Finger Drop, Head Drop, and Asymmetric Hypokalemic Paralysis: An Atypical Presentation of Gitelman Syndrome

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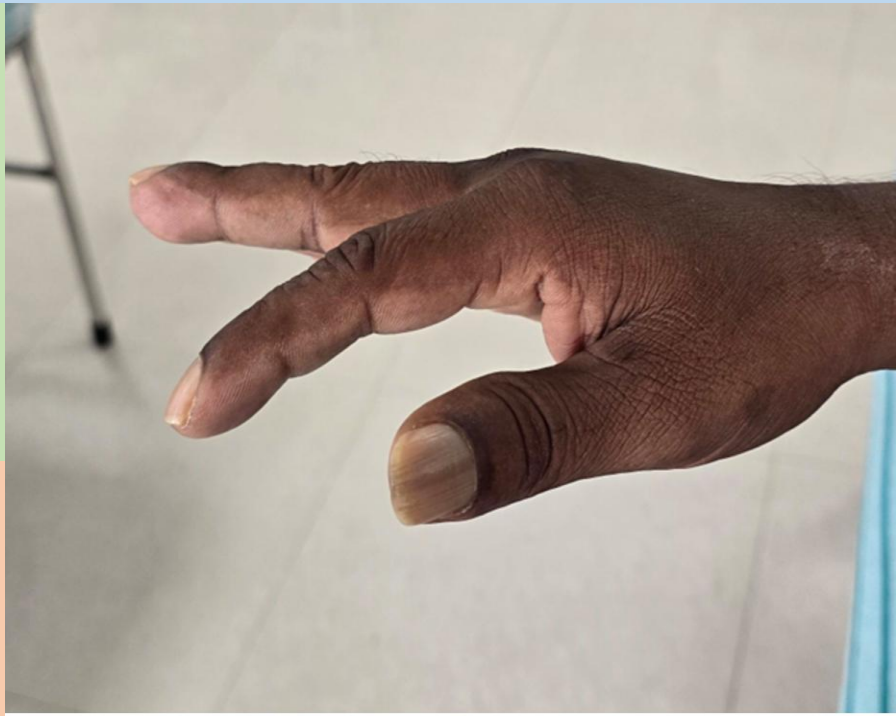
AIMS-Gitelman syndrome(GS), also known as familial hypokalemia-hypomagnesemia, is a rare, inherited salt-losing tubulopathy classically characterized by hypokalemia, hypomagnesemia, metabolic alkalosis, and hypocalciuria[1]. While it often presents with mild, non-specific symptoms like fatigue or cramps, severe neuromuscular manifestations such as acute flaccid paralysis are exceedingly rare[2]. We report a unique case of GS presenting with severe, progressive, and asymmetric quadriparesis with focal features, initially mimicking Guillain-Barré syndrome (GBS)[3].

CASE HISTORY -A 46-year-old man with diabetes and chronic alcohol use presented with acute, progressive neurological decline. His illness began with sudden-onset neck pain, followed within 24 hours by pain and weakness in the left upper limb, which progressed within hours to involve the right upper limb, with difficulty holding objects and finger drop of the right index finger and thumb.



Index and thumb drop

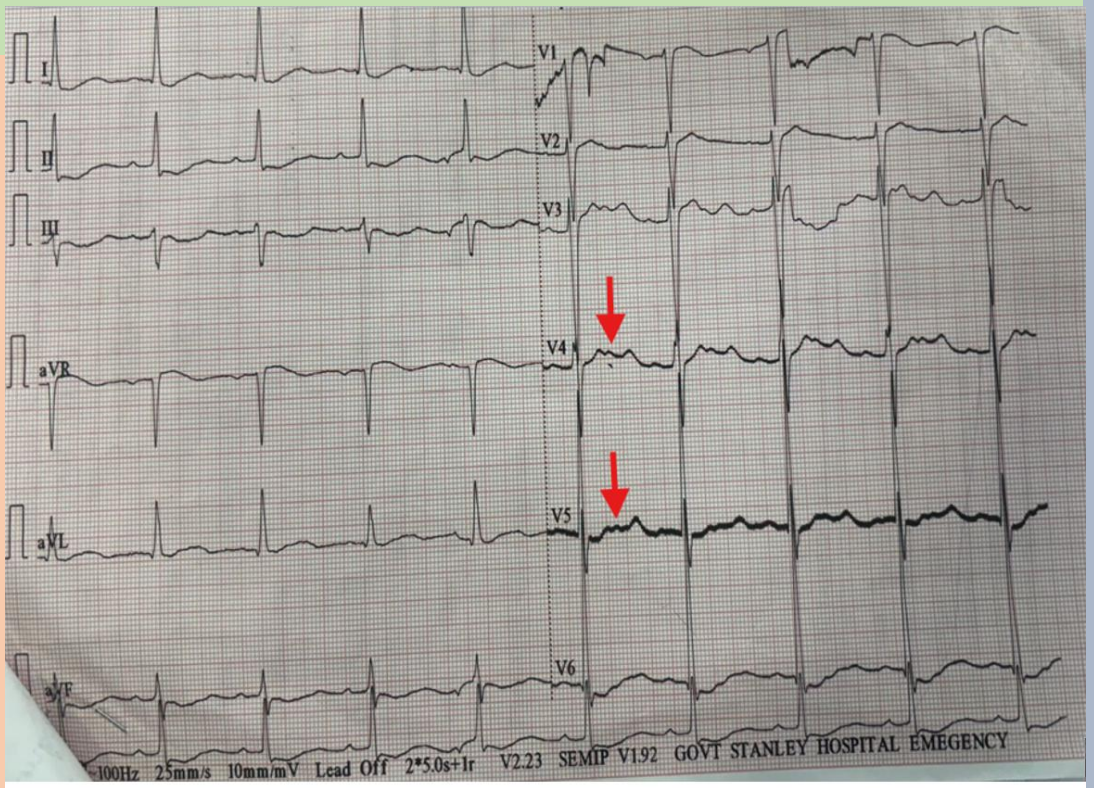
Over the next two days, weakness extended asymmetrically to the lower limbs (right > left), causing gait difficulty and inability to rise from a squatting position. The following day, he developed a head drop. There was no history of diuretic/laxative use, vomiting, diarrhea, or hearing loss. On examination, the patient was alert and oriented, with a blood pressure of 128/82 mmHg. Neurological evaluation showed asymmetric, proximal-predominant flaccid quadriparesis: upper limbs left weaker (4/5) than right (4+/5), lower limbs right weaker (4/5) than left (4+/5). Marked neck extensor weakness explained his head drop. Deep tendon reflexes were globally reduced, while sensation was preserved.

Materials and Methods -

→ Laboratory investigations showed **severe hypokalemia** with a serum potassium of (2.3 mmol/L), hypomagnesemia(1.3mg/dl), and metabolic alkalosis. An electrocardiogram (ECG) demonstrated prominent U waves. Serum creatine phosphokinase (CPK) was markedly elevated at 10,904 U/L and urine dipstick test was positive for heme pigment (in the absence of red cells on microscopy –suggestive of myoglobinuria), confirming rhabdomyolysis[4], which subsequently decreased with treatment. A 24-hour urine potassium excretion of more than 30 mEq/day indicates inappropriate renal potassium wasting. Our patient's 24-hour urine potassium excretion was 34 mEq/day, and his urine chloride was high at 144 mEq/L. Persistently high urine chloride (>20 mEq/L) in the setting of hypokalemia and metabolic alkalosis is a key feature that helps differentiate renal salt-wasting tubulopathies from other causes, such as surreptitious vomiting, where urine chloride is typically low (<20 mEq/L). These findings confirmed ongoing renal potassium and chloride losses.

→The combination of **hypokalemia, metabolic alkalosis, normal blood pressure, and renal salt wasting** narrowed the differential diagnosis to an inherited salt-losing tubulopathy, specifically Bartter syndrome (BS) or Gitelman syndrome (GS). The key distinguishing feature between these two is urinary calcium excretion. BS is typically associated with normal or elevated urinary calcium (hypercalciuria), whereas GS is characterized by reduced urinary calcium (hypocalciuria). The patient's spot urine calcium was low at 3 mg/dL, strongly supporting the diagnosis of GS. Furthermore, the presence of significant hypomagnesemia is a classic feature of GS, whereas it is usually absent in the most common adult form of BS (type 3)[1,2].

Nerve conduction studies (NCS) revealed absent sensory nerve action potentials (SNAPs) in all limbs and reduced compound muscle action potentials (CMAPs).



U waves

→ These findings were interpreted as an acute metabolic insult superimposed on a background of chronic axonal sensorimotor neuropathy, possibly related to his long-standing, undiagnosed metabolic condition or comorbidities such as diabetes and alcohol use. Although genetic testing is the gold standard for confirming GS, it was not performed in this case; however, the classic biochemical profile made the clinical diagnosis highly reliable [3].

DISCUSSION

- This case is notable for its highly atypical presentation of Gitelman syndrome, with asymmetric weakness, finger drop, and head drop—features rarely reported and initially mimicking GBS[2,3].
- The occurrence of severe rhabdomyolysis (CPK >10,000 U/L), likely from profound hypokalemia impairing muscle perfusion and Na^+/K^+ ATPase activity, further adds to its uniqueness[4].
- The pathophysiology of GS involves a defective NCC transporter in the DCT that impairs NaCl reabsorption, causing volume depletion and activating the renin-angiotensin-aldosterone system (RAAS). The increased distal delivery of sodium to the collecting ducts, combined with high aldosterone levels, promotes sodium reabsorption in exchange for potassium and hydrogen ions, leading to persistent hypokalemia and metabolic alkalosis. Hypomagnesemia likely results from TRPM6 downregulation in the atrophied DCT, while hypocalciuria arises from increased proximal calcium reabsorption due to volume contraction and enhanced distal calcium reabsorption via TRPV5 channels[1].

CONCLUSION

Gitelman syndrome, though typically a mild disorder, can manifest as a severe neurological emergency with acute, asymmetric, and focal hypokalemic paralysis that can mimic Guillain-Barré syndrome. This case underscores the importance of considering inherited tubulopathies in the evaluation of acute flaccid weakness, where timely serum electrolyte testing and prompt replacement can rapidly reverse paralysis and prevent misdiagnosis[1,2,4].

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