

GITELMAN SYNDROME: THREE CASES OF A RARE GENETIC DISORDER IN A TERTIARY CARE HOSPITAL

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Gitelman syndrome is a salt-losing tubulopathy caused by mutation of genes encoding sodium chloride (NCCT) and magnesium transporters in the thiazide-sensitive segments of the distal nephron. It is characterized by renal potassium wasting, hypokalemia, metabolic alkalosis, hypocalciuria and hypomagnesemia. Here we present three cases those we encountered in Shaheed Suhrawardy Medical College Hospital. A 20-year-old male who was a soldier of Border Guard Bangladesh presented to us with muscle cramps and tingling & numbness for last 05 days. On examination, we found normotension with positive trousseau's sign. Investigation revealed hypokalemia with hypomag-nesemia. 24 hours urinary electrolytes were indicating urinary loss. As ABG revealed metabolic alkalosis and urinary calcium creatinine ratio was <0.15, we sealed our diagnosis to Gitelman syndrome. A 34-year-old normotensive school teacher who experienced hypokalemia four years ago and was on potassium supplement on demand came to us with sudden onset, rapidly progressive quadriplegia for last 05 days. This time symptom didn't resolve after taking potassium supplement. On Examination, we found flaccid quadriplegia with diminished jerks. Investigation revealed hypokalemia with hypomagnesemia. On further digging, we found diminished 24 hours urinary calcium as well as metabolic alkalosis on ABG. The final one that we encountered was a 40-year-old female normotensive housewife who presented with generalized weakness as well as tingling & numbness for last 10 days. Physical examination was inconclusive but Investigations revealed hypokalemia, hypomag-nesemia&Metabolic alkalosis. Hypocalciuria was evident from both 24 hours urinary calcium & urinary calcium creatinine ratio. In all occasions, all other possible differentials were excluded. We opted for genetic mutation study as well as family screening but in vain. In a nutshell, any electrolyte imbalance shouldn't be only managed by supplementation rather finding out the route of loss & arterial blood gas analysis may be a key to the ultimate diagnosis.

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