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Association of *XRCC5* polymorphisms with COPD and COPD-related phenotypes in the Han Chinese population: a case-control cohort study

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ABSTRACT. Genome-wide association studies (GWAS) and integrative genomic approaches have demonstrated significant associations between chronic obstructive pulmonary disease (COPD) and polymorphisms of the X-ray repair cross-complementing protein 5 gene (*XRCC5*) in non-Asian populations. We investigated whether *XRCC5* polymorphisms might be associated with COPD susceptibility and COPD-related phenotypes in the Chinese Han population. Nine single nucleotide polymorphisms (SNPs) (rs3821104, rs12470053, rs207936, rs3770498, rs6704622, rs3770492, rs4674066, rs7573191, and rs207906) in the *XRCC5* gene were genotyped in a case-control study including 680 COPD patients and 687 controls. To estimate the strength of association, odds ratios (ORs) were calculated and the effects of potentially confounding variables were tested by logistic regression analysis. The association between haplotypes

and COPD outcome was also assessed. Our data identified that the SNP rs207936 was associated with COPD with an adjusted P value of 0.038, which was also found when analyzing only data of current smokers (P = 0.046). No significant associations were found between any of the SNPs and pulmonary function. Eight SNPs (rs3821104, rs12470053, rs207936, rs3770498, rs6704622, rs3770492, rs4674066, and rs7573191) showed strong linkage disequilibrium ($R^2 \geq 0.9$). Two major haplotypes were observed and showed a significant difference between case and control groups (P = 0.0054 and 0.0081, respectively). The present study showed that the *XRCC5* locus might be a contributor to COPD susceptibility in the Chinese Han population.

Key words: Association analysis; Chronic obstructive pulmonary disease; COPD-related phenotypes; Case-control study; *XRCC5*

