CASE REPORT

Localised normocalcaemic tetany secondary to dehydration in an individual with Gitelman syndrome

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SUMMARY

Dehydration is a common presentation to any emergency department with symptoms ranging from lethargy, confusion, oliguria as well as those specific to the underlying cause. In this case we describe a young patient who following a short history of vomiting and abdominal pain developed carpopedal spasm and distal paraesthesia on a background of Gitelman syndrome. Biochemical blood analysis showed a marked hypokalaemia, hypomagnesaemia and mild metabolic alkalosis in addition to a prolonged QTc interval of 592 ms seen on ECG. Following fluid replacement and electrolyte correction his clinical symptoms resolved along with QTc normalisation. This case demonstrates a patient with a rare and interesting renal disorder who presented with typical biochemical and ECG abnormalities in addition to tetany in the presence of normal plasma calcium.

BACKGROUND

This case demonstrates a patient with a rare and interesting renal disorder who presented with typical biochemical and ECG abnormalities in addition to tetany in the presence of normal plasma calcium.

CASE PRESENTATION

A 25-year-old man presented by ambulance to our emergency department (ED) complaining of a short 1 day history of nausea, vomiting, intermittent upper abdominal pain and hand cramping. No respiratory, cardiovascular, urinary symptoms or altered bowel movements were noted. He drank alcohol approximately 10 units a week and denied any recent episodes of excessive drinking. There was no history of laxative or diuretic medications; however, he regularly took two SandoK tablets five times a day. Normally he remarked being fit and

well, playing active sports three times a week while working professionally 5 days a week. Interestingly, he disclosed that he had a rare renal disorder called Gitelman syndrome for which he was seen in a tertiary centre. Previously on two separate occasions he had been admitted to hospital due to complications of the condition.

Cardiac and respiratory systems were normal with abdominal examination noting mild epigastric tenderness only. There were no peripheral stigmata suggestive of any vitamin or mineral deficiencies (eg, koilonychia or leukonychia). Peripherally he was cool with a capillary refill time of 4 s. Neurological exam demonstrated contracture of the hands with inward digital deviation and abnormal distal sensation to both sharp and light touch. The remaining upper limbs were normal with the exception of excitable reflexes throughout. Chvostek's sign was negative.

INVESTIGATIONS

Blood tests showed no evidence of infection or derangement of liver enzymes, albumin, amylase, urea and creatine. A venous blood gas sample (see table 1) showed a metabolic alkalosis with pH 7.470 and hypokalaemia of 2.4. ECG demonstrated normal sinus rhythm of 83 bpm and a prolonged QTc interval of 592 ms (see figure 1A). Laboratory blood analysis confirmed hypokalaemia and in addition hypomagnesaemia of 0.62. Corrected calcium was normal at 2.23.

DIFFERENTIAL DIAGNOSIS

- Hypocalcaemia
- Hypomagnesaemia
- Hypoparathyroidism
- Bartter's syndrome
- Acute intermittent porphyria

Table 1 Electrolyte imbalances during admission. All samples venous.

Parameter	ED blood gas Day 0	ED lab sample Day 0	PCU lab sample Day 1	PCU blood gas Day 1	Ambulatory clinic Day 3
pH (7.35–7.45)	7.470	n/a	n/a	7.460	n/a
K ⁺ (3.5–5.5)	2.4	2.5	2.8	3.0	3.7
Mg ²⁺ (0.7–1)	n/a	0.62	0.53	n/a	0.70
cCa ²⁺ (2.1–2.6)	n/a	2.23	2.20	n/a	n/a
Na ⁺ (135–145)	136	136	139	140	141
HCO ₃ ⁻ (22–28)	33.6	n/a	n/a	36	n/a
BE (–2 to +2)	+9	n/a	n/a	+10	n/a

PCU, progressive care unit; n/a, not applicable

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Figure 1 (A) ECG on admission to A&E resus exhibiting prolonged QT interval. QT 504/QTc 592 ms. (B) ECG on admission to progressive care unit K⁺ 2.5, QT 449/QTc 511 ms. (C) ECG following further intravenous electrolyte correction K⁺ 3.0, QT 372/QTc 407 ms.

TREATMENT

A diagnosis of dehydration-induced severe electrolyte disturbance secondary to GS was made. The patient was treated in the resuscitation area of the ED while on cardiac monitoring. There

he received 1 litre of 0.9% normal saline stat followed by 1 litre of 0.9% normal saline with 40 mmol of potassium chloride over 4 h. Additionally, an infusion of 20 ml of 10% calcium gluconate was given for suspected hypocalcaemia following the

initial clinical and biochemical features of carpopedal tetany and a prolonged QTc interval.

Subsequently, he was admitted to the progressive care unit (PCU) under the acute medical team for monitoring, fluid resuscitation and electrolyte stabilisation. He received further intravenous infusions of 0.9% normal saline and electrolyte correction with potassium chloride for hypokalaemia as well as magnesium sulfate for hypomagnesaemia confirmed on venous blood sampling.

Collectively, in the first 24–26 h the patient received 6.25 litres of 0.9% normal saline, 160 mmol of potassium chloride, 20 ml of 10% calcium gluconate and 5 g of magnesium sulfate.

OUTCOME AND FOLLOW-UP

Serial ECGs with repeat blood sampling showed stabilisation of the QT/QTc interval as K^+ and Mg^{2+} levels normalised (see table 1 and figure 1). No other significant arrhythmias were seen to develop. The localised tetany visibly improved and the patient self-discharged after approximately 30 h feeling clinically back to baseline despite the advice that further monitoring was needed. Two days later in an ambulatory clinic, clinical review and repeat blood tests showed sustained improvement.

DISCUSSION

GS, also known as familial hypokalaemia–hypomagnesaemia was first described in 1966¹ as a rare autosomal recessive salt-losing renal tubular disorder characterised by hypomagnesaemia, hypocalciuria and hypokalaemic metabolic alkalosis. Although generally deemed rare with a prevalence of 1:40 000, it is one of the most commonly inherited renal tubular disorders.² This patient presented to our ED with typical GS features of tetany, muscle weakness, paraesthesia and abdominal pain in the context of an acute ‘salt-losing’ event (vomiting). Other GS symptoms along the spectrum include salt-craving, thirst, nocturia and chondrocalcinosis.³

Hypokalaemic metabolic alkalosis shown in the venous blood gas fittingly reflects the pathophysiology of GS. Genetic mutations result in defective NaCl reabsorption in the distal convoluted tubules with compensatory increased plasma renin activity and subsequent secondary hyperaldosteronism.⁴ The ensuing secretion of K^+ into tubular lumen and secretion of hydrogen ions in exchange for Na^+ into cortical collecting tubules generates a hypokalaemic metabolic alkalosis.

The initially prolonged QTc seen in this patient is characteristic of GS with up to 50% of affected individuals having a slight-to-moderate QTc lengthening.³ This is theorised to stem from hypokalaemia and hypomagnesaemia prolonging duration of cardiomyocyte action potentials. Fortunately, clinically significant arrhythmias or even cardiac arrests are rare. Intriguingly, a

prolonged QTc interval is often unaffected by electrolyte levels; however, in this patient this was seen to normalise following plasma K^+ and Mg^{2+} correction.

This case highlights classic clinical and biochemical manifestations of a rare and poorly recognised genetic renal disorder. As such there has been no report of a similar ED presentation of an individual with GS in the literature. Given its incidence and yet poor recognition, an interesting question presents itself regarding whether significant numbers of GS-affected individuals who seek medical help remain undiagnosed. The condition itself is variable with some 180 speculated NaCl cotransporter gene mutations identified and subject to differing phenotypic penetrance. Absence of unique clinical symptoms such as tetany or paraesthesiae may contribute to lack of suspicion among clinicians. Fortunately in this case, despite an unclear initial diagnosis, employment of methodical and structured management enabled identification of abnormalities and subsequent resolution of symptoms with treatment.

Learning points

- ▶ Gitelman syndrome is a unique renal disorder often not recognised or suspected due to the lack of clinician awareness.
- ▶ Potentially significant ECG changes could develop which may or may not be independent of electrolyte states.
- ▶ Focus of treatment lies in the correction of biochemical abnormalities.
- ▶ Patients require close follow-up and in some long-term management may necessitate daily electrolyte supplements.

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Competing interests None.

Patient consent Obtained.

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