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Genetic variation contributes to pulmonary fibrosis risk

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A newly published study of patients with pulmonary fibrosis has discovered multiple genetic variations that should help with future efforts to treat the disease.

Pulmonary fibrosis is a condition where lung tissue becomes thickened, stiff and scarred. Currently in the United States, there are no drugs approved for use in cases of the condition's most common and severe form, which is known as idiopathic pulmonary fibrosis (IPF) because the cause of the disease is not known. In those cases, the median survival time after diagnosis is two to three years and lung transplants are the only intervention known to prolong life.

This new study found evidence that common genetic variation is an important contributor to the risk of developing IPF, accounting for approximately one-third of the risk of developing disease. The study identified seven novel genetic risk loci that include genes involved in host defense, cell-cell adhesion, and DNA repair. These findings suggest that the disease is primarily initiated by defects in the lung's ability to defend against internal and environmental challenges.

This international collaborative research was led by scientists at the University of Colorado.

"The insightful leadership of Tasha Fingerlin, extraordinary contributions of Elissa Murphy, and active participation of many others ensured the success of this research and, in aggregate, we have

established the scientific basis for early recognition and have identified novel therapeutic targets for this untreatable disease," says David A. Schwartz, MD, chairman of the Department of Medicine at the University of Colorado School of Medicine and senior author of the study. "These findings will change the way we think about pulmonary fibrosis and should eventually enhance the diagnostic and therapeutic options for our patients."

Fingerlin, PhD, and Murphy, MS, also authors of the study, are researchers at the Colorado School of Public Health and the CU School of Medicine.

The study, published in the April 14 edition of the journal *Nature Genetics*, is the first study to map out genes associated with IPF risk on a genome-wide scale. Three previously known genetic links were confirmed and seven novel loci were identified by studying the entire genome in this progressive incurable disease.

The work was supported by the National Heart, Lung, and Blood Institute (NHLBI). "In addition to expanding the library of genetic changes that can underlie pulmonary fibrosis, this study's findings demonstrate that both rare and common genetic variants contribute significantly to pulmonary fibrosis risk," says James Kiley, PhD, Director of NHLBI's Division of Lung Diseases. "A key next step for research is figuring out how these genetic variants work with environmental factors in the development of the disease."

Provided by University of Colorado Denver