



Genomics and Society Working Group

National Advisory Council for Human Genome Research
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Stephanie Malia Fullerton, DPhil
Professor and Interim Chair, Department of Bioethics & Humanities

UNIVERSITY *of* WASHINGTON

GSWG Mission

- > **Provide NACHGR advice on short- and long-term planning and priority setting for genomics and society activities at NHGRI, particularly the ELSI Research Program**
- > **Provide input about:**
 - **ELSI research priorities**
 - **Appropriate balance between investigator- and program-initiated research**
 - **Best use of limited budgetary and staff resources**

Current GSWG Members



Co-Chair:
Malia Fullerton, DPhil



Co-Chair:
Kyle Brothers, M.D., Ph.D



Shawneequa Callier, J.D.,
M.A



Stephanie Russo Carroll
DrPH, MPH



Marsha Michie, Ph.D.



Gabriel Lázaro-Muñoz,
Ph.D., J.D.



Lisa Parker, Ph.D.



Holly Landrum Peay,
Ph.D., M.S.



Beth Tarini, M.D., M.S.



Genevieve Wojcik, Ph.D.

Reminder: Sept 2022 GSWG Report

- > *ELSI research and its role in guiding efforts to address structural inequities in genomics research*
- > *Prioritize population-level analyses, including responsible incorporation of SDOH, sustained community involvement in research, and consideration of health equity*
- > *Broaden ELSI expertise to include new perspectives*

ELSI Research Priorities

Continuing , and New in 2023

- > **Health equity**
- > **Structural factors and social justice**
- > **Genomics beyond healthcare**
- > **Gene and environment interaction research**
- > **Social Determinants of Health (SDOH)**

- > **Diversity in the ELSI Research Portfolio**
- > **Follow-up of NASEM Population Descriptors Report**

ELSI Priority: Health Equity

- > **Questions asked today are similar to those asked decades ago**
 - Action is needed now to ensure that genomic studies generate knowledge that will benefit diverse patients and communities
- > **Review criteria that address health disparities and health equity as explicit outcomes of genomic research will be critical**
- > **While enhanced workforce diversity is important, genomics cannot rely solely on people from underrepresented groups to break down research and translational barriers**

ELSI Priority: Structural Factors and Social Justice

- > **Structural factors and structural racism continue to be important challenges for both ELSI and genomics**
- > **ELSI Research Program areas have been reorganized to highlight research areas related to Sociocultural Values and Genomics at the Institutional and System Level**
- > **To better address these areas, ELSI must attract more scholars and fields with expertise in structural inequities and justice theory**

NEW: Diversity in the ELSI Research Portfolio

- > **At the request of the GSWG, NHGRI staff provided preliminary data from two portfolio analyses**
 - Minority health/health disparities research at NHGRI
 - Research and Institution Diversity within the ELSI Program
- > **Measures of diversity:**
 - Project measures (e.g., diversity-related NIH RCDC categories, health disparities research, minority health research)
 - Institutional measures (e.g., Minority Serving Institution (MSI), IDeA state)
 - *Future analyses will include investigator measures, as available (requires DUA)*
- > **Preliminary findings provoked rich discussion about ways to continue to monitor and promote diversity moving forward**

ELSI Priority: Genomics Beyond Healthcare

- > **Genomics research has expanded beyond traditional (health-focused) applications, e.g., sociogenomics, forensic uses**
 - **Appropriate, indeed essential, for ELSI research community to engage with non-health applications**
 - **Simultaneously, care should be taken to not let 'easy villains' distract us from broader structural concerns**
- > **ELSI can work with and support the rest of the genomics community in combating misinformation**

ELSI Priority: Gene and Environment Interaction

- > **Genomic ELSI often focuses on individual's risks and benefits while environmental sciences focus on community level**
 - As genomics moves into population health, we need to think about population-level ELSI, and learn from environmental studies
 - Groups that have been previously harmed, minoritized, and/or abused in research are as concerned about harms to groups as to individuals
- > **ELSI can help guide the responsible conduct of G x E research**
 - This may require ensuring community members are involved in all aspects of research, including study design, implementation, dissemination
 - Recognition that many environmental risk factors are not under individual control

ELSI Priority: Social Determinants of Health

- > Inclusion of SDOH in genomics research is a scientific issue first and foremost, with related ethical, legal and social implications
- > Improving work on SDOH must involve:
 - Understanding how and why studies of complex traits might avoid genomic determinism
 - Ongoing extensive community interaction and engagement, and
 - Developing sustained partnerships with SDOH researchers
- > *Last year:* NASEM report on population descriptors *will* identify related methods and challenges to be considered

NEW: NASEM Report is Now Out

About

Announcements

● Publications

Resources

Description

Frequently Asked Questions

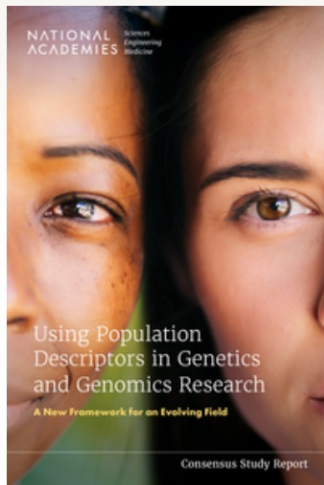
Committee

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Past Events

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Publications



2023

Using Population Descriptors in Genetics and Genomics Research: A New Framework for an Evolving Field

Genetic and genomic information has become far more accessible, and research using human genetic data has grown exponentially over the past decade. Genetics and genomics research is now being conducted by a wide range of investigators across disciplines, who often use population descriptors inconsistently and/or inappropriately to capture the complex patterns of continuous human genetic variation.

[Read Full Description](#)

RESOURCES

 [Report Highlights](#)

 [Recommendations](#)

 [Action Guide: Professional Societies and Research Journals](#)

 [Action Guide: Research Institutions and Funders](#)

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<https://www.nationalacademies.org/our-work/use-of-race-ethnicity-and-ancestry-as-population-descriptors-in-genomics-research>

NEW

Explainer

Use of Population Descriptors in Genomics



Explore this page

[What are population descriptors? >](#)

[Understanding genetic ancestry, race and ethnicity >](#)

[How well can researchers determine genetic ancestry? >](#)

[Are population descriptors social constructs? >](#)

[Why should researchers be intentional about how population descriptors are used in genomics research and health? >](#)

[Why does NHGRI care about this issue? >](#)

<https://www.genome.gov/about-genomics/policy-issues/population-descriptors-in-genomics>

Population Descriptors in Genomics: GSWG Recommendations to Council

1. Consider requiring applicants to explain and justify their use of population descriptors within grant applications, as appropriate.
2. Consider how best to incentivize the adoption of NASEM recommendations by investigators and consortia. Resources and tools which are foundational or widely utilized would, ideally, be prioritized.
3. Consider support of novel research designed to foster the development of new research frameworks and methods related to genetic similarity. Collaborative teams could include both genomic scientists and ELSI researchers, as appropriate.

Conclusions

- > **ELSI priorities continue to focus on considerations of health equity, social justice, and the responsible collection and use of SDOH in conjunction with genomic research**
- > **New recommendations for the use of population descriptors in genomics are now codified and the GSWG urge the NACHGR to consider how best to ensure their rapid implementation**
- > **Continuing efforts to collect data on diversity in the NHGRI (ELSI and other) research portfolio are welcome**



ELSIcon2024

THE 6TH ELSI CONGRESS

*Reimagining the Benefits of
Genomic Science*

Columbia University • June 10-12

<https://www.mhe.cuimc.columbia.edu/6th-elsi-congress>



smflrtn@uw.edu