

Genetic variant may control lung function and risk of COPD

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Researchers have discovered evidence that suggests a genetic variant may be associated with better preserved lung function among children with asthma and adults who smoke, according to a new study funded by the National Heart, Lung, and Blood Institute (NHLBI), which is part of the National Institutes of Health. The study also found an association between the genetic variant and a lowered risk of developing chronic obstructive pulmonary disease (COPD) in adults who smoke. COPD is a lung disease most common among smokers, which makes it difficult to breathe.

The study is published online by The [New England Journal of Medicine](#) on Wednesday, Dec. 16. The team of researchers found that a DNA single nucleotide polymorphism, or SNP, was associated with better preserved lung function among children with asthma and in former or current smokers. The study also found a lower risk for developing COPD in adults who smoke. A SNP is a single base pair in a person's DNA which often varies among individuals. Adult patients with this SNP had a 35 percent reduction in the risk of onset of COPD.

Researchers examined the genes and the breathing capacity of more than 8,300 child and adult participants from seven different studies, including the NHLBI-funded [Childhood Asthma](#) Management Program and National Emphysema Treatment Trial. Using data from all seven studies, researchers found a link between a SNP in MMP12, a gene that encodes matrix metalloproteinase 12, and better preserved lung function among both children with asthma and adults who are former or current smokers.

The same SNP was also found to be linked to a reduced risk of developing COPD. MMP12 is produced by inflammatory cells called macrophages, which are found in the lung.

Results from the study support the theory that asthma and COPD may share some common mechanisms, even though the two diseases affect patients differently.

[Asthma](#) is one of the most common chronic conditions in the United States and affects more than 22 million people of all ages. COPD affects primarily older individuals and is the fourth leading cause of death in the United States. The number of people who have COPD is on the rise — more than 12 million are currently diagnosed with it, and it is estimated that another 12 million may have undiagnosed COPD.

Provided by NIH/National Heart, Lung and Blood Institute

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