

Environmental associations with genes may yield opportunities for precision medicine

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Chris Gignoux, Ph.D., University of Colorado Anschutz Medical Campus. Credit: Dr. Gignoux

A new approach to genetic analysis finds associations between environmental factors and pharmacogenes—genes associated with a person's response to drugs—sparking ideas for new research at the interface of population genetics and medicine. Findings were presented at the American Society of Human Genetics (ASHG) 2018 Annual Meeting in San Diego, Calif.

"Humans have developed and used pharmaceutical drugs for a few centuries, but their genes have been functioning on their own and interacting with other environmental factors for long before that," explained presenting author Chris Gignoux, Ph.D., of the University of Colorado Anschutz Medical Campus. Like changes in the physical environment, drugs affect the micro-environment within the body, which alters the way its cells and genes function. This suggests that genes with pharmacogenomic relevance may also

be useful in studying broader correlations between genetics and environment.

To explore a variety of environmental factors, Dr. Gignoux collaborated with Elena Sorokin, Ph.D., of Stanford University, who created a geocoded resource of over 20 climate, geographic, and ecological variables, using data from NASA, the World Wildlife Fund, and other sources. With collaborators from across the United States, they examined samples from the Population Architecture using Genomics and Epidemiology (PAGE) Study, a large initiative to highlight the utility of studying clinically and epidemiologically relevant variation in 51,698 individuals from 99 global populations. In a new type of analysis they termed an Enviro-WAS (environmental-wide association study), the researchers examined 19,690 pharmacogenomically-relevant variants to identify associations between genotypes and the 20 [environmental variables](#).

Dr. Sorokin described the reasons behind building the geocoded resource and applying it to PAGE. "This work was inspired by previous research on human adaptation, coupled with our interest in performing large-scale association studies across diverse populations," she said. "We can use our rich environmental database to identify correlations with genetic variants from globally representative populations in the PAGE Study."

The Enviro-WAS identified novel associations between certain genetic variants and ecological zones and replicated previously known associations between certain variants and environmental variables such as latitude and altitude. The geocoded resource of environmental factor data will be made available for other researchers to use and collaborate on, in hopes of promoting and refining the Enviro-WAS approach and triggering further investigation into environmental factors and genomics.

"We believe these associations may be of interest to both evolutionary and medical geneticists, since this method can show how genes have reacted to environments throughout history," said Dr. Gignoux.

In addition, studying a diverse, global population allows for further examination of a variety of [environmental factors](#) and adds to information missing in pharmacogenomics. Such research can help to address unequal representation in clinical genomics.

"Pharmacogenomics data currently have a heavy bias towards people of European descent, and there's a lack of knowledge about diverse populations," said Dr. Gignoux. "This sampling gives us more diversity and could lead to better targeted treatments for subgroups worldwide."

Provided by American Society of Human Genetics

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