The investigation of DNA repair polymorphisms with histopathological characteristics and hormone receptors in a group of Brazilian women with breast cancer

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ABSTRACT. The association of tumor differentiation and estrogen receptor expression with the prognosis of breast cancer has been well established. Nevertheless, little is yet reported about the association of morphological characteristics of the tumor, estrogen receptor status and polymorphisms in low penetrance genes. The aim of the present study was to investigate a possible association between DNA repair gene polymorphisms (XRCC1, XPD, XRCC3, and RAD51) with histological type, grade and hormone receptor expression in a series of breast cancers. A cross-sectional study was carried out to evaluate 94 women with breast carcinoma, who had already
been selected and included in a study on the association of DNA repair gene polymorphisms. For immunohistochemistry, formalin-fixed, paraffin-embedded tissue samples from breast tumors were consecutively retrieved from the histopathology files of our institution. DNA obtained from blood samples of the same patients was investigated for the presence of the following polymorphisms: Arg-399Gln located in the XRCC1 gene; 135C/G located in the RAD51 gene; Lys751Gln located in the XPD gene and Thr241Met located in the XRCC3 gene. Polymorphisms were considered to be independent variables and hormone receptor expression and the morphological characteristics of the tumors comprised the dependent variables. No statistically significant association was found between gene polymorphisms and hormone receptor status. The association between XRCC1-Arg399Gln polymorphism and ductal carcinoma was statistically significant (P = 0.02). The association of the XPD-Lys751Gln polymorphism with histological grade was also statistically significant (P = 0.05). In conclusion, the XRCC1 genotype was found to be associated with ductal carcinoma histotypes and XPD genotype with low histological grade, which is the most frequent pattern of sporadic breast carcinomas.

**Key words:** Breast cancer; Estrogen; Polymorphisms; Hereditary disease