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Genetic variants in FAM13A and IREB2 are associated with the susceptibility to COPD in a Chinese rural population: A case-control study - Jin Zhang - Ningxia Medical University

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Introduction & Objective: Genome-wide association studies identified several genomic regions associated with the risk of Chronic Obstructive Pulmonary Disease (COPD), including the 4q22 and 15q25 regions. These regions contain the FAM13A and IREB2 genes, which have been associated with COPD but data are lacking for Chinese patients. The objective of the study was to identify new genetic variants in the FAM13A and IREB2 associated with COPD in Northwestern China.

Method: This was a case-control study performed in the Ningxia Hui autonomous region between January 2014 and December 2016. Patients were grouped as COPD and controls based on FEV1/FVC, 70%. Seven tag Single-nucleotide Polymorphisms (SNPs) in the FAM13A and IREB2 genes were genotyped using the Agena MassARRAY platform. Logistic regression was used to determine the association between SNPs and COPD risk.

Results: rs17014601 in FAM13A was significantly associated with COPD in the additive (odds ratio [OR]=1.36, 95% confidence interval [CI]:1.11???1.67, P=0.003), heterozygote (OR=1.76, 95% CI:1.33???2.32, P=0.0001), and dominant (OR=1.67, 95% CI:1.28???2.18, P=0.0001) models. Stratified analyses indicated that the risk was higher in never smokers. rs16969858 in IREB2 was significantly associated with COPD but in the univariate analysis only and the multivariate analysis did not show any association.

Conclusion: The results suggest that the new variant rs17014601 in the FAM13A gene was significantly associated with COPD risk in a Chinese rural population. Additional studies are required to confirm the role of this variant in COPD development and progression.

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