ORIGINAL ARTICLE

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Association between the Length of CA Dinucleotide Repeat in the PIK3CA gene with Risk of Breast Cancer

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Abstract

Introduction: The phosphatidylinositol 3-kinase (PI3K) signaling pathway regulates a variety of biological processes including proliferation, cell survival, apoptosis, motility, cell migration and insulin metabolism. ***The*** human p110α ***catalytic subunit*** is ***encoded*** by the phosphatidylinositol-3,4-bisphosphonate 3-kinase, catalytic subunit alpha (**PIK3CA*).*** To our knowledge this is the first report about polymorphic CA dinucleotide repeat of PIK3CA gene and its relation to cancer risk. In the present study we investigate the CA dinucleotide repeat polymorphism in the intron 1 of *PIK3CA* gene among breast cancer patients and healthy individuals and its relation to risk of breast cancer.

Method: we performed a case control study of 200 patients with breast cancer from Omid (Seyedoshohada) Cancer Hospital of Isfahan city and 200 healthy blood donors visiting hospitals for regular health checks. After DNA extraction from blood, CA dinucleotide region was amplified by PCR technique and the number of CA repeats was determined by polyacrylamide gel electrophoresis and direct sequencing.

Results: Five different length of CA repeat in the range of 10-14 and 10 allele combinations (genotypes) were observed among patients and controls. CA13 was the most common allele between patients and controls. Our findings demonstrate that women who carry one or two alleles shorter than 13 CA repeat are at significantly higher risk of developing breast cancer. Carrier of smallest allele (CA10) are at the greatest risk against breast cancer with an odds ratio of 4.26 (p = 0.006), in contrast, women with one or two alleles longer than 12 are at lower risk of breast cancer.

Conclusion: Our findings indicate a direct relationship between the numbers of repetitive sequences of PIK3CA gene intron 1 and increased risk of breast cancer.

Keywords: Breast cancer, PIK3CA, CA repeats polymorphism.