Abstract

Introduction: Gitelman syndrome (GS) is a rare inherited renal tubulopathy characterized by metabolic abnormalities, including hypokalemia, metabolic alkalosis, hypomagnesemia, and hypocalciuria. It results from mutations in the SLC12A3 gene, primarily affecting the sodium-chloride cotransporter (NCC) in the distal convoluted tubule. GS diagnosis can be challenging due to variable features and overlapping conditions like Bartter syndrome. While traditionally considered benign, GS can present with severe manifestations. Genetic testing aids diagnosis but is costly. This case report highlights the complexities in diagnosing refractory hypokalemia associated with GS.

Case presentation: An 18-year-old female presented with one-week-long generalized body weakness and myalgia, primarily in her lower limbs, unrelated to meals or specific times. Her medical history was remarkable with recurrent hypokalemic episodes treated with oral potassium replacement therapy for six months. Physical examination revealed prominent proximal muscle weakness than distal weakness but the rest of the examination was unremarkable. Further extensive evaluation revealed GS as the cause for this chronic hypokalemia leading to muscle weakness.

Conclusion: Gitelman syndrome is a rare, complex disorder with diverse biochemical abnormalities and varying presentation. Diagnosis is challenging due to these variations and limited genetic testing. Hence, when dealing with a patient suffering from chronic hypokalemia, conducting thorough investigations is vital. This comprehensive evaluation aids in accuratelydiagnosisand developing a lifelong treatment plan aimed at minimizing potential complications.