

Estrogen receptor- α gene, codon 594 (G3242A) polymorphism among Iranian women with breast cancer: a case control study

ABSTRACT

A case-control study was conducted to establish a database of ESR1 polymorphisms in Iranian population in order to compare Western and Iranian (Middle East) distributions and to evaluate ESR1 polymorphism as an indicator of clinical outcome. The ESR1 gene was scanned in Iranian patients newly diagnosed invasive breast tumors, (150 patients) and in healthy individuals (147 healthy control individuals). PCR single-strand conformation polymorphism technology and direct sequencing was performed. The silent single nucleotide polymorphism (SNPs) was found, as reported previously in other studies, but at significantly different frequencies. The frequency of genotype 01 in codon 594 (ACG-ACA), (G3242A), exon 8 was significantly higher in breast cancer patients (48.0%) than in control individuals (1.4%; $p = 0.001$). The allele 1 in codon 594 was significantly more common in breast cancer patients with age at menarche ≤ 12 (40.8%) than in those which their menstruation began at older than 12 years old (23.9%; $p = 0.002$). The allele 1 in codon 594 exhibited, the greater the frequency, the lesser the likelihood of LN metastasis. Present results demonstrated that this particular SNP marker may increase accuracy in predicting LN. Therefore, this SNP marker further increased predictive accuracy in Iranian population. These data suggest that ESR1 polymorphisms are correlated with various aspects of breast cancer in Iranian ESR1 genotype, as determined during pre-surgical evaluation, might represent a surrogate marker to increase predicting breast cancer in Iranian population.

Keyword: Breast cancer; Estrogen receptor- ; Lymph node metastases PCR-SSCP; Polymorphism