DETECTION OF TaqI, ApaI, BmsI e FokI POLYMORPHISMS IN VIT.D RECEPTOR (DVR) GENE

AMPLI Set VDR polymorphisms Cat. n. 2.020

D vitamin promotes intestinal and renal calcium assimilation and it is essential for the bone mass development and the support. D vitamin is involved in controlling the proliferation, cellular differentiation and immuno-modulation. For instance, in the immune system vitamin D promotes the monocytes differentiation and inhibits lymphocytes trough cytokines increase (IL-2, IL12 or interferon). In some carcinoma vitamin D has a kind of anti-proliferative activity. D Vitamin's effects are mediated by its nuclear receptor (VDR)as it sets up an heterodimeric complex with the retinoic acid receptor and it interacts with trascription factors. VDR (12q12-14) codifies a protein of 427 amino acids, which regulates calcium homeostasis and it has probably a great genetic effect on bone densitometry (BMD). In region 3 of VDR human gene some polymorphic sites were found and identified by the endonucleases restriction T*aqI and BsmI*, *ApaI;* there is an other polymorphic variation, known as *FokI*. It has been extensively shown that there is a functional participation of VDR alleles in calcium homeostasis and in bone mineralization. Some studies have been underlined the VDR gene interaction with calcium, calcium absorption and calcium level in the diet. Allelic variations clarifies 70% of genetic effects on bone density. The **polymorphism *Fok*1** consists of nucleotide substitution T-C in the first codon of the translated region of the VDR gene . This polymorphism causes a difference in the protein length; the shortest one is the most efficient, whereas the longest one is associated with low BMD. The **polymorphism *BsmI***, localized in gene VDR intron 8, a nucleotidic variation A-G,is associated with a stability variation of the transcript and with a BMD value decrease. The **polymorphism *ApaI*,** in the intron 7, is a nucleotide variation T-G. It influences the mineral bone density depending on the presence or the absence of A variance. The **polymorphism *TaqI***, in gene VDR exon 9, is nucleotide variation T-C. This polymorphism is associated with an turnover increase of bone cells and, therefore, with an increase of osteoporosis risk. The method consists in amplification by PCR with specific primers and by enzymatic digestion with specific endonucleases (TaqI, ApaI, BsmI e FokI).

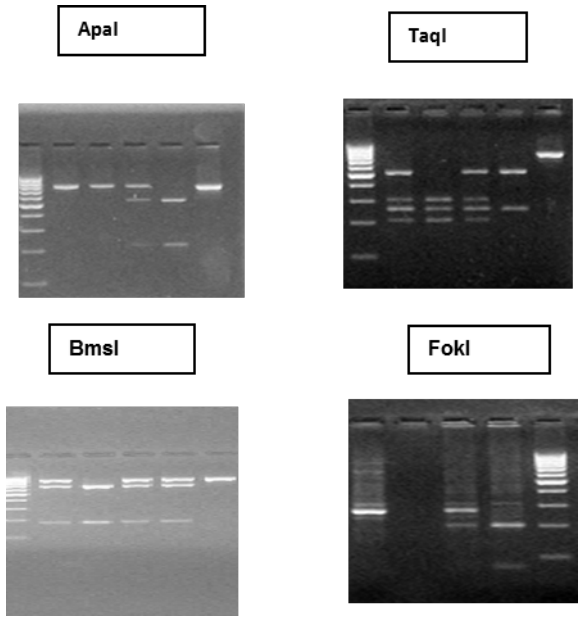
Principle of assay : A) extraction of genomic DNA B) amplification C) enzymatic digestion D) detection on agarose gel
Applicability: genomic DNA extracted by whole blood.
Number of tests : 24 x 4 polymorphisms

KIT CONTENTS AND STORAGE

AMPLIFICATION and DIGESTION	
ApaI- TaqI PCR mix	-20°C
BmsI PCR mix	-20°C
FokI PCR mix	
sterile H_2O	-20°C
Taq Polymerase (5U/µl)	-20°C
ApaI enzyme (10 U/µl)	-20°C
TaqI enzyme (10 U/µl)	-20°C
BmsI enzyme (10 U/µl)	-20°C
FokI enzyme (2 U/µl)	-20°C
Digestion BUFFER 10X for Apa I	-20°C
Digestion BUFFER 10X for TaqI	-20°C
Digestion BUFFER 10X for Bms I	-20°C
Digestion BUFFER 10X for FokI	-20°C
Mutated control	-20°C
WT control	-20°C

Stability: over 18 months if correctly stored.

ANALYSIS OF RESULTS



References:
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