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Title: Association of FAM13A polymorphisms with COPD and COPD-related phenotypes in Han Chinese

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Body: Background: Genetic factors are known to contribute to COPD susceptibility and these factors are not fully understood. Genome-wide association studies (GWAS) and integrative genomics approaches have demonstrated significant associations between chronic obstructive pulmonary disease (COPD) and FAM13A polymorphisms in non-Asian populations. Objectives: To investigate whether FAM13A polymorphisms would be associated with COPD susceptibility and COPD-related phenotypes in Han Chinese. Methods: Seven single nucleotide polymorphisms (SNPs) in FAM13A gene were genotyped in a case-control study (680 COPD patients and 687 controls). Results: Statistical analysis revealed that SNP rs7671167 was associated with COPD in former smokers with adjusted P-value of 0.026. Five SNPs (rs7671167, rs2869966, rs2869967, rs2045517, and rs6830970) were associated with FEV1/FVC ratio in the entire cohort and rs6830970 was associated with FEV1/FVC ratio in COPD cases (P range 0.003-0.034). Six SNPs (rs7671167, rs2869966, rs2869967, rs2045517, rs1903003, rs6830970) showed strong linkage disequilibrium ($r^2 \geq 0.9$). Four major haplotypes were observed but showed no significant difference between case and control groups (P = 0.2356, 0.1273, 0.6266 and 0.3006 respectively). Conclusions: The current study suggests that the FAM13A locus might be a contributor to COPD susceptibility in Han Chinese.