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Validation of associations between *ESR1* variants and breast cancer risk in Chinese cohorts

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ABSTRACT. Estrogen receptor-α (ER) protein plays a key role in breast carcinogenesis, and common genetic variants in the corresponding gene locus have been associated with breast cancer risk in different populations. Here, we analyzed estrogen receptor 1 (*ESR1*) associations in two hospital-based studies of patients from the south of China. Three single-nucleotide polymorphisms (SNPs; rs3757318, rs2046210, and rs3734805) in *ESR1* were selected from previous genome-wide association study results and were genotyped using the Sequenom MassARRAY® iPLEX System in 845 breast cancer patients and 882 healthy controls. Association analysis based on unconditional logistic regression was carried out to determine the odds ratio (OR) and 95%

confidence interval (95%CI) for each SNP. Stratified analyses according to the status of ER and progesterone receptor (PR) were also performed. Of the three SNPs, rs3757318 did not pass the Hardy-Weinberg equilibrium test and was excluded from the subsequent analysis. The other two SNPs (rs2046210 and rs3734805) were strongly associated with susceptibility to breast cancer. Allele T of rs2046210 and allele C of rs3734805 were risk alleles and the adjusted ORs were 1.348 (95%CI = 1.172-1.550, P = 0.0001) and 1.319 (95%CI = 1.144-1.522, P = 0.0001), respectively. Furthermore, the risk allele of rs2046210 gave negative results for ER and PR expression in an immunohistochemical test, with ORs of 0.602 (95%CI = 0.384-0.944, P = 0.027) and 0.532 (95%CI = 0.338-0.837, P = 0.006), respectively. Our study further supports associations between rs2046210 and rs3734805 and breast cancer risk in Chinese women.

Key words: Breast cancer; Estrogen receptor 1; Susceptibility; Single-nucleotide polymorphism