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Researchers identify gene variant associated with chronic obstructive pulmonary disease

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Researchers from Boston University School of Medicine (BUSM) have, for the first time, identified a gene variant on chromosome 4 that may be a potential risk factor for chronic obstructive pulmonary disease (COPD). These findings will be published in *PLoS Genetics* on March 20th.

COPD is the fourth leading cause of death in the United States and one of the most prevalent disabling diseases of adults. According to the researchers, cigarette smoking is the primary risk factor for impaired lung function, yet only 20 percent of smokers develop COPD. This observation, along with family studies of lung function and COPD, suggests that genetic factors influence susceptibility to cigarette smoke.

The researchers performed a genome-wide association study on 7,691 Framingham Heart Study participants to identify a relationship between common genetic variants and measures of lung function. The identified variants on chromosome 4 were then examined and confirmed in an independent set of 835 Family Heart Study participants.

"Several interesting genes are present in the region that we identified, including a gene (HHIP) interacting with a biological pathway involved in lung development, but it is not yet clear which gene in the region explains the association," said lead author Jemma Wilk, D.Sc., an assistant professor of neurology at BUSM. "Our results identified a region of chromosome 4 that warrants further study to understand the genetic effects influencing lung function," she added.

Source: Boston University