(CT/GA)n polymorphisms of GATA3 gene and risk of breast cancer

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Background and Aims: GATA3 is an enriched transcription factor in mammary epithelium. In present study we determined the allelic length of two short tandom repeats in 5'UTR and intron3 of GATA3 gene, in breast cancer patients and matched controls. We also evaluated risks for breast cancer associated with these STRs.

Methods: The GA expansion in 5'UTR and CT repeat in intron3 of GATA3 gene were amplified by PCR and alleles in variable sizes were selected by polyacrylamid gel and sequenced directly. These sequenced alleles were used as allele specific markers.

Results: In this research, we studied the GA and CT repeat polymorphisms in GATA3 gene among breast patients and healthy controls in Isfahan city of Iran. We also evaluated the relation of these STRs with breast cancer risk. Several different alleles of GATA3 for CT repeat were detected in both pateints and controls. However, there were significant differences in the distributions of alleles with 17 repeats for CT expansion between the patients and the controls. No polymorphism was observed for GA repeat.

Conclusions: These results suggest a significant association between CT repeat length in GATA3 gene and risk of breast cancer in Isfahan population of Iran. Identification of other polymorphic sites in GATA3 gene may be useful for identifying individuals predisposed to breast cancer.

Keywords: GATA3; CT/GA repeats; Breast cancer; Polymorphism