ORIGINAL ARTICLE

Iranian Quarterly Journal of Breast Disease 2014; 7(3).

Study of estrogen receptor-α A908G (K303R) mutation with breast cancer risk

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Abstract

Background: Genetic mutations in premalignant breast lesions may have a role in malignancy progression or influence the behavior of subsequent disease. A point mutation in estrogen receptor-α (ESR1) as A908G (Lys303→Arg) was originally involved to hypersensitive to estrogen breast hyperplasia. We detected this mutation among Iranian women with invasive breast cancer.

Methods: A population-based case-control study was conducted in 150 newly diagnosed invasive breast cancer and 147 healthy control individuals to screen for presence of the ESR1 A908G mutation by using single-strand conformation polymorphism (SSCP) analysis and 33Pcycle DNA sequencing.

Results: We detected the 10.7% ESR1 A908G mutation in the form of heterozygote genotype only among cancer patients (P=0.00). The allelic frequency of mutant allele AGG in codon 303 was significantly (P=0.001) higher in patients with the family history of breast cancer (28.9%) than those without the family history of breast cancer (1.9%).

Conclusion: Our data suggest that ESR1 codon 303 mutations are correlated with various aspects of breast cancer in Iran. ER-α genotype might represent a surrogate marker for predicting breast cancer developing later in life.

Keywords: Breast cancer, mutation, estrogen receptor, PCR-SSCP, lymph node metastasis.