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Presentation Abstract

Title: E-P12.12 - The ESR1 Gene rs1801132 variation and Breast Cancer risk in

Iran

Keywords: breast cancer; estrogen receptor-a; SNP

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Abstract: Abstract

Iranian breast cancer patients are relatively younger than their Western counterparts. Evidence suggests that alterations in estrogen signaling pathways, including ESR1 (estrogen receptor- α), occur during breast cancer development in Caucasians. Epidemiologic studies have revealed that age-incidence patterns of breast cancer in Asians differ from those in Caucasians. Genomic data for ESR1 in either population is therefore of value in the clinical setting for Iranian breast cancer.

A case-control study was conducted to establish a database of *ESR1* polymorphisms in Iranian women population in order to compare Western and Asian with Iranian (Asian-Caucasians) distributions and to evaluate *ESR1* polymorphism as an indicator of clinical outcome. DNA was extracted from Iranian women with breast cancer referred to Imam Khomeini Hospital Complex clinical breast cancer group (150 patients) and in healthy individuals (147 healthy control individuals). PCR single-strand conformation polymorphism technology was performed.

A site of silent single nucleotide polymorphism (SNP) rs1801132 was found, The frequency of allele 1 in codon 325 (CCC \rightarrow CCG) was significantly higher in breast cancer patients (39.6%) than in control individuals (28.9%; P = 0.007). The allele CCG had also significant association with the occurrence of lymph node metastasis.

Data suggest that *ESR1* polymorphisms in exon 4 codon 325 is correlated with various aspects of breast cancer in Iran. *ESR1* genotype, as determined during presurgical evaluation, might represent a surrogate marker for predicting breast cancer lymph node metastasis.

Keywords: breast cancer, estrogen receptor-α, SNP, PCR-SSCP. This research supported by Tehran University of Medical Sciences and Health Services grant numbers of 3054.

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