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Title: Genome-wide association study identifies new COPD risk locus, PDE4D, in Korean population

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Body: Background. Chronic obstructive pulmonary disease (COPD) is a complex disease in which genetic background is a major risk factor. It is assumed that genetic factor is very important in its development. However the exact role of genetic abnormality in the pathogenesis of COPD remains unclear. Objectives: The present study was conducted to define the COPD related single nucleotide polymorphism (SNP) using GWAS in Korean population. Material and Methods. We performed GWAS by whole-genome SNP genotyping for 102 COPD cases and 544 normal controls using Affymetrix SNP 5.0 array. TaqMan analysis was employed for replication in different set. In subgroup analysis,patients were classified into four subgroups according to DLco and FEV1 data. Results: We identified that three SNPs satisfied the significance threshold of $P < 10\text{-}6$ (unadjusted). Among the top 3 significant SNPs, Second and third significant SNPs were in the promoter region of Phosphodiesterase-4D (PDE4D) gene [rs16878037 (C>T), $P = 4.63 \times 10\text{-}7$; rs16878035 (G>T), $P = 4.63 \times 10\text{-}7$]. Both SNPs are in the same haplotype block. The association of rs16878037 polymorphism was successfully replicated in the independent set ($P = 0.024$, OR= 3.479, 95% CI 1.105 - 10.953). CT and TT phenotypes were significantly more frequent in mild mixed and bronchitis dominant subgroup ($P=0.022$ and $P=0.045$). CT and TT genotype was significantly more frequent in DLco ≥ 50% group and minor allele frequency was also significantly frequent in same group ($P=0.016$) Conclusions. The results of this study suggested that PDE4D SNP were associated with the risk of COPD development especially chronic bronchitis phenotype in Korean population.