Rethinking the use of race and other labels in genetics research

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Charmaine Royal's work on a national committee reviewing the use of population descriptors in genetics and genomics research will help guide researchers to conduct more rigorous research and build stronger relationships with the communities they are studying. Credit: Duke University

For decades genetics and genomics researchers have used race, ethnicity, ancestry and other population descriptors in research that has opened

powerful areas of study of human history and evolution, biology, diseases and heritable traits.

But these descriptors are slippery words with hard to pin down meanings. They are also contested words, and the use of "race" as a surrogate for biology or genetics has in particular led to confusion, discrimination and increased race-based health inequities.

In March, a national committee co-chaired by Duke Professor Charmaine Royal issued a report that emphasized the high stakes of ensuring that genetics research benefits all groups in society and mitigates harm.

Released after a year of public hearings and study, the committee's report recommended that researchers tailor their use of populations descriptors to the type of study and the research questions they are pursuing. The committee also generally recommended avoiding the use of "race" in genetics research, and using other population descriptors only when they state clearly and precisely what purpose they serve in the study.

"There may be times when using 'race' as a descriptor could be helpful, such as understanding the impacts of racism," said Royal, Robert O. Keohane Professor of African & African American Studies, Biology, Global Health, and Family Medicine & Community Health. "But race should never be used as a proxy for human genetic variation."

The report's recommendations present not a menu but a process, Royal said, encouraging researchers to consider what concept of human difference they wanted study and use terms that most effectively capture that information.

Too often in past and current genetics research, the population descriptor

is used when other factors are not considered, too difficult to study, or beyond the expertise of the researcher. For example, Royal said the committee found examples of genetic studies where race was used to the exclusion of environmental data, even when such data would provide more precise information. Noting that genetics research often requires a variety of disciplinary approaches, the panel recommended that genetics researchers collaborate with social scientists, epidemiologists and other relevant experts whenever possible.

The report also emphasized the ethical obligations in genetics research of engaging and respecting the community under study.

"Researchers should consider whether the benefits of the research are equitable and avoid exacerbating existing inequities or creating new ones. We provide them with a roadmap of sorts to help them conduct research that is more scientifically and ethically sound. We need to change the thinking behind and approaches to a lot of genetics and genomics research to produce science that is meaningful not just for the researchers, but for the participants and their communities as well."

The committee was formed under the auspices of the National Academies of Sciences, Engineering, and Medicine at the request of the National Institutes of Health. Royal served as co-chair along with Aravinda Chakravarti, director of the Center for Human Genetics and Genomics at New York University.

Royal serves as director of Duke's Center on Genomics, Race, Identity, Difference and the Duke Center for Truth, Racial Healing & Transformation. She is internationally known for her research, scholarship, and teaching on the ethical, social, scientific, and clinical implications of human genetics and genomics research, and her appointment as co-chair of the committee is a sign of Duke's leadership in the intersection of basic science, humanities, social science and health

research.

Seventeen scholars from the biomedical and social sciences took part, bringing a variety of perspectives to the discussion. The year-long study was challenging, Royal said, but in the end, she came away encouraged by the work.

"As an interdisciplinary researcher in human genetics, I'm always thinking and talking about the importance of environmental and social factors even in studying genetic diseases such as sickle cell. The committee's work was challenging and intense, but to hear other geneticists emphasizing in this report the value of including environmental factors in genetics research was particularly encouraging.

"There was an acknowledgement that our work in human genetics and genomics is incomplete without a better understanding of the lived experiences of the individuals and communities we study. A more integrative and humane approach is a key component of the way forward to better science."

Select recommendations

- Not use race as a proxy for human genetic variation. In particular, they should not assign genetic ancestry labels to individuals based on their race, regardless of whether the label was self-identified.
- Apply labels consistently to all participants. For example, if ethnicity is the most appropriate descriptor, all participants should be assigned an ethnicity label, rather than labeling some by race and others by ethnicity.
- Be attentive to the connotations and impacts of terminology they use to label groups. The report points to the term "Caucasian" as an example, explaining it should not be used under any

circumstance because it was originally coined to convey the notion of white supremacy.
- Disclose the process by which they select and assign group labels. If researchers develop new labels for existing samples, researchers should provide a description of the differences between the new and old labels.

More information: Using Population Descriptors in Genetics and Genomics Research (2023). DOI: 10.17226/26902

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