

# Asthma treatment may be headed toward personalized medicine

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Asthma patients could clearly benefit from personalized medicine, a new study suggests. However, the new discovery of a key gene, while exciting, does not mean that day is here quite yet.

Asthma treatment response depends on whether [patients](#) have two copies of a [common genetic variation](#), researchers reported in the September 26 online edition of the [New England Journal of Medicine](#) (*NEJM*).

The discovery raises hopes that [personalized medicine](#) and [genetic testing](#) might one day be used to improve [patient outcomes](#) for asthma and other [respiratory conditions](#) treated with the same class of drugs.

“This is a newly identified genetic variation, and the finding needs to be replicated in larger studies,” says UCSF’s Esteban Burchard, MD, MPH, who also studies genes in asthma. “But it could lead to a change in how we manage severe asthma.”

One-in-six study participants had two copies of a variation in the gene called GLCCI1. Their lung function was less than half as likely to improve to a clinically significant degree in response to inhaled corticosteroid treatment.

About 30 million people in the United States have had asthma or currently have asthma, according to the US Centers for Disease Control and Prevention. The disease is responsible for about 17 million trips to the doctor each year, including nearly one-half million hospital stays.

For decades, first-line treatment for all but the mildest cases has included an inhaler, which patients use to deliver corticosteroid medicine into the lungs' airways.

The medicines have side effects, especially for children, so physicians often initially prescribe a lower dose and increase dosage if the patient's asthma does not improve. But one-third or more of patients do not respond to treatment with any standard dosage of an inhaled corticosteroid.

“It can be a long period of wasted time,” Burchard says. “If we knew that you had severe asthma and we knew which genetic mutations were associated with differences in [drug](#) response, then we could test you, and we would then know and be able to discuss which drugs were likely to work or not work for you.”

The researchers, led by Harvard Medical School scientists, surveyed the entire genomes of pediatric [asthma patients](#) and their parents, using a method called a genome-wide association study (GWAS). Enlisting parents of asthmatics, including many who also had been affected by asthma, allowed researchers to use fewer study participants to home in on genetic variants likely to affect [treatment response](#).

The scientists discovered the gene in blood from participants in a study called the Childhood Asthma Management Program. They then found a significant association between the genetic variant and treatment responses in patients from three out of four additional small studies examined.

In addition, the scientists studied the affect of the genetic mutation on cells grown in the lab and showed that the cells' behavior was consistent with what one might expect to observe in patients with a diminished response to corticosteroid treatment.

The NEJM study relied on clinical records for data collected over years on a standard measurement known as forced expiratory volume, in which patients inhale as much air as they can and then exhale as much as they can, as quickly as they can.

The study included fewer than 1,000 individuals, small by GWAS standards. “They have small samples sizes in all five of the cohorts they studied,” Burchard says. “The result is encouraging, but I think it needs to be replicated in a larger, prospective study in which treatment is assigned at the beginning of the study.”

The researchers analyzed data for white participants, and the findings cannot be assumed to apply to other ethnic groups. Burchard searches for asthma risk genes using similar GWAS methods, and recently was part of a research team that identified an asthma risk gene that appears unique to populations of African descent.

Although the findings require confirmation before they merit broader application, “This is probably one of the more significant publications in a while for the pharmacogenetics of [asthma](#),” Burchard says.

**More information:** [www.nejm.org/doi/full/10.1056/NEJMoa0911353](http://www.nejm.org/doi/full/10.1056/NEJMoa0911353)

Provided by University of California, San Francisco

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