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Novel Genetic Variants in Koreans causing Gitelman Syndrome

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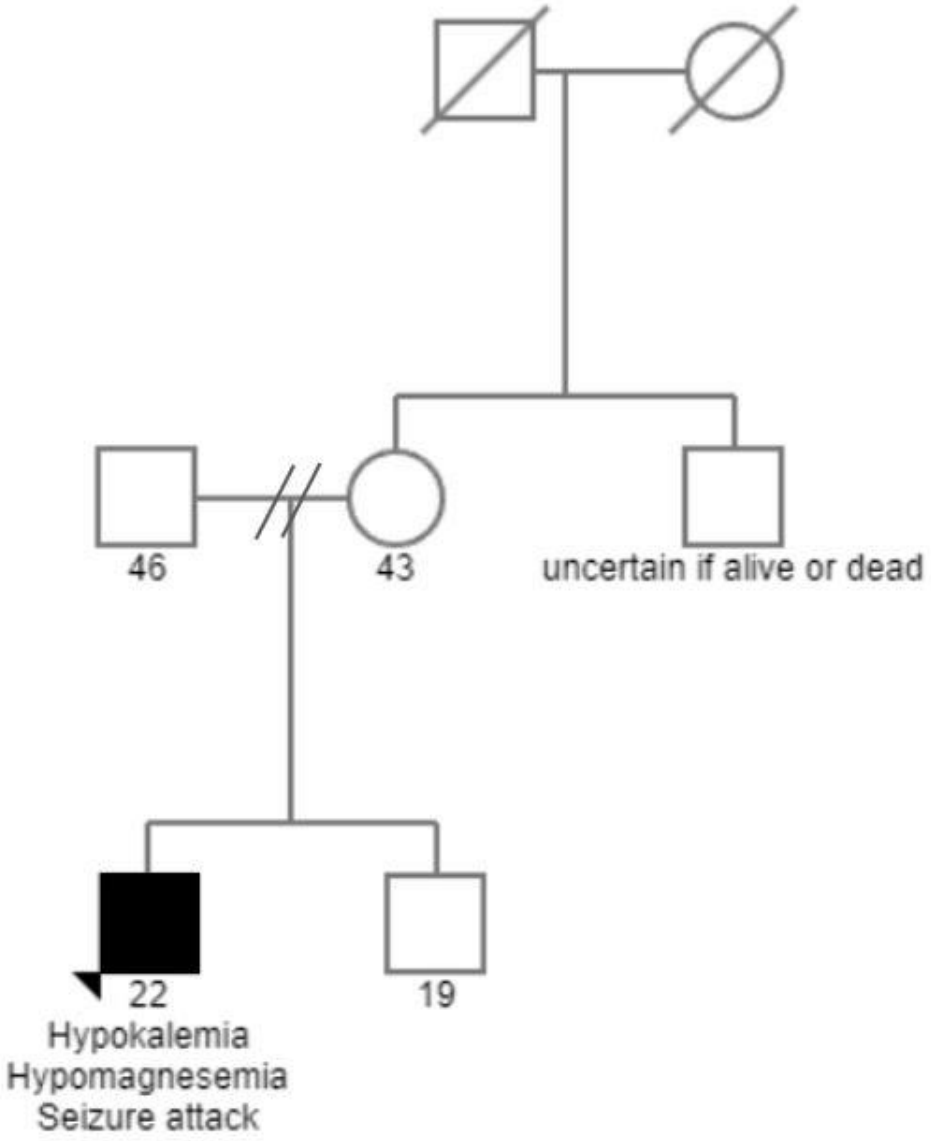
Case Study : Gitelman syndrome, a genetic disorder characterized by renal electrolyte absorption abnormalities, manifests as hypokalemia, metabolic alkalosis, hypomagnesemia, reduced urinary calcium excretion, and hypermagnesuria. It results from loss-of-function mutations in the sodium-chloride cotransporter in the distal tubule epithelium, specifically in both alleles of the SLC12A3 gene on chromosome 16's long arm. The frequency and clinical manifestation of these mutations exhibit significant ethnic and geographical variation. This case represents the first identified genetic variants of Gitelman syndrome in a Korean population. A 22-year-old male, presenting with hyperventilation, limb numbness, and general weakness, was admitted to the emergency room. The patient had a history of repeated symptoms and seizures, which had resulted in multiple hospitalizations at various medical institutions. Laboratory tests revealed hypokalemia (serum potassium level of 2.25mEq/L) and hypomagnesemia (serum magnesium level of 1.03mg/dL), necessitating admission to the nephrology department. Following aggressive intravenous and oral potassium supplementation, the patient showed significant improvement. Additional laboratory findings including urinary potassium excretion of 18.15mEq/gCr, metabolic alkalosis on arterial blood gas analysis, urine chloride level of 131mEq/L, and a low urine calcium to creatinine molar ratio of 0.004, leading to suspicion of Gitelman syndrome. Next-generation sequencing panel testing, targeting the SLC12A3 gene mutation, identified pathogenic variants, particularly the mutations c1387G>A and c2129C>A, with minor allele frequencies of 0.0028% and 0.0021% in the general population, respectively. Notably, these genetic variants had not been previously documented in the Korean population. This case represents the first reported genetic variants of Gitelman syndrome in Koreans, underscoring the importance of continued research and documentation of genetic variations across populations.

GITELMAN PEDIGREE.jpg


46
43
uncertain if alive or dead
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Hypokalemia
Hypomagnesemia
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