Journal of Clinical Respiratory Open Access

Extended Abstract
Vol. 2, Iss. 1
2020

COPD 2019: Genetic variants in FAM13A and IREB2 are associated with the susceptibility to COPD in a Chinese rural population: A case-control study, Ningxia Medical University, China

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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.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 associationbetween SNPsand COPDrisk.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.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. Chronic obstructive pulmonary disease (COPD) could be a major explanation for morbidity and mortality worldwide. it's characterized by persistent respiratory symptoms and limitation of air flow.1 within the People's Republic of China, the prevalence of COPD in individuals of $ 40 years old is estimated at 8.2% 2 or varies from 5% to 13% in several provinces. / cities.3 Cigarette smoking is taken into account the foremost important risk factor, but genetic characteristics play a crucial role in susceptibility to COPD. Genome-wide association studies (GWAS) identified several genomic regions related to an increased risk of COPD. Some GWAS loci are found on the FAM13A gene on chromosome 4q22 and at the 15q25 locus, which has the IREB2 gene. Considered to be an indication transduction gene because of the RhoGAP functional domain within the exon region, 9 but it's now known to be related to signaling of □-catenin, which is generally activated during injury repair and tissue regeneration10, eleven. Hypoxia often accompanies COPD and improves FAM13A expression.9 Furthermore; Kim et al12 showed that FAM13A SNPs related to an increased risk of COPD were also related to an increased expression of FAM13A within the lungs, suggesting a possible causal association with pathological changes within the lung. Corvol et al13 showed that the association of the FAM13A gene with lung function parameters (FEV1% predicted and FEV1 / FVC) was observed in several independent cohorts, suggesting that FAM13A is related to a selected COPD phenotype. Furthermore, Choo ET al14 demonstrated an association between the CTGA diploma in FAM13A and therefore the emphysema phenotype of COPD, and Jiang et al11 provided the premise for the role of FAM13A within the development of emphysema. A recent study by Corvol ET al15 showed that FAM13A and therefore the epithelial-mesenchymal transition (EMT) of the airways are closely associated with fibrocystic disease of the pancreas. EMT is additionally believed to play a crucial role in airway remodeling in COPD.16 Taken together, these results suggest that FAM13A is involved within the etiology of lung disease and COPD, iron is found in smoke. cigarette smoking17 and has been shown to disrupt lung homeostasis, making lung tissues more liable to damage from any cause.18 IREB2 could be a gene that ends up in iron regulatory protein 2 (IRP2), which plays a task in key role in iron homeostasis. IREB2 is in an exceedingly strong linkage disequilibrium with the nicotine receptor genes (CHRNA3 and 5) .5 Expression of IREB2 increases within the lungs of COPD patients.19 IRP2 regulates cellular iron homeostasis and mitochondrial function.20, 21 Some variants of IREB2 are reported to affect COPD within the presence of high levels of iron because of exposure to cigarette smoke.18 Therefore, there can be some association between IREB2 and respiratory conditions like COPD.

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This abstract is partly presented at the 9th International Conference On COPD and Lungs on August 19-20, 2019 at Tokyo, Japan