VARIANTS POLYMORPHISMS OF GENES VITAMIN D RECEPTOR (VDR)

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A polymorphism is a genetic variant that appears in at least 1% of the population. These changes can occur in non-coding parts of the gene (introns), so they would not be seen in the protein product. Changes in these regulatory parts of the gene would then affect the degree of expression of the gene, and thus the levels of the protein. For instance, chanes in the 5'-promoter of the VDR gene can affect mRNA expression patterns and levels, while 3' untranslated region (UTR) sequence variations can affect the mRNA stability and protein translation efficiency. However, the changes can take plays in exonic parts of the DNA, then leading to changes in the protein sequence. Nonetheless, changes in exonic sequences of the DNA which do not alter the protein structure are also possible, and are called synonymous polymorphisms. Often these changes create or abolish sites for restriction enzymes to cut the DNA. Digestion with the enzyme then produces DNA fragments of a different length which can be detected by electrophoresis. These polymorphisms are called Restriction Fragment Length Polymorphisms (RFLPs). The existence of several RFLPs in the VDR gene has been described using different restriction enzymes. Examples of these include the *Tru*91, *Taq*I, *Bsm*I, *Eco*RV, and *Apa*I. All of these RFLPs are located between the 8 and 9 exons and lay in an area with unknown function. A different case of RFLP is the so-called *Fok*I. This polymorphism was described in the early nineties in exon 2, and consisted of T to C change. The next polymorphisms (Cdx2) consisted in a G to A change in the promoter region of the VDR gene. That change is within the binding site for an intestinal-specific transcription factor called CDX2. Recently the polymorphism has also been described among different racial groups. The 3' UTR of the VDR gene is also a source of several different polymorphisms. However, conflicting reports over the number and position of the polymorphisms exist in the literature. Morrison et al. and Durrin et al. reported 1 and 7 different polymorphisms, respectively. The discovery of genetic variants linked with susceptibility of diseases can be the key to advances in preventive medicine.