**عنوان مقاله:**

Molecular epidemiology of breast cancer among Iranian-Azeri Patients based on p53 research: cutting the Gordian knot

**محل انتشار:**

نهمین کنگره بین المللی سرطان پستان (سال: 1392)

تعداد صفحات اصل مقاله: 1

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**خلاصه مقاله:**

Background: There is no doubt that the future of cancer treatment relies on a further understanding about personal medical and population genomics data. In this study we tried to have a better understanding about molecular and epidemiological feature of breast cancer among the Azeri population with emphasis on detection of TP53 gene mutations and the role of p53codon 72 polymorphisms in susceptibility to breast cancer.Materials and Methods: Tumor and control samples were collected from 102 patients and 99 controls. TP53 gene mutations in exons 4-9 and adjacent intronic regions were detected by direct sequencing and/or single strand conformation polymorphism (SSCP). p53 codon 72 polymorphisms were identified by allele specific PCR amplification procedure. Pearson $\chi 2$ test was used and statistical significance level was set to < 0.05. Results: We found 19.6 % alterations at both exonic and intronic regions of TP53 gene. These mutations comprised of 18 single-base substitutions (11 transitions + 7 transversions), 1 deletion and 1 complex. Exon 6 was a highly mutable region in this study and we have detected 9 of all 20 (45%) observed mutations in this exon. Frequency of Arg/Arg at codon 72 was 30.3%, in controls and 49% in patients, the Arg / Pro frequency was 50.5% and 30.4%, Pro/Pro genotype was 19.2% in controls and 20.6% in patients. Distribution differences in the p53 codon 72 polymorphism between the cases and controls were statistically significant (p<0.05) We found that 24% of patients carrying Arg/Arg also had TP53 mutations in tumors compared with 14% of Pro/Pro carriers.Conclusions: The high proportion of mutations occurred in exon 6, a distinct pattern of TP53 mutation spectrum and differences in the hotspot distribution may reflect geographical and ethnic disparity in this study. Our results addressed to significant association between the codon 72 polymorphism (Arg/Arg genotype) and risk of breast cancer in Azeri population. Also our findings have a bias to previously reported results that the prevalence of cases with p53 mutations within Arg72-containing allele was higher than Pro72-containing allele, although it isn't statistical significance in our study. We hope that our team works can establish the first molecular database in this geographically and genetically distinct region.

**کلمات کلیدی:**

breast;cancer;p53;codon 72;mutation;Azeri patients