

Supplemental information

Recommendations on the use and reporting of race,

ethnicity, and ancestry in genetic research:

Experiences from the NHLBI TOPMed program

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Additional background

From 2014-2020, the TOPMed Data Coordinating Center (DCC) was housed in the Genetic Analysis Center (GAC) in the Department of Biostatistics at the University of Washington¹. The GAC has performed scientific, analytical, and/or administrative coordination for a range of human genomics consortia and programs over the past 15 years including the NHGRI Gene Environment Association Studies consortium (GENEVA, 2007-2011), NHGRI Genomics and Randomized Trials Network (GARNET, 2009-2012), and the NHLBI Hispanic Community Health Study/Study of Latinos (HCHS/SOL, 2013-2016). Through these efforts, we have established standards for genotypic data quality assurance that account for population structure², grappled with how to analyze and report genetic diversity, e.g. among Hispanic/Latino groups³, and developed statistical methods and software for analyzing diverse datasets⁴⁻⁷. In 2018, GAC staff initiated monthly internal discussions on the use of race, ethnicity, and ancestry in genetics research—engaging with academic literature, public media, and our own experiences working in TOPMed and prior genetics consortia. We discussed a variety of articles across disciplines⁸⁻¹¹ and invited guest speakers on topics such as statistical rationale for stratified analyses and the co-opting of population genetic research by white supremacists on social media¹². Discussions were informed by a range of training and experience at the DCC, including biostatistics; statistical genetics; science communication; public health genetics; and ethical, legal, and social implications (ELSI). From these discussions, we recognized the opportunity as a DCC to help establish recommendations for TOPMed researchers that address the challenges of working with diverse data and incorporate anti-racist principles¹³ into the research process.

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