CONCURRENT GITELMAN SYNDROME-LIKE TUBULOPATHY AND GRAVE'S DISEASE

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Gitelman syndrome (GS) is a rare autosomal recessive disorder characterized by the loss of function mutation of the solute carrier family-12 member-3 (SLC12A3) gene, encoding for sodium-chloride cotransporter of the distal convolute tubule. GS is characterized by hypokalemia, hypomagnesemia, metabolic alkalosis, and hypocalciuria. GS-like syndrome has been described rarely. Hyperthyroidism due to Grave's disease (GD) is characterized by the presence of autoantibodies to thyrotropin receptors. Concurrent occurrence of GS and GD is rarely reported, that too exclusively from far-east Asian populations.

We describe a case of a 45-year-old man who presented with severe muscle weakness; the evaluation showed volume depletion, hypokalemia, hypomagnesemia, renal potassium and magnesium wasting, metabolic alkalosis, and hypocalciuria. A tentative diagnosis of Gitelman's Syndrome was made on the above findings. He was also detected to have Grave's Disease based on hyperthyroidism with low serum TSH levels and the presence of anti-TSH receptor antibodies. He received intravenous potassium chloride (KCl) in 0.9% saline for the first 3 days and oral KCl for the next 6 days till the serum potassium was normalized [Figure 2]. He also received intravenous magnesium sulphate 2 gm on day 2 and oral magnesium oxide varying from 800 to 1200 mg/day.

He also received neomercazole 10 mg three times a day for hyperthyroidism. A month later, serum potassium was 3.7 mEq/L and serum magnesium was 1.5 mg/dL while on eplerenone 25 mg per day and magnesium oxide 1200 mg/day. Genetic analysis by target gene sequencing showed an autosomal dominant heterozygous mutation in exon 14 of TRPM4 gene that resulted in the amino acid substitution of asparagine for serine at codon 633 of chromosome 19, and no mutations were detected in SLC12A3 and CLCNKB genes.

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