VDR GENE ANALYSIS RESULTS OF FOUR PATIENTS WITH 1,25-DIHYDROXYVITAMIN D RESISTANT RICKETS

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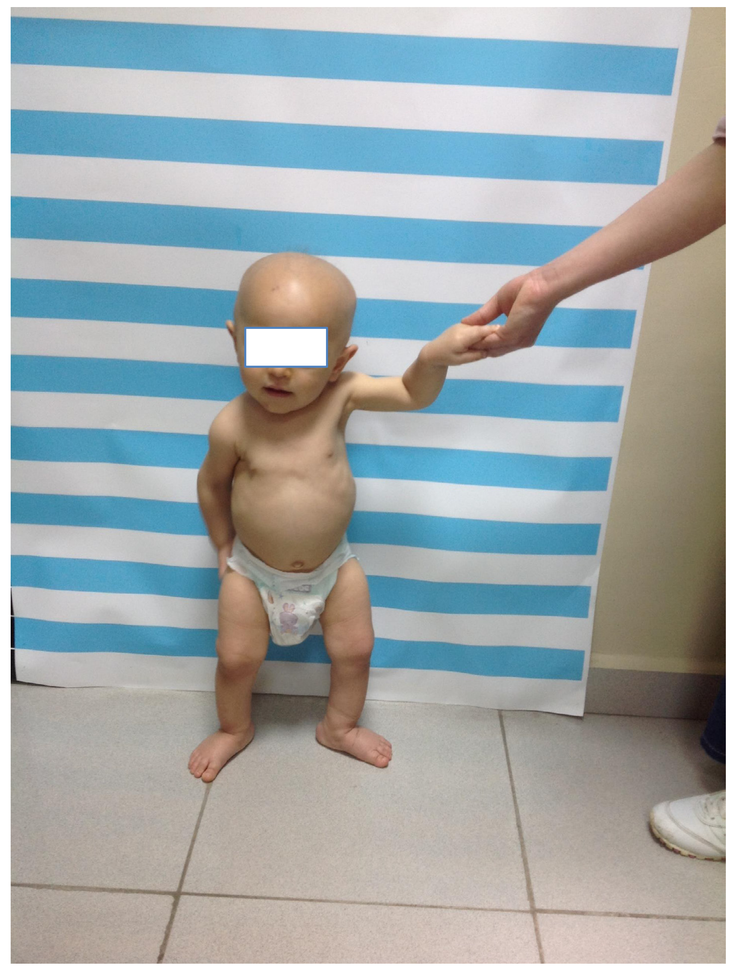
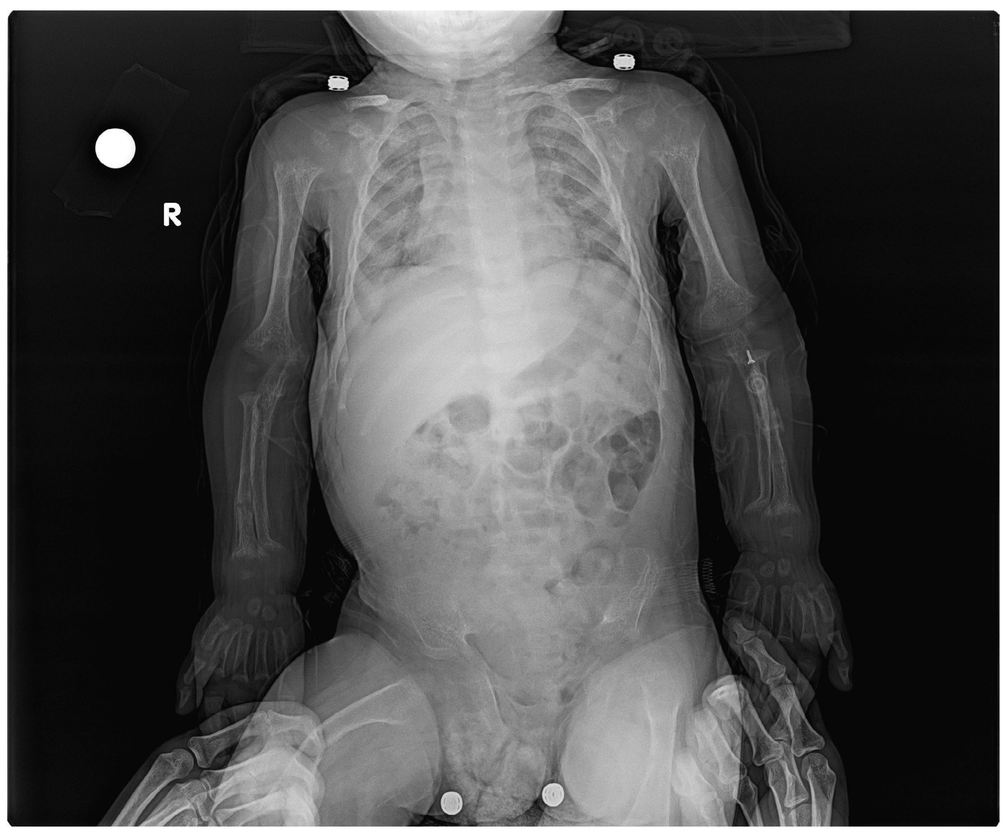
OBJECTIVE: Mutations in the vitamin D receptor (VDR) are associated to hereditary 1,25-dihydroxyvitamin D-resistant rickets (HVDRR). We performed VDR gene analysis of four patients with clinical diagnosis of HVDDR presenting alopecia, skelethal dysplasia and hypocalcemia.

METHOD: Genomic DNA extracted from peripheric blood sample. Whole gene sequence analysis performed.

RESULTS: We found homozigot p.Q152X(c.454G>T) mutation in three patients. Two of this three patients were siblings. There is homozygous p.R50X(c.148C>T) mutation in one patient.

	CA (mg/dL)	P (mg/dL)	ALP (U/L)	25OHVİTD (ng/mL)	PTH (pg/Ml)	CILINICAL FINDINGS	MUTATION
PATIENT 1	7.1	3	738	45	485	Alopecia Bone deformities Papular skin lesions	p.Q152X (c.454 G>T)
PATIENT 2	7.5	3.3	1250	33.9	840	Alopecia Bone deformities Papular skin lesions	p.Q152X (c.454 G>T)
PATIENT 3	7,6	3,2	956	33	562	Alopecia Bone deformities	p.Q152X (c.454 G>T)
PATIENT 4	8.03	2.69	1081	42	712	Alopecia	p.R50X(c.148 C>T)





DISCUSSION AND CONCLUSION : HVDRR is a rare autosomal ressesive disease caused by the inactivating mutations in VDR gene. Early onset ricketsial changes in skelethal system, alopecia, hypocalcemia, secondary hyperparathyroidism and elevated 1,25 Dihydroxyvitamin D levels are characteristic findings of this disease. Here we report clinical and laboratory findings of four patients.

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