MedicalXpress

Lung disease may be genetic, despite lack of family history

May 17 2010

Patients who encounter serious lung diseases in middle age, despite an absence of family history or other predisposing factors, may still have their genes to blame, according to a new study conducted by researchers at the National Institute of Allergy and Infectious Diseases, part of the National Institutes of Health.

The study also determined that the use of a simple screening test may help identify those genetic abnormalities and allow detection before the onset of disease.

"Earlier reports have indicated a correlation between certain nontuberculous mycobacterial respiratory infections and specific gene abnormalities," noted lead author Kenneth Olivier, M.D., M.P.H., NIAID staff clinician. "The results of this study confirm the correlation, and indicate the usefulness of simple testing in identifying these abnormalities, especially in an older population."

The results will be reported at the ATS 2010 International Conference in New Orleans.

The study of 32 patients focused on the movement of cilia, the tiny hair-like filaments that line the respiratory tract. In normal respiration, cilia help move dust and other fine particles out of the airway, keeping passages clear of harmful bacteria and other pathogens. When cilia are altered as the result of genetic mutation, they may fail to function properly, allowing disease-causing bacteria to build up.

"Genetic abnormalities in this clearance mechanism may predispose some older individuals to the development of certain pulmonary diseases, even though those individuals do not appear to have any predisposing factors," Dr. Olivier said.

Physicians can determine whether patients are at risk of the cilia not functioning correctly through a simple, noninvasive test that measures the levels of nitric oxide produced in the nose and sinuses. In patients who are predisposed to nontuberculous lung diseases, Dr. Olivier noted nitric oxide levels significantly lower than those of healthy individuals.

"Measurement of nasal nitric oxide production is easily performed, and can be an effective and noninvasive screen for identifying patients who may have abnormal ciliary function," he said. However, the screening test may also be abnormal for some patients with cystic fibrosis or acute viral respiratory infections, he added.

In patients with no known risk factors for lung disease, identifying which individuals are most likely to develop illness has posed a dilemma for physicians. Dr. Olivier said the results of this study may help researchers determine additional genetic abnormalities that could cause lung disease to develop, and may even lead to effective treatments.

"These results may lead to better understanding of predisposing genetic factors that will allow identification of at-risk individuals before the typical middle-age disease onset," he noted. "It may also allow development of preventive strategies or therapeutic interventions aimed at correcting airway clearance deficiencies."

"The next step is to focus on the identification of mutations in genes associated with ciliary dysfunction, to search for as yet unidentified novel cilia genes, and to utilize evolving technologies to better characterize genetic risks in patients with these diseases," he added.