

Genetic ancestry data improve diagnosis in asthma and lung disease

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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 a study to be released July 7. 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, spanning a dozen research centers and pooling data on more than 3,000 patients, a team of researchers led by UCSF and Northwestern University 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, researchers say. Findings will appear in the July 22 print edition of the "[New England Journal of Medicine](#)" and online on July 7 at [nejm.org](#).

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.

Standard race categories, however, don't capture the extent of our ancestral diversity, according to the paper's senior author, Esteban G. Burchard, MD, MPH, who is director of the UCSF Center for

Genes, Environment and Health, and a member of the Department of Bioengineering and Therapeutic Sciences, a joint department between the UCSF schools of medicine and pharmacy.

"People throughout the world have a richer genetic heritage than can be captured by our current definitions of race," Burchard said, noting that almost every continent has large populations that are known to be genetically mixed. "When we force patients into an individual box, such as 'African-American' or 'Caucasian', we're missing a lot of genetic information."

While this study focused on patients who define themselves as African-Americans, the participants' actual [genetic ancestry](#) ranged broadly and included Caucasian and African heritage.

"Since genetic ancestry improves our definition of normal lung function, it may be relevant for determining the severity of all lung diseases, as it was for asthma in this study," adds Rajesh Kumar, MD, an attending physician at Children's Memorial Hospital, associate professor at Northwestern University Feinberg School of Medicine and the lead and corresponding author on the paper.

"Taking genetic ancestry into account could result in more appropriate treatment for patients."

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.

"This study provides new evidence that genetic ancestry correlates to physiologic measures," added Burchard. "With it, we're one step closer to personalized medicine."

The authors acknowledge some important limitations with the study. For example, the association between [lung function](#) and ancestry could be influenced by factors other than genetics, such as social and environmental exposures.

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