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 ± 8.55; duration of the disease 13.40 ± 7.41; HbA1c 8.25±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-TaqI - 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 %.

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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.