

**STUDY OF THE Arg194Trp AND Arg399Gln POLYMORPHISMS IN THE  
*XRCC1* GENE IN THE POPULATION OF RIO DE JANEIRO AND THE GENETIC  
SUSCEPTIBILITY TO BREAST CANCER**

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The State of Rio de Janeiro is considered as the Brazilian state with the highest number of cases of breast cancer. In the present study we evaluated the alleles and genotypes frequencies of Arg194Trp (exon 6) and Arg399Gln (exon 10) polymorphisms, in the *XRCC1* gene, in the population of Rio de Janeiro (n=418). The results are in accordance to Hardy-Weinberg Equilibrium. Also, the distribution of these polymorphisms was analyzed in 104 women with family history of breast cancer in relation to controls, represented by 216 healthy women without family history of cancer. Interestingly we found a tendency that women with first-degree relatives with breast cancer present, preferentially, the Arg194Trp polymorphism ( $p=0.0538$ ; IC=1.039-3.487). In conclusion, although the difference between the polymorphism distribution in breast cancer family women and controls were not statistically significant, there is a tendency that women with first-degree relatives preferentially present the Arg194Trp polymorphism in the *XRCC1* gene. This finding points to the importance of analyzing the distribution of repair genes polymorphisms in the breast cancer families.

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Key words: breast cancer, family history, polymorphisms, *XRCC1* gene.