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TNFRSF11B Gene Polymorphisms A163G and G11811C in Prediction of Osteoporosis Risk

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Abstract : Osteoporosis is a complex health disease characterized by low bone mineral density, which is determined by an interaction of genetics with metabolic and environmental factors. Current research in genetics of osteoporosis is focused on identification of responsible genes and polymorphisms. TNFRSF11B gene plays a key role in bone remodeling. The aim of this study was to investigate the genotype and allele distribution of A163G (rs3102735) osteoprotegerin gene promoter and G1181C (rs2073618) osteoprotegerin first exon polymorphisms in the group of 180 unrelated postmenopausal women with diagnosed osteoporosis and 180 normal controls. Genomic DNA was isolated from peripheral blood leukocytes using standard methodology. Genotyping for presence of different polymorphisms was performed using the Custom Taqman®SNP Genotyping assays. Hardy-Weinberg equilibrium was tested for each SNP in the groups of participants using the chi-square ($\chi 2$) test. The distribution of investigated genotypes in the group of patients with osteoporosis were as follows: AA (66.7%), AG (32.2%), GG (1.1%) for A163G polymorphism; GG (19.4%), CG (44.4%), CC (36.1%) for G1181C polymorphism. The distribution of genotypes in normal controls were follows: AA (71.1%), AG (26.1%), GG (2.8%) for A163G polymorphism; GG (22.2%), CG (48.9%), CC (28.9%) for G1181C polymorphism. In A163G polymorphism the variant G allele was more common among patients with osteoporosis: 17.2% versus 15.8% in normal controls. Also, in G1181C polymorphism the phenomenon of more frequent occurrence of C allele in the group of patients with osteoporosis was observed (58.3% versus 53.3%). Genotype and allele distributions showed no significant differences (A163G: $\chi 2=0.270$, $p=0.605$; $\chi 2=0.250$, $p=0.616$; G1181C: $\chi 2= 1.730$, $p=0.188$; $\chi 2=1.820$, $p=0.177$). Our results represents an initial study, further studies of more numerous file and associations studies will be carried out. Knowing the distribution of genotypes is important for assessing the impact of these polymorphisms on various parameters associated with osteoporosis. Screening for identification of “at-risk” women likely to develop osteoporosis and initiating subsequent early intervention appears to be most effective strategy to substantially reduce the risks of osteoporosis.

Keywords : osteoporosis, real-time PCR method, SNP polymorphisms
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