**VDR Gene Analysis of Four Patients with Hereditary 1,25 Dihydroxyvitamin D-Resistant Rickets**

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We present the VDR gene analysis results of four hereditary 1,25-dihydroxyvitamin D-resistant rickets (HVDRR) patients with severe skeletal dysplasia, alopecia, and hypocalcaemia. Genomic DNA was extracted from peripheral blood samples of these four patients. Whole gene sequencing was performed for VDR gene. We identified the same p.Q152*(c.454G>T) homozygous mutation in VDR gene in three of these patients. One of the patients had a homozygous p.R50*(c.148C>T) mutation in this gene. HVDRR is an autosomal recessive disease caused by mutations in VDR gene. We reported four patients, one of whom had a new mutation in VDR gene.

**Key words:** Hereditary 1,25-dihydroxyvitamin D resistant rickets, VDR gene

**A Case of Odontohypophosphatasia and Family Investigation**

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**Introduction:** Early tooth loss could be the consequence of local or systemic diseases. We present an odontohypophosphatasia case with autosomal dominant mutation in ALPL gene.

**Case:** A three-year-old boy was admitted to our pediatric endocrinology clinic with tooth loss without any other dental or gingival diseases. His serum levels of calcium, phosphorus, alkaline phosphatase, parathormone, and 25-hydroxy vitamin D levels were 9.7 mg/dL, 5.9 mg/dL, 70 U/L, 32.2 pg/mL, and 18.9 ng/mL, respectively. We considered that the patient has odontohypophosphatasia. ALPL gene was analyzed and heterozygous autosomal dominant c.346G>A (p.A116T) mutation was detected. ALPL gene analysis was performed in all members of the family. While his father has no mutation, his mother, brother, and sister have the same heterozygous mutation in the same locus.

**Conclusion:** Odontohypophosphatasia should be considered in patients with early tooth loss. It can present without extremely low alkaline phosphatase levels.

**Key words:** Hypophosphatasia, odontohypophosphatasia, ALPL gene