscle pain, abdominal pain, vomiting. Patients may have sudden cardiac death rarely [1].

CASE REPORT:

A 49-year-old lady with a background history of Hypertension and was being treated with calcium channel blocker for the same presented with complaints of bilateral jaw pain for the past four days. She reported of a strange habit of excessive craving for salt intake. The jaw pain was acute on onset and was present throughout the day with no diurnal variation; however, it increased while chewing, biting and yawning. There was restricted movement of bilateral temporomandibular joints as well as tingling sensation and numbness of the skin around the mouth. She also complained of generalised weakness. There was also recurring history of similar episodes in the past.

On examination, the following signs were observed: Heart rate 76/min, regular, normal in volume and character; Respiratory Rate 18/min, regular, no accessory muscles of respiration working; SpO2 98% in room air; Blood Pressure 130/70mm of Hg. There was no pallor, icterus, oedema, clubbing or cyanosis. Jugular venous pressure was not elevated. On local examination of face, there was painful and restricted mouth opening with. On palpation, there was tenderness around both the temporomandibular joints. Chvostek's sign and Trousseau's sign were found to be positive. There was no other local or systemic positive finding.

The patient underwent emergency ABG which detected respiratory alkalosis with low potassium and calcium corrected conservatively. All other blood parameters were sent for investigation. The ECG did not show any significantly abnormal changes. USG of neck with bilateral parotids was within normal limits. She underwent CT of temporomandibular joints and parotids, which were normal. The NCV and EMG of facial muscles and muscles of mastication were normal. The laboratory investigations which were done during hospital stay are given in Table 1 which is given below.

Table 1: Investigations done during Hospital Stay

	Day 0	Day 2	Day 4
Haemoglobin	11		
TLC	9600		
ESR	20		
Urea	32		
Creatinine	0.8		
Albumin	4.2		4
Sodium	134	133	137
Potassium	2.4	3.1	3.6
Magnesium	0.8	1.6	1.9
Calcium	4.1	6.2	7.2
PTH		32	
Vitamin D		24	
24 Hr Urine Potassium			655
24 Hr Urine Magnesium			45
24 Hr Urine Calcium			31

Therefore, she was diagnosed with Hypocalcaemic tetany with hypokalaemia and hypomagnesemia. Opinion of the Nephrology department was taken and urine was also examined. The patient was found to have normal urine with routine findings of Hypocalciuria, hyperkaliuria and hypermagnesiuria. She was hence diagnosed with Gitelman Syndrome and was treated conservatively with potassium, magnesium and calcium supplementation. Additional blood test showed ANA negative. As a result, anti-hypertensive was changed to angiotensin receptor blocker losartan. She improved with all conservative medications and was discharged with advice to continue those.

DISCUSSION:

Our patient presented with acute pain around both temporomandibular joints with restricted jaw movement, perioral tingling sensation and numbness in the backdrop of a history salt craving. Clinically, she had features of hypocalcaemia, which was confirmed on blood and urine reports. She was also found to have hypokalaemia. She had history of similar episodes in the past, which was associated with generalised weakness.

Our patient had the symptom of salt craving. It can be a rare presenting symptom in Gitelman Syndrome, as reported by Merwe et al. after studying African population. It is probably caused by volume depletion secondary to renal sodium loss [2].

Calcium is essential for muscle contraction and neurotransmitter release. Hypocalcaemia is defined as serum calcium below 8.6mg/dl. Symptoms may range from asymptomatic to life-threatening conditions and they require urgent care. Disorders maybe divided into PTH-related and non-PTH-related. Our patient had normal PTH level and was thus evaluated for non-PTH-related causes. Intravenous calcium is essential for treatment of life-threatening features of hypocalcaemia, as were present in our patient. Oral calcium is required for long-term treatment [3].

Potassium imbalance is one of the most common electrolyte imbalance

encountered in clinical practice. Hypokalaemia is defined as potassium level below 3.5mEq/L. It can be asymptomatic or can cause life-threatening cardiovascular disease. It can be caused when there is increased intracellular shift of potassium or increased renal or digestive losses. Urinary potassium excretion and acid-base assessment are helpful for the diagnosis [4]. Our patient had recurrent episodes of generalised weakness with hypokalaemia, which was suggestive of hypokalaemic periodic paralysis. In a study conducted in India for aetiology of hypokalaemic periodic paralysis, thyrotoxicosis was found to be the leading cause followed by distal renal tubular acidosis and Gitelman Syndrome [5].

Hypomagnesemia is defined as serum Magnesium level below 1.8mg/dl. It may be caused by decreased oral intake, gastrointestinal or renal losses or redistribution of magnesium from extracellular to intracellular space. Fractional excretion of magnesium above 2% with normal renal function indicates renal magnesium wasting. Drugs, Bartter Syndrome and Gitelman Syndrome are the leading causes of renal magnesium loss [6].

All clinical symptoms as well as the blood and urine examination confirmed the diagnosis of Gitelman Syndrome. The presentation of Gitelman Syndrome has been very rarely reported, as described by Gandhi et al. [7]. The most probable cause of hypocalcaemia in Gitelman Syndrome is chronic hypomagnesemia which causes impaired parathyroid hormone secretion or refractoriness of bone and renal tubules to PTH [8]. This case presentation highlights a rare disorder of Gitelman Syndrome which becomes even rarer when associated with hypocalcaemic tetany.

CONCLUSION:

Our patient presented with complaints of bilateral jaw pain and spasm with a history of craving salt and recurrent episodes of bilateral upper and lower limb weakness. Clinical examination showed positive Trousseau's and Chvostek's signs. Blood investigations showed hypokalaemia, hypomagnesemia and hypocalcaemia. Urine investigations revealed hypocalciuria, hyperkaliuria and hypermagnesiuria. Hence, she was diagnosed with Gitelman Syndrome with hypocalcaemic tetany with hypokalemic periodic paralysis. Thus, we report a rare case of Gitelman Syndrome which presented with hypocalcaemic tetany.

She was diagnosed with meticulous clinical examinations, appropriate investigations and had complete recovery following conservative treatment.

PATIENT CONSENT: TAKEN.
CONFLICT OF INTEREST: NONE.

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