CASE REPORT

Gitelman’s syndrome
A RARE PRESENTATION MIMICKING CAUDA EQUINA SYNDROME

We describe a case of bilateral weakness of the lower limbs, sensory disturbance and intermittent urinary incontinence, secondary to untreated Gitelman’s syndrome, in a 42-year-old female who was referred with presumed cauda equina syndrome. On examination, the power of both legs was uniformly reduced, and the perianal and lower-limb sensation was altered. However, MRI of the lumbar spine was normal. Measurements of serum and urinary potassium were low and blood gas analysis revealed metabolic alkalosis. Her symptoms resolved following potassium replacement.

We emphasise the importance of measurement of the plasma and urinary levels of electrolytes in the investigation of patients with paralysis of the lower limbs and suggest that they, together with blood gas analysis, allow the exclusion of unusual causes of muscle weakness resulting from metabolic disorders such as metabolic alkalosis.

Hypokalaemia is a rare cause of paralysis. We describe the clinical course of a 42-year-old woman who presented with extensive neurological symptoms which were found to be due to Gitelman’s syndrome; a renal tubular disorder characterised by hypokalaemia, metabolic alkalosis and often hypomagnesaemia. Patients with this syndrome may be hypotensive and may have low levels of urinary calcium with elevated levels of plasma renin. A mutation in the NCCT gene, which codes for the sodium chloride co-transporter at the distal convoluted tubule of the kidney, leads to increased excretion of sodium. This causes a drop in plasma volume and activation of the renin-angiotensin-aldosterone system. Angiotensin stimulates resorption of sodium at the level of the collecting duct via the epithelial sodium channels at the expense of increased secretion of potassium and hydrogen. This leads to hypokalaemia and alkalosis. At a cellular level, the juxtaglomerular apparatus is usually hyperplastic. Gitelman’s syndrome has an autosomal recessive inheritance pattern and usually manifests in the second and third decades.

Case report
A 42-year-old female presented with a 12-hour history of a sudden onset of bilateral weakness and loss of sensation in the lower limbs, decreased perineal tone and episodic urinary incontinence, associated with a seven-day history of progressively worsening pain in the lower back. Bowel habit was normal. It was thought that she had cauda equina syndrome.

Three years previously, she had sustained an anterior wedge fracture of the eighth thoracic vertebra following a fall. This was managed conservatively with rest and subsequent physiotherapy and the fracture healed with a less than 10% reduction in the anterior height of the vertebral body.

On admission her medication included daily oral magnesium and potassium which had been prescribed by her family doctor, but she was uncertain for what reason.

Neurological examination revealed normal tone and reflexes with down-going plantars bilaterally, but reduced power (MRC grade 2/5) in both legs on all movements about the hip, knee and ankle joints. Straight leg raising was possible to 70° on the right side but was limited to 50° on the left by pain in the leg. Sensation was globally abnormal in the right leg and diminished in the left leg in a variable distribution from L4 to L5-S1. In the lumbar area, there was altered sensation extending to T12, but no corresponding change in sensation was found over the abdomen. Neurological examination of the upper limbs was normal and coordination was unaffected. There was normal perivaginal sensation but decreased perianal sensation and absent anal tone.

Plain radiographs of the thoracic and lumbar spine revealed the healed fracture of T8,
with nothing to suggest compromise of the spinal canal at any level. Urgent MRI of the lumbar spine was normal.

An electrocardiogram showed a resting sinus tachycardia of 120 beats per minute and U waves, which are present in patients with hypokalaemia. The blood pressure was 96 mmHg/50 mmHg. Initial biochemical investigations revealed a serum potassium level of 2.2 mmol/L (normal range (n) = 3.5 to 5) and magnesium of 0.72 mmol/L (n = 0.7 to 1.0). Arterial blood gas analysis, performed with the patient breathing room air, demonstrated a pH of 7.45 (n = 7.35 to 7.45) and a bicarbonate level of 27.2 mmol/L (n = 22 to 24). The urinary potassium was 23 mmol/L (n = 25 to 120) and the urinary chloride was 165 mmol/L (n = 20 to 250). The pH of the urine was 8 (n = 5 to 7).

Based on the patient’s normal MRI, her low levels of serum and urinary potassium in combination with a metabolic alkalosis and hypotension, and the emergence from the condition of a positive family history for the condition, a diagnosis of Gitelman’s syndrome was made. Appropriate treatment was started, with intravenous potassium replacement and administration of spironolactone oral tablets of spironolactone 25 mg and 36 mmol potassium and magnesium a decade earlier. Although our patient had never been given a formal diagnosis, she had been started on oral potassium and magnesium supplementation of potassium and magnesium a decade earlier. It seems that her sister had previously been diagnosed with Gitelman’s syndrome. Although our patient had never been given a formal diagnosis, she had been started on oral supplementation of potassium and magnesium a decade prior to presentation by her family doctor, based on serial recordings of low serum potassium. In addition she admitted that she had, in the past, experienced similar but less severe episodes of muscle weakness, for which she had been advised to consume high-potassium foods, which resulted in resolution of the symptoms.

**Discussion**

Also known as familial hypokalaemia-hypomagnesaemia, and one of the most commonly inherited renal tubular disorders, 2 Gitelman’s syndrome was first described in 1966 3 and is considered to be a clinical subtype of Bartter’s syndrome. 7,8 Both syndromes can present with a variety of additional symptoms including polydipsia, polyuria, fatigue, muscle cramps, weakness, tetany and hypotension. 1,4,5 Chondrocalcinosis may be a late feature of Gitelman’s syndrome secondary to magnesium depletion, although this is rare. 9 While it tends to present in the second and third decades, 10 early onset of disease and consequent retardation of growth have been described. 11

The diagnosis is based on a combination of clinical and biochemical findings. The urinary calcium is often low as a result of the increased resorption of calcium via apical calcium channels due to hyperpolarisation of cells, which occurs secondary to impaired resorption of sodium and loss of chloride ions. 12

Gitelman’s syndrome arises as a result of a mutation in the NCCT (sodium chloride co-transporter) gene, 13 most commonly SLC12A3. More than 140 different mutations have been identified. 2 It is typically inherited in an autosomal recessive pattern and has a prevalence of 1 in 40 000. 4 Phenotypic heterogeneity is a feature of Gitelman’s syndrome and it has been suggested that females with the condition may be less severely affected than males. 14 The prognosis is generally excellent and progression to end-stage kidney disease is rare. 2

This unusual case demonstrates the importance of measuring plasma and urinary levels of electrolytes in the investigation of patients with bilateral weakness of the lower limbs. Failure to recognise hypokalaemia on presentation in this instance may have led to an alternative course of treatment and the potentially fatal side-effects of low serum potassium, 15 while rapid potassium replacement resulted in resolution of symptoms for our patient. Although arterial blood gas analysis may not always be indicated in the investigation of paralysis, this case demonstrates that it should be considered in cases where plasma and urinary levels of electrolytes are abnormal, in order to exclude an inherited renal tubular disorder such as Gitelman’s syndrome.

No benefits in any form have been received or will be received from a commercial party related directly or indirectly to the subject of this article.

**References**


