

Gene variant linked with development of COPD in men

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Researchers have linked a variant in the vitamin D receptor gene (VDR) with the onset of chronic obstructive pulmonary disease (COPD) in Caucasian men. The study population consisted of participants in the Veterans Administration Normative Aging Study, a multidisciplinary study of aging that began in 1963.

The VDR study will be presented at the ATS 2011 International Conference.

"Our results show that this gene variant is associated with development of COPD in [Caucasian men](#), and provides support for the notion that vitamin D [metabolic pathways](#) may affect COPD risk," said Audrey Poon, PhD, postdoctoral fellow at Meakins-Christie Laboratories, McGill University Health Centre in Montreal. The study was conducted while Dr. Poon was doing her first postdoctoral fellowship at Channing Laboratory, Brigham and Women's Hospital in Boston.

Although cigarette smoking is considered to be the main risk factor for chronic obstructive diseases such as emphysema and [chronic bronchitis](#), only a proportion of smokers develop clinical disease. Researchers believe genetic factors also contribute to the risk of developing COPD. The vitamin D [metabolic pathway](#) has been implicated in the development of COPD.

"Several variants of genes that control vitamin D function and metabolism have been associated with COPD and other [lung diseases](#),

but results have been conflicting," Dr. Poon said. "In this study we investigated variants in two vitamin D pathway genes and their association with development of COPD."

Using DNA data from the VA study, the researchers determined the genotypes of 24 variants in the vitamin D receptor gene and 12 in the vitamin D binding [protein gene](#) in a total of 1,215 men. All subjects were free of known chronic conditions, including [coronary heart disease](#), hypertension, [chronic lung disease](#), asthma and diabetes at the time of recruitment. The VA study also offered data from repeated lung function measures conducted over 40 years, as well as smoking information for the participants.

The researchers used the lung function data to measure the time it took for participants to develop COPD, evaluating all 36 gene variants. They found variant rs3847987 of the VDR gene was found to influence the time to onset of COPD in the study population.

"We had the expectation that we would find an association of variants in one of these genes with the development of COPD," Dr. Poon said.

"However, we did not expect that this particular variant in the VDR gene would be associated, since it has not been reported to be associated with COPD before."

Future studies will need to clearly determine the function of the gene variant, she added.

"More questions need to be answered before we can take any of these findings to clinical practice," Dr. Poon said. "For instance, we do not know what effect, if any, vitamin D levels would have on the risk of developing COPD and whether circulating vitamin D levels interact with genetic variants."

"Furthermore, we only selected two genes in the pathway, and there are numerous genes that are involved," she added. "If these findings are validated, then investigating the effect of this particular variant in the function of the vitamin D receptor will be important."

Provided by American Thoracic Society

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