Genetic Ancestry Data Improve Diagnosis in Asthma and Lung Disease

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Denver, CO — Americans with lung disease may face a far greater level of lung damage than either they or their doctor suspect, depending on their individual genetic heritage, according to research published online July 7 in the *New England Journal of Medicine.* The research implications range from diagnosing the severity of asthma, to disability decisions or eligibility for lung transplants, researchers say.

In the largest study of its kind to date, which pooled data on more than 3,000 patients, researchers at National Jewish Health and several other institutions found that patients' precise genetic background told far more about their potential lung function - and therefore any damage that has occurred - than the self-identified racial profile commonly used in such tests. The results point to a more precise method of assessing patients' lung function, as well as the potential impact of using precise genetic benchmarks for assessing health overall.

"Standard racial classifications do not capture the mixed genetic heritage of most populations," said Dr. Max A. Seibold, a Research Instructor at the Center for Genes, Environment and Health at National Jewish Health and one of four lead authors on the paper. "Our findings show that accounting for genetic ancestry improves prediction of normal lung function over a simple race-based classification. This improvement in lung function prediction, using genetic ancestry, may result in reclassification of severity in many lung diseases, similar to what we observed for asthma in this study. We feel these findings bring us closer to personalized medicine."

Lung function is one of many medical assessments that use standard benchmarks, such as age, sex and race, to determine the normal expected range for an individual patient. Similar criteria also are used in assessing kidney function or the risk for some cancers. In patients with lung disease, those benchmarks help physicians assess the severity of damage represented by a patient's lung function test and are often used to determine whether patients have severe asthma, or whether they may be eligible for disability insurance or a lung transplant.

The study used recently developed genetic tools to estimate individual genetic ancestry, and found a significant association between ancestry and lung function. Advances in technology have reduced the cost of those tools significantly, according to the researchers. As a result, this could be a viable method of dramatically improving patient care at relatively low cost.

The researchers used five large-scale, independent health studies with self-identified African American populations, ranging in age from 18 to 93 years, to examine the impact of genetic ancestry on measures of lung function.

The team found a significant link between African ancestry and pulmonary measurement in both men and women across all ages.

The paper had four co-first authors. In addition to Dr Seibold, they include Rajesh Kumar, MD, of Children's Memorial Hospital and Northwestern University School of Medicine, Chicago; Melinda Aldrich, PhD, MPH, with the University of California San Francisco; and L. Keoki Williams, MD, MPH, with the Center for Health Services Research and the Henry Ford Health System, Detroit. A complete list of authors, their affiliations and disclosures are available with the article, at NEJM.org.

National Jewish Health is known worldwide for treatment of patients with respiratory, cardiac, immune and related disorders, and for groundbreaking medical research. Founded in 1899 as a nonprofit hospital, National Jewish remains the only facility in the world dedicated exclusively to these disorders. Since 1998, *U.S. News & World Report* has ranked National Jewish the #1 respiratory hospital in the nation.

National Jewish Health is the leading respiratory hospital in the nation. Founded 120 years ago as a nonprofit hospital, National Jewish Health today is the only facility in the world dedicated exclusively to groundbreaking medical research and treatment of patients with respiratory, cardiac, immune and related disorders. Patients and families come to National Jewish Health from around the world to receive cutting-edge, comprehensive, coordinated care. To learn more, visit the media resources page.

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