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Genetic variation in ERCC1 and XPF genes and breast cancer risk

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ABSTRACT. Breast cancer is one of the most frequently diagnosed cancer in women worldwide, and we conducted a case-control study by genotyping seven potentially functional SNPs, three in ERCC1 and four in XPF, in a Chinese population of 417 breast cancer cases and 417 cancer-free controls. Three SNPs in ERCC1 and four SNPs in XPF were genotyped by using the Taqman Universal PCR Master Mix in the GeneAmp® PCR System 9700 with Dual 384-Well Sample Block Module, and assays were performed on a 384-well plate on the Sequenom MassARRAY platform. We found that elevated breast cancer risk was associated with those who had a family history of breast cancer and history of breast disease, and those who were over 25 years old at first full-term pregnancy. We found that decreased risk of breast cancer was associated with those who had a history of full-term pregnancies. Compared with the ERCC1 rs11615 T/T genotype, a

significantly higher risk of breast cancer was found in the C/C genotype in codominant and dominant models after adjusting for potential risk factors. Similarly, we found that ERCC1 rs3212986 C/C genotype was associated with an increased risk of breast cancer in codominant, dominant and recessive models. Our study indicated that the ERCC1 rs11615 and rs2298881 polymorphisms are associated with breast cancer in a Chinese population. Further studies with large sample size are greatly needed to elucidate the SNPs of ERCC1 and XPF genes in the development of breast cancer.

Key words: Excision repair cross-complimentary group 1; Susceptibility; Xeroderma pigmentosum complementation group F; Breast cancer; Polymorphisms

