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Genetic Analysis of the Gitelman Syndrome Coexisting with Osteogenesis Imperfecta

Se Jin Park[1], Ju Hyung Kang[1], Haing-Woon Baik[2]
[1]Department of Pediatrics-Nephrology, Eulji University Hospital, Korea, Republic of
[2]Department of Biochemistry & Molecular Biology, Eulji University School of Medicine, Korea, Republic of

Case Study

Gitelman syndrome (GS) is an autosomal recessive disorder caused by loss-of-function mutations in *SLC12A3*, which encodes the Na-Cl cotransporter (NCC). Osteogenesis imperfecta (OI) is an autosomal dominant disorder caused by the inheritance of mutations mainly in the *COL1A1* gene, resulting in bone fragility and deformity. In this study, we aimed to investigate the clinical and genetic manifestations in a 7-year-old boy with OI, who had electrolyte abnormalities and his four family members.

Complete sequence analysis of *COL1A1* revealed a novel mutation, c.268G>T, p.Glu90del. The gene mutation of OI in the patient's older brother was inherited from his mother, and the younger brother had no mutation. Two pathogenic mutations (c.179C>T, p.Thr60Met and c.1763C>T, p.Ala588Val) in *SLC12A3* resulting in GS were also identified in the patient. The OI-related genetic mutation in the patient was consistent with that in the patient's mother. The GS-related genetic mutations were inherited from each parent.

This study is the first to identify compound heterozygous variants in the *SLC12A3* gene and a novel mutation in the *COL1A1* gene in patients with OI and GS. Our findings indicate that genetic analysis is recommended to differentiate GS from BS, as clinical manifestations do not provide an accurate diagnosis.