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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 (*XRCC*1, *XPD*, *XRCC*3, and *RAD*51) 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 *XRCC*1 gene; 135C/G located in the *RAD*51 gene; *Lys751Gln* located in the *XPD* gene and *Thr241Met* located in the *XRCC*3 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 *XRCC*1-*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 *XRCC*1 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

