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Tetany caused by functional hypoparathyroidism in Gitelman Syndrome: a case report

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Abstract

Gitelman syndrome is an autosomal recessive salt-wasting tubulopathy affecting the distal convoluted tubule, characterised by hypokalaemia, metabolic alkalosis, hypomagnesaemia, hypocalciuria, and secondary hyperreninemic hyperaldosteronism, mimicking the effects of chronic thiazide diuretic use. Although hypocalcaemia is uncommon, it can occur due to hypomagnesaemia-induced suppression of parathyroid hormone (PTH) secretion, leading to functional hypoparathyroidism. Symptomatic hypocalcaemia and tetany are extremely rare presentations of Gitelman syndrome.

We report a 30-year-old man who presented with transient loss of consciousness and muscle cramps. He had experienced similar episodes over the preceding year, accompanied by peri-oral numbness and salt cravings. He had previously been diagnosed with primary hypoparathyroidism based on persistently low serum parathyroid hormone (PTH) levels despite calcium supplementation. Examination revealed carpopedal spasms and a positive Chvostek sign.

Laboratory investigations revealed hypokalaemia, metabolic alkalosis, hypomagnesaemia, hypocalciuria, and elevated renin and aldosterone levels, consistent with Gitelman syndrome and hypocalcaemia. Treatment with intravenous calcium, magnesium, and potassium resulted in symptom resolution. Due to persistent hypokalaemia, spironolactone was added. Genetic testing confirmed mutations in SLC12A3 consistent with Gitelman syndrome.

This case highlights the importance of evaluating serum magnesium in patients with unexplained or treatment-refractory hypocalcaemia. Magnesium deficiency can mimic hypoparathyroidism by suppressing PTH secretion and impairing the end-organ responsiveness to PTH. The combination of hypokalaemia, hypomagnesaemia, and hypocalciuria is characteristic of Gitelman syndrome and should prompt further evaluation, including genetic

Key words: hypocalcaemia, tetany, Gitelman syndrome, functional hypoparathyroidism, hypomagnesemia

Introduction

Gitelman syndrome (GS) is a rare autosomal recessive salt-wasting tubulopathy affecting the distal convoluted tubules (DCT), characterised by hypokalaemia, metabolic alkalosis, hypomagnesemia, hypocalciuria, and secondary hyperreninemic hyperaldosteronism. It essentially mimics chronic thiazide diuretic use due to the defective thiazide-sensitive Na-Cl cotransporter in the DCT.1 The estimated prevalence of GS is about 1 in 40,000 (approximately 25 per 100,000). Patients typically present during adolescence or adulthood with muscular weakness, cramps, or fatigue, although some remain asymptomatic. Hypocalcaemia is extremely rare in GS and, when present, is usually secondary to chronic magnesium deficiency-induced suppression of parathyroid hormone (PTH) release. In severe hypomagnesaemia, magnesium loss leads to functional

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hypoparathyroidism by inhibiting PTH secretion and inducing end-organ PTH resistance.[2,3] This mechanism can lead to refractory hypocalcaemia until magnesium levels are restored.

We present a case of GS in a young man who was previously misdiagnosed with primary hypoparathyroidism, highlighting the importance of recognising magnesium deficiency as a reversible cause of hypocalcaemia and the unusual presentation of GS with symptomatic hypocalcaemia and tetany.

Case presentation

A 30-year-old man presented after a transient loss of consciousness while climbing stairs, accompanied by generalised painful muscle cramps. He had experienced similar symptoms for approximately one year, often with peri-oral numbness and salt cravings during episodes. There were no palpitations, chest pain, tonic-clonic movements, incontinence, or post-ictal confusion.

His first syncopal episode occurred a year earlier, during which evaluation revealed hypocalcaemia with inappropriately low PTH, normal vitamin D and phosphate levels. Based on these findings, he was diagnosed with primary hypoparathyroidism and commenced on vitamin D (1000 IU/day) plus elemental calcium (2 g/day). Despite treatment, he required five hospital admissions over the year for recurrent, similar episodes.

On examination, he appeared well-nourished with a BMI of 23 and had no dysmorphic features. He had carpopedal spasms and a positive Chvostek sign. He was normotensive with a regular pulse, and no focal neurological deficits were noted. The electrocardiogram revealed a prolonged QTc interval of 480 ms. Laboratory results (with reference ranges) were as follows: serum potassium 3.1 mmol/L (3.5-5.0), magnesium 0.53 mmol/L (0.70-1.10), total calcium 1.86 mmol/L (2.20-2.60), intact PTH 20 pg/mL (15-65), 25-OH vitamin D 30 ng/mL (sufficient), phosphate 1.2 mmol/L (0.8-1.5). Renal and liver functions were normal.

Arterial blood gas revealed a metabolic alkalosis (pH 7.53, HCO_3^- 33 mmol/L, base excess +9.3). Urine studies revealed inappropriately high urine potassium (kaliuresis), low urine calcium excretion (calcium/creatinine clearance ratio <0.1), high urine chloride levels, and a urine pH of 7.5. Plasma renin activity was 228 μU/mL (4.4-46) and aldosterone level was 1102 pmol/L, consistent with secondary hyperaldosteronism.

These findings narrowed the differential diagnosis to either Bartter syndrome, Gitelman syndrome, or surreptitious diuretic use. The presence of profound hypomagnesaemia and hypocalciuria favoured Gitelman syndrome. He was admitted and treated with intravenous calcium gluconate, potassium chloride, and magnesium sulphate. His symptoms of tetany resolved with electrolyte repletion. He was transitioned to oral calcium and magnesium supplements. However, persistent hypokalaemia necessitated the addition of spironolactone (50 mg daily) for its potassium-sparing effects. This led to the normalisation of his serum potassium levels and the resolution of his muscle cramps. The patient remained normocalcaemic and asymptomatic on follow-up. Subsequent genetic testing confirmed biallelic SLC12A3 mutations, establishing the diagnosis of Gitelman syndrome.

Discussion

This case highlights the diagnostic challenge of hypocalcaemia with low PTH levels and emphasises the vital role of magnesium in calcium regulation. Primary hypoparathyroidism, whether postsurgical, autoimmune, or congenital, is a common cause of hypocalcaemia with inappropriately low PTH, but magnesium deficiency is a frequently overlooked cause of a similar biochemical pattern. Severe hypomagnesaemia can inhibit PTH secretion and cause resistance to PTH action, a phenomenon sometimes called “functional hypoparathyroidism”.[2,3] In such cases, patients present with hypocalcaemia and low or normal-low PTH despite normal parathyroid glands.[3] Recognising this is crucial, as calcium levels may not recover until magnesium is replenished. Indeed, our patient’s hypocalcaemia and inappropriately low PTH persisted despite calcium and vitamin D supplementation, which aligns with magnesium deficiency impairing PTH release. In our patient, the combination of hypokalaemia, metabolic alkalosis, hypomagnesaemia, and hypocalciuria was diagnostic of GS. Both GS and Bartter syndrome (BS) are hereditary renal tubulopathies that cause renal potassium wasting, volume contraction, and secondary hyperaldosteronism; however, there are essential differences in their typical features. Urinary calcium excretion is a key distinguishing factor: GS is classically associated with low urinary calcium (hypocalciuria), whereas BS is typically associated with normal or high urinary calcium (hypercalciuria). This reflects the different nephron segments involved: GS involves the DCT, where thiazide-like effects enhance calcium reabsorption. In contrast, BS involves the thick ascending limb, producing a loop diuretic-like effect and leading to calcium wasting. Magnesium handling also differs significantly. Hypomagnesaemia is present in most GS patients (due to impaired magnesium reabsorption in DCT). In contrast, BS patients typically have normal serum magnesium levels (only ~20-30% of classic BS cases have mild hypomagnesaemia).

Table 1. Diagnostic criteria for GS as per Kidney Disease: Improving Global Outcomes (KDIGO)

Diagnostic criteria as per KDIGO	Our patient
Chronic hypokalaemia (<3.5 mmol/l) with inappropriate renal potassium wasting (spot potassium-creatinine ratio >2.0 mmol/mmol [>18 mmol/g])	Serum potassium 2.8 mmol/L Spot urine potassium 38 mmol/L
Metabolic alkalosis	pH – 7.53 Bicarbonate – 33 mmol/L Base excess 9.3
Hypomagnesaemia (<0.7 mmol/l [<1.70 mg/dl]) with inappropriate renal magnesium wasting (fractional excretion of magnesium >4%)	Serum magnesium 0.53 mmol/L Renal magnesium wasting not performed.
Hypocalciuria (spot calcium-creatinine ratio <0.2 mmol/mmol [<0.07 mg/mg]) in adults.	Urine calcium to creatinine ratio – 0.023 mg/mg
High plasma renin activity or levels	Plasma Renin activity – 228 μU/ml (4.4-46)
Fractional excretion of chloride > 0.5%	Not performed
Low or normal-low blood pressure	Blood pressure 105/65 mmHg
Normal renal ultrasound	Normal

Clinically, BS often appears in infancy or early childhood, and antenatal forms can manifest with polyhydramnios and failure to thrive. It may also be associated with nephrocalcinosis or sensorineural deafness in specific subtypes. In contrast, GS usually presents later (in adolescence or adulthood) with muscle cramps, fatigue, or periodic paralysis, and patients are often normotensive and have normal growth. Another differentiating factor is prostaglandin E_2 activity: patients with BS have elevated renal prostaglandin E_2 production and frequently benefit from nonsteroidal anti-inflammatory drugs (NSAIDs) to reduce vasodilatory prostaglandins, whereas prostaglandin levels are normal in GS.[4,5]

These distinctions guided our evaluation – the presence of severe hypomagnesemia and marked hypocalciuria strongly indicated Gitelman syndrome over Bartter syndrome, despite the unusual finding of hypocalcaemia. This initially raised consideration of BS, since calcium is typically normal in GS. Genetic testing ultimately confirmed SLC12A3 mutations consistent with GS; however, in practice, a presumptive diagnosis can often be made based on clinical and biochemical features alone.[1] It is essential to note that surreptitious diuretic abuse can mimic GS or BS and must be excluded. Our patient denied any diuretic use, and the persistent electrolyte abnormalities during hospitalisation argued against a factitious cause. Diuretic screening can be used if there is suspicion of covert diuretic use.

Another notable aspect of this case is the presentation of symptomatic hypocalcaemia (tetany) as a feature of GS. Although GS typically does not cause hypocalcaemia, there have been scattered reports of hypercalcaemic tetany in GS attributable to magnesium depletion-induced hypoparathyroidism. Desai et al., reported a 28-year-old woman with GS who presented with carpopedal spasm and seizures due to hypocalcaemia that was unresponsive to calcium until magnesium was corrected.[6] Our case adds to the literature on this uncommon clinical presentation. Significantly, the patient's hypocalcaemia improved with magnesium repletion, reinforcing that hypomagnesaemia was the cause of his hypocalcaemia, via functional PTH suppression. The management of GS primarily focuses on correcting electrolyte abnormalities

and alleviating symptoms, as there is currently no cure for the underlying transporter defect. Lifelong oral supplementation with potassium and magnesium is the mainstay of management.[1,7] Importantly, the aim is to keep the patient asymptomatic rather than to fully normalise serum levels, since aggressive repletion may be limited by gastrointestinal tolerance and urinary losses.

According to consensus recommendations (KDIGO), reasonable targets in GS are to maintain $K^+ \geq 3.0$ mmol/L and $Mg2^+ \geq 0.6$ mmol/L, as full normalisation is often impractical.[1] Our patient required high doses of oral magnesium oxide and potassium chloride; despite this, his potassium remained in the low-normal range with ongoing symptoms. In cases of persistent hypokalaemia or symptomatic fluctuations, addition of a potassium-sparing agent can be beneficial.[1] We initiated low-dose spironolactone in our patient, which helped raise his potassium and reduce the aldosterone-driven renal losses. Alternatives include amiloride or eplerenone, which directly block distal tubular sodium reabsorption and thus decrease potassium excretion. Ensuring adequate dietary intake of sodium to help expand extracellular volume and consuming magnesium-rich foods is also advised. Finally, patient education is crucial; they should understand the benign nature of the condition in terms of life expectancy and the importance of adhering to supplements and follow-up. With proper management, most individuals with GS can maintain a good quality of life.[1] Long-term follow-up should include periodic monitoring of electrolytes and renal function. Although rare, complications such as arrhythmias (due to hypokalaemia), nephrocalcinosis (uncommon in GS due to hypocalciuria), or chondrocalcinosis should be monitored for. Genetic counselling might be provided, particularly for family screening and during pregnancy planning.

Conclusion

This case demonstrates a rare presentation of Gitelman syndrome with recurrent syncope and tetany caused by hypocalcaemia. It highlights the importance of testing serum magnesium levels in any hypocalcaemic patient, as this can differentiate true hypoparathyroidism from a reversible, magnesium-dependent PTH suppression. The classic laboratory triad of hypokalaemic metabolic alkalosis, hypomagnesaemia, and hypocalciuria should prompt confirmation of Gitelman syndrome through genetic testing or clinical criteria, as early diagnosis can prevent unnecessary treatments.

Author declaration

Conflict of interests

The authors declare that they have no conflict of interests.

Consent for publication

Informed written consent for publication was obtained from the patient.

Consent for publication

SI acquired and analysed the patient data and drafted the final manuscript. NJD substantively revised the manuscript. All authors read and approved the final manuscript.

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