Abstract

Gitelman syndrome(GS) is rare, inherited renal tubulopathy that presents with low serum potassium concentration and normal blood pressure. Underlying pathogenesis is secondary to dysfunction of a Na/Cl co-transporter (NCCT) located in the distal convoluted tubule, secondary to genetic mutations in SLC12A3 gene.

It is an association of hypokalemic metabolic alkalosis, and low-normal blood pressure coupled with hypomagnesaemia and hypocalciuria. Majority of the patients present during childhood or early adulthood. Gitleman syndrome is not frequently accompanied with growth retardation.

Here we describe a 14-year-old Sri Lankan school boy who presented with a febrile illness for three day duration without any evidence of systemic involvement. His height was 135cm (<<3rd percentile), body weight was 27kg (<<3rd percentile) with a with pre pubertal external genitalia development, (Tanner stage 1.) His blood pressure was in low normal range (90/50 mmHg) and rest of the system examination was normal.

His biochemical investigations revealed persistent hypomagnesaemia, hypokalemia, metabolic alkalosis, hypocalciuria which favour a diagnosis of Gitleman syndrome with a short statue.