

## عنوان مقاله:

Association of CDH1 and TERT single nucleotide polymorphisms with susceptibility to familial breast cancer risk

## محل انتشار:

چهاردهمین کنگره بین المللی سرطان پستان (سال: 1397)

تعداد صفحات اصل مقاله: 1

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## خلاصه مقاله:

**Introduction:** Breast cancer (BC) is multistep disease that is thought to result from an interaction between genetic background and environmental factors. In Iran, one of the strongest risk factors for developing breast cancer is a positive family history of the disease. Recently, various polymorphisms of E-cadherin (CDH1) and TERT have been found to be associated with increased breast cancer risk worldwide. This study aimed to analyze the association of CDH1 and TERT single nucleotide polymorphisms with susceptibility to familial BC risk in the Iranian patients. **Materials and method:** One hundred and five patients with familial BC (FBC) and 110 non-familial BC (NFBC) were genotyped to elucidate the potential association between CDH1 rs5030625 polymorphism and TERT rs2736098 polymorphism by RFLP-PCR. Then, results were evaluated by electrophoresis and Epi Info(TM) 2012 software. **Results:** A significant association was found between CDH1 rs5030625 GAGA genotype and FBC risk. Compared with the control group, the FBC patients had a lower frequency of GG genotype (69% vs 85%) and a higher frequency of GAGA (5% vs 2%,  $P < 0.02$ ). Furthermore, the patients with FBC had a lower frequency of TERT rs2736098 GG genotype (38% vs 49%,  $P = 0.001$ ) and a higher frequency of rs2736098 AA genotype (12% vs 5%,  $P = 0.001$ ) compared with the NFBC. In other hand, The TERT rs2736098 GG genotype potentially increased the recurring risk of FBC ( $OR = 3.17$ ,  $P < 0.01$ ). Allele genotypic frequencies in the FBC patients differed from those of the controls. Interestingly, tumors in FBC patients with rs2736098 GG genotype and rs5030625 GAGA exhibited higher mitotic activity, higher grade, lower ER and PR than the other genotypes. In conclusion, CDH1 rs5030625 GAGA genotype and TERT rs2736098 GG genotype in combination with clinical parameters may be a prognostic factor rather than a susceptibility factor during the progression of FBC.

## کلمات کلیدی:

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