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

Esra Deniz Papatya Çakır[1], Özgür Aldemir[2], Seyit Ahmet Uçaktürk[3], Erdal Eren[4], Samim Özen[5]

[1]Bakirköy Dr. Sadi Konuk Training and Research Hospital, Clinic of Pediatric Endocrinology, İstanbul, Turkey

[2]Mustafa Kemal University Faculty of Medicine, Department of Medical Biology and Genetics, Hatay, Turkey

[3]Dışkapı Pediatric Hematology and Oncology Research and Training Hospital, Clinic of Pediatric Endocrinology, Ankara, Turkey

[4]Uludağ University Faculty of Medicine, Department of Pediatric Endocrinology, Bursa, Turkey

[5]Ege University Faculty of Medicine, Department of Pediatric Endocrinology, İzmir, Turkey

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

Esra Deniz Papatya Çakır[1], Mehmet Türe[2], Halil Sağlam[3], Seyit Ahmet Uçaktürk[4], Şahin Erdöl[5], Erdal Eren[6], Tahsin Yakut[2], Ömer Tarım[6]

[1]Bakırköy Dr. Sadi Konuk Training and Research Hospital, Clinic of Pediatric Endocrinology, İstanbul, Turkey

[2]Uludağ University Faculty of Medicine, Department of Medical Genetics, Bursa, Turkey

[3]Uludağ University Faculty of Medicine, Department of Pediatric Endocrinology and Metabolism, Bursa, Turkey

[4]Dışkapı Pediatric Hematology and Oncology Research and Training Hospital, Clinic of Pediatric Endocrinology, Ankara, Turkey

[5]Uludağ University Faculty of Medicine, Department of Pediatric Metabolism, Bursa, Turkey

[6]Uludağ University Faculty of Medicine, Pediatric Endocrinology, Bursa, Turkey

Introduction: Early tooth loss could be the consequence of local or systemic diseases. We present an odondohypohosphatasia 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 odontohypohosphatasia. *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