New Genetic Discoveries in Lung Health Offer Hope for Better Prevention and Treatment for Lung Diseases

CHARLOTTESVILLE, Va., September 26, 2011 – An international group of scientists, including University of Virginia researchers, have discovered 16 new sections of the genetic code that relate to lung health -- opening up the possibility for better prevention and treatment for lung diseases.

An international consortium of 175 scientists from 126 centers in Europe, the United States and Australia identified genetic variants associated with the health of the human lung. Their discovery sheds new light on the molecular basis of lung diseases like Chronic Obstructive Pulmonary Disease (COPD).

“COPD is the fourth most common cause of death worldwide; it is a progressive disease that makes it increasingly hard for people to breathe,” says Steven Rich, PhD, director of the University of Virginia Center for Public Health Genomics and a major contributor to the new study. “Although smoking is the most important risk for developing COPD, not all smokers are equally likely to develop COPD. We believe that these differences in risk are due, in part, to the genetic variants people carry.”

This research, published online September, 25, 2011 in *Nature Genetics*, marks the first time these 16 common genetic variants have been definitively linked with lung function. Researchers say these newly discovered pathways could be prime targets for medications.

Rich, the Harrison Professor of Public Health Sciences at the UVA Health System, provided a wealth of data from participants of the MESA (Multi-Ethnic Study of Atherosclerosis) cohort, supported by a contract from the National Heart, Lung and Blood Institute (NHLBI) of the National Institutes of Health (NIH).

The pioneering study was led by Professor Martin Tobin from the University of Leicester, Professor Ian Hall from the University of Nottingham, Dr. Stephanie London from the National Institute of Environmental Health Sciences, and Dr. Stephen Rich of the University of Virginia School of Medicine.

The genetic study involved 2.5 million genetic variants in each of 48,201 people across the world. A smaller number of the most promising variants were then studied in a further 46,411 individuals. The research was funded by the UK Medical Research Council (MRC), the Wellcome Trust, and the NIH.

These recent discoveries build on research published by the same authors last year, bringing the total number of genetic variants associated with lung function to 26. The same authors also showed, in research published in the *American Journal of Respiratory and Critical Care Medicine* in June 2011, that variants predicting lung function also predict the disease, COPD.

“This study identifies the parts of the human genome that contain genes that have genetic variants that are common in the population,” explains Rich. “Studies such as this one, and others that our group at the University of Virginia are conducting in lung disease, may help us understand the biologic mechanisms and inform improved patient care. However, until we identify the genes, their disease-causing variants and the pathways that we can target for therapy, the best way to prevent COPD is to stop smoking.”

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UVA Health System is an academic health system that includes a 589-bed Magnet® hospital, the UVA School of Medicine, a level I trauma center, nationally recognized cancer and heart centers and primary and specialty clinics throughout Central Virginia. UVA is recognized for excellence by *U.S. News & World Report*, *Best Doctors in America*, *America’s Top Doctors* and Thomson Reuters’ *Top 50 Hospitals for Inpatient Cardiovascular Care*. 