

Scientists scour genes of over 53,000 people to better battle dangerous diseases

5 April 2021



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A new analysis of the entire genetic makeup of more than 53,000 people offers a bonanza of valuable insights into heart, lung, blood and sleep disorders, paving the way for new and better ways to treat and prevent some of the most common causes of disability and death.

The analysis from the Trans-Omics for Precision Medicine (TOPMed) program examines the complete genomes of 53,831 people of diverse backgrounds on different continents. Most are from [minority groups](#), which have been historically underrepresented in [genetic studies](#). The increased representation should translate into better understanding of how heart, lung, blood and sleep disorders affect minorities and should help reduce longstanding health disparities.

"The Human Genome Project has generated a lot of promises and opportunities for applying genomics to [precision medicine](#), and the TOPMed program is a major step in this direction," said Stephen S. Rich, Ph.D., a genetics researcher at the University of Virginia School of Medicine who helped lead the project. "An important feature of

TOPMed is not only publishing the genomic data on 53,000 people with massive amounts of data related to heart, lung, blood and sleep disorders but also the great diversity of the participants who donated their blood and data."

Historic Genome Analysis

The groundbreaking work identified 400 million genetic variants, of which more than 78% had never been described. Nearly 97% were extremely rare, occurring in less than 1% of people. This sheds light on both how genes mutate and on human evolution itself, the researchers say.

Of the groups studied, people of African descent had the greatest genetic variability, the researchers found. The resulting data is the best ever produced on people of African ancestry, the scientists report in the prestigious journal *Nature*.

The work also offers important new insights into certain gene variants that can reduce people's ability to benefit from prescription drugs. This can vary by race and ethnic group.

"TOPMed is an important and historic effort to include under-represented minority participants in genetic studies," said Rich, who served on the project's Executive Committee and chaired the Steering Committee. "The work of TOPMed should translate not only into better [scientific knowledge](#) but increase diversity at all levels—scientists, trainees, participants—in work to extend personalized medicine for everyone."

Rich was joined in the effort by UVA's Ani Manichaikul, Ph.D.; Joe Mychaleckyj, DPhil; and Aakrosh Ratan, Ph.D. All four are part of both the Center for Public Health Genomics and UVA's Department of Public Health Sciences.

More information: et al, Sequencing of 53,831 diverse genomes from the NHLBI TOPMed

Program, *Nature* (2021). DOI:
[10.1038/s41586-021-03205-y](https://doi.org/10.1038/s41586-021-03205-y)

Provided by University of Virginia

APA citation: Scientists scour genes of over 53,000 people to better battle dangerous diseases (2021, April 5) retrieved 12 July 2022 from <https://medicalxpress.com/news/2021-04-scientists-scour-genes-people-dangerous.html>

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