ASHG
American Society of Human Genetics

Diversity in Genetics and Genomics Studies

DIVERSITY IN GENETICS AND GENOMICS: AN EQUITY IMPERATIVE

It is imperative for participation in genetics and genomics research to reflect humanity's diversity so that all people can enjoy its benefits. This will take a concerted effort by the research community and research funders and must include greater engagement with individuals and communities underrepresented in research.

What is genetic diversity?

Each person's genome – the combined set of all their genes and other DNA – is unique. While we, as human beings, are 99.9 percent identical in our genetic makeup, there is variation in the remaining 0.1%. This variation in the human genome is why, for instance, height and dimples vary between people. Some genetic variants influence one's risk of disease or how the body responds to medications, stress, or environmental factors. The number and frequency of variants is known to differ across populations. It is critical for scientists to fully understand this variation in the human genome to ensure that clinical and societal advances resulting from research benefit all.

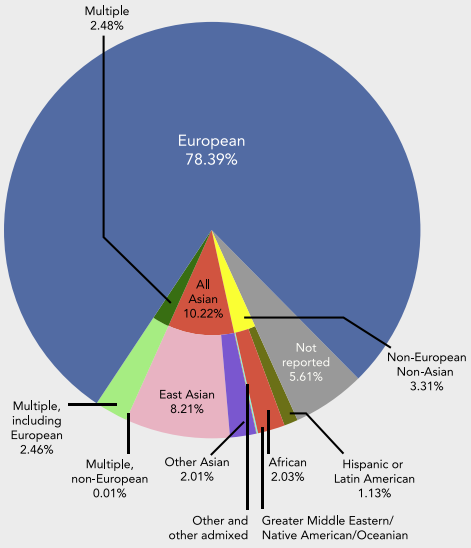
Improving Healthcare for All

Research with diverse populations greatly enhances our understanding of variation in the genome, yet most participants recruited into genetics and genomics research studies are of European ancestry.[1]

As genetics research is foundational to our understanding of human biology, gleaning the full scope of genetic variation will improve both healthcare and health equity. Inclusion of populations from diverse ancestries in studies is revealing novel insights about drug responses, diagnostic accuracy, and disease risk, demonstrating the need for increased diversity in research studies.[2]

- ***Improving diagnoses:*** A genetic variant discovered by sampling largely European ancestry groups was initially believed to increase one's risk for hypertrophic cardiomyopathy. However, the variant was subsequently found to be common in African ancestry populations and unlikely to be a risk factor for the disease.[3]
- ***Understanding differences in drug response:*** Asthma is the most common chronic childhood disease. In a study with participants from Latino ethnic groups and African Americans, researchers discovered genetic variants that may help explain the different responses to albuterol asthma inhalers in people of all ancestries.[4]
- ***Developing new drugs:*** In a study including African American research participants, researchers discovered a genetic variant associated with low cholesterol levels in a gene called PCSK9. This helped lead to the development of PCSK9 inhibitors to treat hypercholesterolemia, benefitting patients regardless of ancestry.[5]

Ancestry category distribution of individuals in human genome studies catalog



Source: Source: Sirugo G, Williams SM, Tishkoff SA. The Missing Diversity in Human Genetic Studies. Cell. 2019 May 2;177(4):1080.

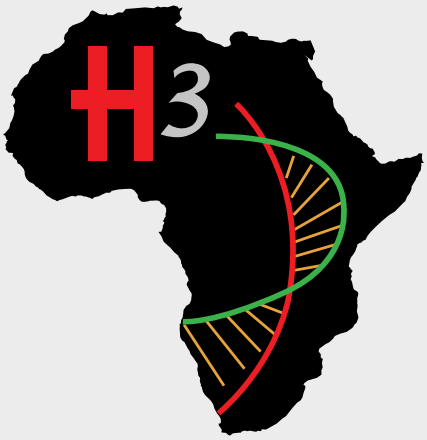
Genetics & Genomics: Striving for Health Equity

The COVID-19 pandemic has disproportionately affected racial and ethnic minorities in the U.S., illuminating that there are social factors in this country that cause major health disparities. It is imperative that the application of genetic science in healthcare does not worsen existing health disparities[6] but instead advances health to benefit all Americans. Participation of individuals from diverse groups is essential if we are to realize the full promise of genomics research and the equitable application of genetic discoveries in healthcare and society. Achieving greater participation comes with fundamental responsibilities to conduct research in ways that address the distinct needs of vulnerable populations.

Federally funded research programs and efforts to advance diversity and equity in research

- **UNITE** – this new initiative established by the National Institutes of Health (NIH) aims to "establish an equitable and civil culture within the biomedical research enterprise." One objective of UNITE is to "address long-standing health disparities and issues related to minority health inequities in the United States."[7]
- **All of Us** – created with the goals of having the research cohort reflect the diversity of the United States, and of having research participants involved in study development.[8]
- **H3Africa** – focuses on the vast genetic diversity in the African continent. [9]
- **Southern Community Cohort Study** – established to understand the root causes of cancer health disparities and includes high representation of African Americans and rural communities in southeastern U.S. [10]
- **Jackson Heart Study** – comprehensive, longitudinal study that focuses on African Americans, with strong community basis and community engagement. [11]
- **Hispanic Community Health Study/Study of Latinos** – long-term study of health, disease, and possible causes of health disparities among Hispanics and Latinos communities in the U.S.[12]

All of Us Research Program



How Can Congress Support Diversity and Equity in Research?

Congress should support NIH institutes and programs that emphasize diversity and equity in research and clinical trials participation by the public. Congress should also provide sufficient funding for researchers to do the work of engaging populations underrepresented in research.

Additional Resources

UNITE
https://www.nih.gov/ending-structural-racism/unite

All of US
https://allofus.nih.gov/

H3Africa
https://h3africa.org/index.php/about/

Southern Community Cohort Study
https://www.southerncommunitystudy.org/about-the-sccs.html

References:
ashg.org/advocacy/fact-sheets/

Jackson Heart Study
https://www.jacksonheartstudy.org/

Hispanic Community Health Study/Study of Latinos
https://sites.cscc.unc.edu/hchs/StudyOverview