



# VITAMIN D RECEPTOR (VDR) GENE POLYMORPHISMS AND THE RISK OF LOW BONE MASS IN TYPE 1 DIABETIC PATIENTS

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## BACKGROUND AND AIM:

Osteoporosis is a common skeletal disease characterized by low bone mass and microarchitectural deterioration with increased susceptibility to fracture. Osteoporosis has a complex etiology and is considered to be a multifactorial polygenic disease. Low bone mineral density (BMD) and fracture risk are associated with type 1 diabetes (T1D). There are more than 150 genes associated with bone mineral density. Vitamin D receptor (VDR) polymorphisms have been suggested to be associated with the diabetic complications. Our aim was to investigate the frequency of occurrence of vitamin D receptor (VDR) - (FokI, BsmI, Apal, TaqI) - single nucleotide polymorphisms (SNPs) in type 1 diabetic patients.

## SUBJECTS AND METHODS:

✓ We studied 62 type 1 diabetic (T1D) patients (26 men and 36 women; mean age  $31,46 \pm 8,55$ ; duration of the disease  $13,40 \pm 7,41$ ; HbA1c  $8,25 \pm 0,95\%$ ).

✓ Bone mineral density was measured by dual-energy X-ray absorptiometry.

✓ QIAamp DNA Blood Mini Kit» («Qiagen», USA) was used to purify DNA from whole blood, gene polymorphisms were detected in PCR-RFLP (restriction fragment length polymorphism) analysis. The following restriction enzymes were used to determine the appropriate polymorphism:

- VDR-FOKI – FokI (BseGI)
- VDR-Apal – Apal
- VDR-BSMI - BsmI (Mva1269I)
- VDR-Taql - TaqI

## RESULTS:

The presence of the mutant allele VDR-FokI was detected in 75 % of cases (in 40% cases as heterozygotes and in 35% as homozygotes). VDR- BsmI SNPs was found in 91 % of cases (in 56% cases as heterozygotes and in 35% as homozygotes). VDR- TaqI SNPs was found in 53 % of cases (in 47% cases as heterozygotes and in 6% as homozygotes). VDR- Apal SNPs was found in 72 % of cases (in 38% cases as heterozygotes and in 34% as homozygotes). (see Table 1).

Table 1.

Data are expressed as %.

	N	WW	%	WM	%	MM	%
VDR-FokI	62	15	25	25	40	22	35
VDR- BsmI	62	6	9	34	56	22	35
VDR- TaqI	62	29	47	29	47	4	6
VDR-Apal	62	17	28	24	38	21	34

## CONCLUSIONS:

The results of the study reflect the high frequency of vitamin D receptor (VDR) - (FokI, BsmI, Apal, TaqI) SNPs which probably may explain the occurrence of low bone mineral density in type I diabetic patients.