

Abstract

Gitelman syndrome is a disease with an autosomal recessive inheritance pattern which presents with various electrolyte abnormalities including hypokalaemia, hypomagnesemia, hypocalciuria, metabolic alkalosis and hypotension. This syndrome usually becomes apparent from early childhood to early adulthood.

Herein we report a case of a 45-year-old female with a background history of well-controlled type 2 diabetes mellitus presenting with a one-week history of progressively worsening bilateral lower limb weakness. It was followed by bilateral thigh pain and reduced urine output. Examination revealed bilateral lower limb proximal muscle weakness with diminished reflexes and thigh tenderness. There were diminished reflexes in the upper limbs also. Investigations revealed, persistent severe hypokalaemia with metabolic alkalosis and evidence of rhabdomyolysis. Based on the clinical and biochemical parameters, the patient was diagnosed with hypokalemic paralysis with rhabdomyolysis complicated by acute kidney injury after excluding other possible causes. The cause for hypokalaemia was renal loss of potassium with metabolic alkalosis and normal blood pressure. With other supportive evidence, Gitelman syndrome was diagnosed as the cause of hypokalemia but did not undergo confirmation with genetic studies. This was her first presentation with such symptoms. This patient is unique as Gittleman Syndrome as it does not usually present for the first time in a middle-aged person. She was managed with supportive treatment along with oral and intravenous potassium and magnesium supplementation and fluid resuscitation after which she made a successful recovery.

Gitelman syndrome can present in adulthood as a rare cause of hypokalaemia. Therefore, clinicians should consider it as a differential diagnosis in the evaluation of persistent hypokalaemia.