

# Clinical and Genetic Analysis of Five Patients with Vitamin D-Dependent Rickets Type 1A

Bülent Hacıhamdioğlu<sup>1</sup>, Gamze Özgürhan<sup>1</sup>, Zeynep Karakaya<sup>1</sup>, Ece Güven<sup>1</sup>

<sup>1</sup>Health Sciences University Suleymaniye Women Maternity and Child Diseases Training and Research Hospital, Istanbul, Turkey

## Introduction

- 👉 The CYP27B1 gene encodes 25-hydroxyvitamin D-1 $\alpha$ -hydroxylase.
- 👉 Mutations of this gene cause vitamin D-dependent rickets type 1A (VDDR-IA, OMIM 264700), which is a rare autosomal recessive disorder.
- 👉 Here in we report 5 patients with 1 $\alpha$ -hydroxylase deficiencies.

## Method

- 👉 We studied 5 patients from 3 families who diagnosed as 1 $\alpha$ -hydroxylase deficiency genetically.

## Results

- 👉 All patients had hypocalcemia, hypophosphatemia, hyperphosphatasemia, elevated serum PTH, normal or high vitamin D levels, and low or inappropriately normal calcitriol levels at presentation (Table).
- 👉 Patients were diagnosed less than 18 months years old. All patients had a family history of consanguinity.
- 👉 Homozygous mutations in the CYP27B1 gene were found in all the patients.
- 👉 Four of them have splice donor site mutation in intron 1 (c.195 + 2 T>G), causing partial retention of the intron and a shift in the reading frame.

	Calcium (mg/dl)	Phosphorus (mg/dl)	PTH (pg/ml)	25-OHD (ng/ml)	1-25-OHD (pg/ml) N: 25-153	Mutation analyses for CYP27B1 gene
Patient 1	8.0	2.3	441	26	15	Homozygous mutation on intron 1 (c.195 + 2 T>G)
Patient 2	5.9	3.41	925	41	<1.3	Homozygous mutation on egzon 8 p.Phe443Profs*24
Patient 3	6.2	2.8	980	120	12	Homozygous mutation on intron 1 (c.195 + 2 T>G)
Patient 4	7.8	3.0	625	45	6	Homozygous mutation on intron 1 (c.195 + 2 T>G)
Patient 5	8.0	2.9	546	50	11	Homozygous mutation on intron 1 (c.195 + 2 T>G)

## Conclusion

- 👉 Clinically, all the patients required calcium and calcitriol initially and then continuously calcitriol treatment.
- 👉 Autosomal recessive diseases are common in countries where the consanguineous marriages are common.
- 👉 VDDR-IA should be kept in mind patients with vitamin D resistant rickets.

