The Association Between Tri-nucleotide-repeat Containing 9 (TNRC9) /LOC643714 Genetic Variations And Breast Cancer In Egyptian Females

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Introduction and objectives:
Breast cancer is the most frequent cancer in women; over 1,000,000 new cases are diagnosed per year worldwide. Breast cancer is a complex polygenic disease in which genetic factors play a very important role in disease etiology and pathologic process. The TNRC9 / LOC643714 locus on chromosome 16q12 was one among the first breast cancer regions known through genome-wide association study. we aimed to check the association of TNRC9 rs3803662 and LOC643714 rs12922061 and risk of breast cancer in Egyptian females and to correlate them with clinico-pathological features and survival in patients to study their prognostic value.

Methods
This study included seventy-five subjects (fifty breast cancer patients and twenty-five healthy controls). The studied participants were subjected to full history taking, clinical examination, and laboratory investigations include tumor markers (CEA and CA15-3) and genotyping of TNRC9 (rs3803662) and LOC643714 (rs12922061) single nucleotide polymorphisms by using real time-PCR, for patients; pathological, clinical and survival data were analyzed.

Results
Significant higher frequencies of the LOC643714 rs12922061 CT genotype and T allele were observed in breast cancer patients (p=0.016, OR (95%CI); 4.114 (1.30-13.0 and p= 0.044, OR (95%CI); 1.02-5.39 respectively). This significant difference was also observed under a dominant model (CC vs CT+TT OR 3.551; 95%CI 1.26 – 10.02; p= 0.017) and Over-dominant model (CT vs CC+TT OR 3.692; 95%CI :1.19–11.39; p= 0.023). Regards rs12922061 genotype distribution, there was significant difference regards menopausal state under the dominant model (p=0.044) and there was significant difference regards nodal stage under both the dominant model (p=0.049) and over dominant model (p=0.028). No significant association was found between the rs3803662 polymorphism and breast cancer patients. Regarding survival after 2 years of follow up of breast cancer patients both LOC643714 (rs12922061) and TNRC9 (rs3803662) were not associated with significant survival difference C/C versus C/T +T/T and G/G versus A/A+ A/G respectively.

Conclusion
While there was no significant association regarding the TNRC9 rs3803662, our study found that rs12922061 of LOC643714 was related to breast cancer risk. Moreover, the rs12922061 is related to menopausal state and nodal stage.

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