Using Population Descriptors in Genetics and Genomics Research

A New Framework for an Evolving Field

Genetics and genomics research has grown exponentially over the past decade, providing exciting opportunities to transform medicine as we know it. The genetics ecosystem has expanded such that genetics and genomics research is now conducted across a wide variety of disciplines. Researchers have long used population descriptors, or concepts of categorization, to capture information related to the continuous and complex patterns of human genetic variation resulting from history, migration, and evolution. Many of these descriptors focus on *descent-associated* groupings (i.e., populations believed to share characteristics based on a common origin). Examples of population descriptors include race, geography, and nationality, which all have complex social definitions and connotations. The misuse of these descriptors—in particular, race—has persisted and influenced thinking in genetics and genomics that has benefited some while marginalizing others.

Researchers using genetic and genomic data have long struggled with a lack of clear and specific guidance regarding the use of population descriptors. Over the years, many efforts have sought to develop appropriate guidelines with little success. Chapter 5 of this report recommends new approaches for the use of population descriptors according to genomics study types. For both individual and collective behavior to change, incentives, as well as accountability measures, should be in place. There is a great need for partnership among relevant parties to support researchers during the implementation process, as described in Chapter 6.

RECOMMENDED ACTIONS

Offer publicly available tools to facilitate implementation. Although the genomics research ecosystem comprises many different groups, implementation of the recommended changes may disproportionately rely on individual researchers. It is important that other relevant parties, including professional societies and journals, share the responsibility and support researchers to effect lasting change.

Action: Funding agencies, research institutions, research journals, and professional societies should offer tools widely to their communities to facilitate the implementation of these recommendations. The tools should be publicly available, especially when they are supported by public funds. Such tools could include:

- Educational modules for inclusion in human research protection training
- Manuscript submission and review guidelines (example checklist below)

- Grant submission and review criteria
- Training and education of individuals at all levels
- Opportunities for continuing education for researchers
- Informatics tools, such as data structure standards for sharing labels and labeling procedures used within a study

Align policies and procedures and invest in strategies to support implementation.

The ability to advance trust and improve research depends on how groups implement the report's recommendations. Current systems for supporting and rewarding genomics research may impede rather than facilitate implementation. It is important that organizations candidly evaluate how well their current practices and procedures align with the recommendations and course correct where needed. If changes cannot be made to resolve discrepancies, groups should be transparent, provide justification as to why, and determine how misalignment will be mitigated.

> **ACTION:** Key partners, including funding agencies, research institutions, and scientific journals, should ensure that policies and procedures are aligned with these recommendations and invest in developing new strategies to support implementation when needed.

Disclose the process for selection, assignment, and rationale of group labels.

Action: Researchers should disclose the process by which they selected and assigned group labels and the rationale for any grouping of samples. Where new labels are developed for legacy samples, researchers should provide descriptions of new labels relative to old labels.

Professional societies and journals often face the challenge of ensuring their members and authors abide by stated guidelines. One potential strategy for journals is to create a checklist to guide both how descriptors are reported and how the process of selecting them is described in publications. Furthermore, journals could work together to adopt these principles and recommendations via professional organizations focused on changing the publishing culture.

CONCLUSION

To create sustainable change, the full genetics and genomics research ecosystem—research participants, research funders, professional societies, research journals, research institutions, and individual researchers—must work together to implement these recommendations. Both transparency and communication among the relevant groups will be critical. The field is quickly evolving, and the time to meet this need is now.

EXAMPLE CHECKLIST THAT JOURNALS CAN IMPLEMENT FOR GENOMICS RESEARCHERS

- Is there a description in the methods section of population descriptors (e.g., race and/or ethnicity, geography) that were collected?
- Is the source data of each population descriptor reported (e.g., database, electronic health record, survey)?
- Is there a description of how participant population descriptors were selected and how group labels were assigned?
- Is a scientific justification provided for the collection of population descriptor data?
- Are population descriptors being used as proxies for environmental variables? If so, is this noted and explained?
- Are appropriate reference categories for the populations of interest being used as reference categories in analysis? Is there a scientific justification for these approaches?
- In studies of genetic contributions to health disparities, are social influences, environmental exposures, and other likely relevant variables included? If not, is the lack of assessment of the possible roles of these nongenetic factors discussed as a limitation?

SOURCE: Adapted from Genetics in Medicine's Guide for Authors, https://www.elsevier.com/journals/genetics-in-medicine/1098-3600/guide-for-authors.

FOR MORE INFORMATION

This Consensus Study Report Action Guide was prepared by the Health and Medicine Division based on the Consensus Study Report *Using Population Descriptors in Genetics and Genomics Research: A New Framework for an Evolving Field* (2023).

The study was sponsored by the National Institutes of Health (NIH) in the Department of Health and Human Services: All of Us Research Program; Eunice Kennedy Shriver National Institute of Child Health and Human Development; National Cancer Institute; National Heart, Lung, and Blood Institute; National Human Genome Research Institute; National Institute of Dental and Craniofacial Research; National Institute of Diabetes and Digestive and Kidney Diseases; National Institute of Environmental Health Sciences; National Institute of Nursing Research; National Institute on Aging; National Institute on Drug Abuse; National Institute on Minority Health and Health Disparities; NIH Office of Behavioral and Social Sciences Research; and NIH Office of Science Policy. Any opinions, findings, conclusions, or recommendations expressed in this publication do not necessarily reflect the views of any organization or agency that provided support for the project.

This Consensus Study Report is available from the National Academies Press, (800) 624-6242 or https://nap.nationalacademies.org/catalog/26902.

Health and Medicine Division

Division of Behavioral and Social Sciences and Education

NATIONAL ACADEMIES *Sciences Engineering Medicine*