Researchers find genetic risk factor for pulmonary fibrosis

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A paper recently published in the *New England Journal of Medicine* and co-written by physicians and scientists at the University of Colorado School of Medicine finds that an important genetic risk factor for pulmonary fibrosis can be used to identify individuals at risk for this deadly lung disease.

Researchers looked at a fairly common variant of the gene for mucin-5B, a protein that is a component of the mucous produced by the bronchial tubes. While this variant of the MUC5B gene is fairly common, pulmonary fibrosis is an uncommonly reported disease.

In a review of CT scans of more than 2,600 adults who did not have a clinical diagnosis of pulmonary fibrosis, researchers found imaging evidence of lung inflammation and scarring in about 9 percent of those over age 50. In this age group, these abnormal findings on CT scans were significantly more common among the 21 percent people with the MUC5B genetic variant.

Importantly, definite lung fibrosis seen on CT scan was strongly associated with the MUC5B genetic variant. While these abnormalities do not necessarily indicate a disease that will progress, the presence of these abnormalities were associated with more shortness of breath and cough as well as smaller lung sizes and ability to transfer oxygen.

The findings suggest that pulmonary fibrosis, which is a condition where lung tissue becomes thickened, stiff and scarred, may be a part of a

much more common, but likely less severe, syndrome and could potentially be predicted on the basis of the MUC5B genetic variant.

David A. Schwartz, MD, chairman of the School of Medicine's Department of Medicine, was a corresponding and senior author of the paper. Gary M. Hunninghake, MD, MPH, from the pulmonary and critical care division of the Department of Medicine at Brigham and Women's Hospital in Boston was a corresponding and first author of the paper. The research is supported by grants from the National Institutes of Health, the National Heart, Lung and Blood Institute and the U.S. Veterans Administration.

Twenty-one authors shared credit for the paper, including researchers from Brigham and Women's Hospital and Boston University. Joining Schwartz from Colorado were Elissa Murphy, MS, and Marvin I. Schwarz, MD, from the School of Medicine and Tasha E. Fingerlin, PhD, from the Colorado School of Public Health.

David Schwartz this month gave the American Thoracic Society's Amberson Lecture, an honor bestowed annually on an individual with a career of major lifetime contributions to clinical or basic pulmonary research and/or clinical practice.

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