Abstract

Association Study of TP53(Arg72pro ,Intron 3 duplication 16 bp) and XRCC3(Thr241Met) genes polymorphisms in women affected by breast cancer in esternAzarbayjan of Iran

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Introduction: Breast cancer is the second most common cause of cancer related death in women worldwide. TP53 is assumed to be the most important tumour suppressor gene. Mutations in this gene have been shown to be associated with more than 50% of all cancers including breast cancer. Mutations in the p53 gene occur in both coding and non-coding sequences, however 90% of the polymorphisms occur in the noncoding sequences of the gene. This polymorphisms affect DNA-binding-domain and level of gene expression. Arg72Pro and PIN3(16bp duplication) polymorphisms are proposed to have an effective role in structural changes of p53. They have been inspected as a risk factor for breast cancer in different populations.

Objectives: The investigative aim of this study was to examine and define the role of two polymorphisms in p53 gene (changes in exon 4 and intron 3) and polymorphism in exon 7 of XRCC3 may be associated with an increased risk for breast cancer in the female patients from the Eastern Azerbijan of Iran.

Methods: 100 women with breast cancer and 100 healthy women without any background of cancer were included in the presnet study. Genotyping was performed by PCR_RFLP (polymerase chain reaction-restriction fragment length polymorphism) method employing the BstuI and NlaIII restriction enzymes following to DNA extraction. The results was subjected to statistical analysis.

Results: Statistical analysis of the results obtained in the present study did not show a significant correlation between polymorphisms of TP53, R72P (P =0.41) and PIN3, Ins16bp

(P = 0.13) and breast cancer in the population of East Azerbijan of Iran. No significant association were also found between XRCC3 gene polymorphism and breast cancer (P = 0.46).

Conclusion: Presence of P allele at codon 72 (rs1042522) and a doubling of 16bp in TP53 gene (rs17878362) and allele M (rs861539) in the gene XRCC3 can not be considered as a breast cancer risk factor in the population of East Azerbaijan of Iran.

KEY WORDS: Breast Cancer,Polymorphism, TP53, XRCC3