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SPECIAL ARTICLE

Taking an antiracist posture in scientific publications in human genetics and genomics

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From its earliest days, the field of human genetics has had a complex, and at times troubling, connection with racist ideologies. Although the modern field of human genetics and genomics has come a long way from those earlier errors, systemic racism remains ingrained in its institutions and practices. Although a variety of efforts are needed to excise systemic racism, we focus in this commentary on the work that must be done in scientific publishing in genetics and genomics. We propose eight principles that are both scientifically grounded and antiracist that we hope will serve as a foundation for the development of policies by publishers and editorial boards that address the unique needs of the field of genetics and genomics. Publishers and journals must go beyond mere policies, however. Editors and reviewers will need training on these policies and principles, and will benefit from resources like rubrics that can be used for evaluating the adherence of submissions to these guidelines.

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INTRODUCTION

The field of human genetics has had a complex, and at times troubling, connection with what has been called an "ideology of race,"[1] the belief that (1) the human species is comprised of scientifically distinguishable racial groups; (2) these groups are morphologically, behaviorally, and intellectually distinct; and (3) these features allow for racial groups to be ordered in a hierarchy of superiority.[2] This corrupt belief system predated the emergence of human genetics as a distinct scientific field and influenced its early development. Carl Linnaeus, for example, divided the human species into four "varieties" based on continent, skin color, and other "traits."[3] Charles Darwin likewise viewed humanity as comprised of biologically distinct races, and believed that the physical and intellectual differences he perceived among these races were explained by heredity.[2] As human genetics developed in the 19th and early 20th centuries, the "ideology of race" was treated as a background assumption for genetic science. Work during this period thus tended to reinforce these assumptions rather than call them into question.[2]

The tragic result of this history was the emergence of eugenics in the early decades of the 20th century. Early geneticist Francis Galton coined the term "eugenics" and founded the movement, explicitly referencing his half-cousin Darwin as his inspiration.[4] The eugenic conception of "genetic inferiority" was tied indelibly with the belief that one racial group was fundamentally superior to the other racial groups.[4] The racial valence of eugenic conceptions of superiority was manifest not only in the overtly racist ideology of the National Socialist German Workers' Party, but also in the practices of eugenicists in the United States.[5] The eugenics movement in the United States was led by geneticists at the Eugenics Record Office in Cold Spring Harbor, New York, initially directed by Harry H. Loughlin, who lobbied for legislation to restrict immigration and sterilize "defectives."[6] Recent work has further revealed striking trends in American psychiatric facilities that implemented eugenics practices, where Black and Latinx individuals were far more likely to undergo involuntary sterilization compared with White individuals.[7,8]

While the German Nazi Party was defeated and American eugenics laws were repealed, the ideology underlying them—the belief that race has a biological and genetic basis—has persisted. This idea remains a fundamental assumption, for example, of white supremacists in the United States. Some groups make a public display of drinking milk to demonstrate their claimed genetic superiority.[9] Racism is even reflected in federal policy, where control of non-White populations through immigration laws and involuntary sterilization is still a live issue today.[10] The effect of genetics on the way members of the public understand race can also emerge in more subtle ways. For example, some clients of direct-to-consumer genetic ancestry tests demonstrate an increase in racial essentialist beliefs (i.e., that race is fixed and determines innate abilities) after viewing their ancestry results.[11]

These beliefs remain, despite the fact that modern work in genetics and genomics has demonstrated with elaborate detail that the idea of race is logically incoherent and has no biological basis.[12] While human ancestral populations can be discerned from one another by examining the frequency of a large set of genetic variants and descent from common ancestors (i.e., identity by descent), most common variants in the human genome are found across all populations.[13] The discernible genetic differences among populations merely reflect differing frequencies of common variants among these populations, and only reflect a small proportion of the overall human genome that is fundamentally the same across all human groups.

Focusing on the eugenicists of the past and the white supremacists of the present, however, risks causing us to lose sight of the more subtle forms of racism that remain in our field. The so-called "slavery hypothesis," for example, posits that US Black populations face an elevated risk of developing hypertension as a result of the selective pressure experienced by their ancestors during the brutal Middle Passage from West Africa to the Americas and their subsequent enslavement. Given that this hypothesis is unsupported by either genetic or historical evidence, its tenacity among scientists and clinicians seems to reflect racist

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assumptions rooted in genetic determinism and beliefs about the association of genetic "defects" within racial groups.[14]

In this context, however, we have in mind not only the beliefs associated with racism, the "ideology of race," but also the subtle and often unconscious ways these belief systems can influence the way institutions are built and operate, and lead to "a system of advantage based on skin color."[15] Numerous manifestations of this institutionalized form of racism, or *systemic racism*, can be found in the contemporary field of genetics and genomics. The most obvious, perhaps, is the ongoing disparity in the inclusion of non-European populations in genomics research.[16] While efforts have been made to address this discrepancy, recent analysis demonstrates that significantly more work is needed.[17]

This trend is demonstrative of the insidious effect that racism has had in the field of genetics and genomics, in that the cause of this disparity is rarely attributed to racism. And to be sure, the overt form of racism practiced by white supremacists played no significant role in this disparity. Rather, it has resulted from at least two dynamics. On the one hand, this disparity has been driven in part by the so-called "Tuskegee effect," the unwillingness of Black individuals to participate in biomedical research as a result of historical transgressions against Black research participants. On the other hand, it has been driven by a methodological decision starting over a decade ago to stratify genome-wide association studies (GWAS) by continental ancestry, a decision that resulted in the large majority of early GWAS studies being conducted exclusively in populations of European ancestry.

Fundamentally, both of these phenomena reflect the effects of systemic racism. The unwillingness of Black, Latinx, and Asian individuals to participate in biomedical research is not an irrational response driven by paranoia, but rather an entirely rational and understandable response to the systemic racism that has been a part of biomedical research for over a century now.[18–20] In fact, the use of the euphemism "Tuskegee effect" itself reflects the broader societal norm of obscuring the unsavory truth of racism, in this case in the field of biomedical research. This term tends to shift the locus of blame to potential research participants and their unwillingness to participate, rather than to blame the researchers and research institutions that committed these racially driven actions. In fact, most of the researchers responsible for the persistent and widespread abuses that took place in the course of the past century were never associated with Tuskegee University. This is true even of the investigators who worked on the US Public Health Service Syphilis Study itself, which was conceived of, sponsored, and carried out in large part by individuals outside Tuskegee University. In truth, it was not this single event that has dissuaded Black individuals from participating in research; this phenomenon is both historical and contemporary, and involves striking events that have been recognized by the general public as well as more insidious offenses that have taken place repeatedly throughout the biomedical research enterprise. Examples of these abuses date back to at least the mid-18th century, including surgeries conducted without anesthesia on Black slaves and babies in the name of research, and the segregation of health care during the internment of Japanese immigrants in World War II.[21,22] And more recently, abuses of Native Americans in the conduct of genetic research led to lawsuits against research institutions, banishment of researchers from reservations, and moratoriums on genetic research on tribal lands.[23]

The methodological decision to stratify GWAS studies, although rooted in a real scientific challenge, also demonstrates the effects of systemic racism. It was recognized in the early years of this methodology that GWAS analyses needed to be stratified by continental ancestry. While stratification could have been achieved while including multiple ancestry groups in the same study (using either the stratified meta-analysis approach or the joint mixed-model approach),[24] a disproportionate number of studies addressed this issue by simply limiting analyses to populations of European ancestry, frequently because this group was disproportionately represented in the existing databases close at hand. This strategy reflected both opportunism, since these populations were less deterred from research participation due to past transgressions against their community, as well as a subtly racist acceptance that starting with European populations was "good enough" on the logic that other populations could be added later.

This example, then, helps us see that systemic racism, a form of racism that is embedded as normal practice within institutions throughout society, is alive and well in biomedical research, including genetics research, even though the vast majority of scientists and other stakeholders in this work unconditionally reject more overt forms of racism. The uncomfortable truth is that systemic racism remains ingrained in the genetics and genomics research enterprise, where it influences scientific priorities, study design decisions, interpretation of research findings, and even admission and mentorship strategies in research and clinical training programs.

THE DUTY FOR GENETICS AND GENOMICS JOURNALS TO TAKE ACTION

It is evident that the effort to disassemble the structures of systemic racism within the genetics and genomics research enterprise will need to include a broad range of stakeholders. Training programs will require concerted effort by administrators, faculty, and trainees. Research laboratories will need to have tough internal discussions about whether their operations and research practices are being influenced by racist assumptions, and they will need to engage in broader conversations with colleagues, including those being led by organizations like the American Society of Human Genetics (ASHG).[25] And in nearly all of these contexts, efforts are likely to be most effective if "insiders" to the scientific community engage with "outsiders" representing communities that have been underrepresented in research participation and scientific professions.

In the context of this commentary, however, we are speaking out in our roles as editors of a journal that focuses on enhancing the knowledge, understanding, and practice of medical genetics and genomics. As a conduit for the dissemination of scientific findings in genetics and genomics, we recognize that we have both an opportunity and an obligation to join in the effort to excise systemic racism from biomedical research. As we have shown, the field of genetics and genomics has a unique opportunity in this respect, given our ability to interpret and explain the scientific evidence about the biological nonexistence of races, and our historical error as a field in reifying this concept.

As representatives of scientific journals in this field, we have an obligation to ensure that our platform is not used as a conduit for perpetuating scientifically unfounded or discredited ideas. We also have an obligation to lead the community on how best to communicate about scientific insights related to the role of systemic racism in health and the delivery of health care, and the interacting role of genetic ancestry and systemic racism in the development of disease and other traits. Finally, scientific journals have an opportunity to promote interdisciplinary dialogue about controversial ideas in a context where factual claims are subject to peer review.

We seek in this commentary to propose principles that are both scientifically grounded and antiracist, in that they are intended to help excise racist assumptions and practices from scientific publishing in genetics and genomics. We are preceded in this effort by outstanding work that has similarly aimed to address systemic racism in scientific publishing.[26–29] This previous work forms the foundation for the proposals we set forth in this piece. We are inspired in particular by a recent article in *Health Affairs Blog*, in which authors Boyd, Lindo, Weeks, and McLemore call on

journals in the domain of health to adopt an antiracist posture by denouncing biological race and unmasking the role of racism in health and health care, an effect that is often obscured by euphemism and victim blame.[30]

Our goal in this article is to build on this previous work by "drilling down" on the issues raised most acutely by contemporary genetics and genomics research, especially work focused on the role of genetics and genomics in medical care, public health efforts, and human health and wellness. We call on the community of journal editors and publishers working in this domain to develop policies and standards implementing the principles we advance here, and for editors and peer reviewers evaluating the scientific impact and rigor of scientific work to consider these principles in their decision-making:

Principle 1: Race should be used in research studies involving health-care delivery, etiologies of medical conditions, and health outcomes, but only as a sociopolitical category. The inclusion of race variables is especially important in contexts where health disparities are observed. While biological race is scientifically unfounded, race as a socially constructed category plays a major role in health-care delivery and health-care outcomes.[31] This effect is especially apparent in the domain of health disparities, which are largely driven by factors like inequalities in the distribution and delivery of health-care services, residential segregation, poverty, and social injustice, all of which are driven by systemic racism. Failure to include race as a sociocultural variable in scientific studies risks obscuring these effects on health, even for studies focused primarily on genetics and genomics. Utilizing race without a scientific justification, however, risks perpetuating racial essentialism. It is therefore important that when race is used as a sociopolitical category, authors explain the reasons for its inclusion.[32–34]

Principle 2: Genetic ancestry should not be used as a surrogate for sociopolitical race. Sociopolitical race should not be used as a surrogate for genetic ancestry. Data reflecting genetic ancestry is most often used as a surrogate for sociopolitical race in studies involving the secondary use of clinical data collected in contexts where race variables are not documented using rigorous methods. Data reflecting sociopolitical race, on the other hand, is often used as a surrogate for genetic ancestry because it is easier to ascertain than genetic ancestry, such as when a study does not involve genome-wide data. The use of these measures as surrogates for one another is problematic both because it lacks rigor, and because it reifies the mistaken tendency to equate these concepts.

Principle 3: Authors should avoid the use of terms that obscure the distinction between sociopolitical race and genetic ancestry, as well as terms that evoke historical conceptions of racial superiority.[32,33] For example, the terms "Black" and "White" for racial groups in the American context are preferable in genetics and genomics research, since alternative terms like "European American" and "African American" could mistakenly be understood to reference continental genetic ancestry. The term "Caucasian" is particularly problematic; it not only implies genetic ancestry, but also is linked with historical conceptions of White superiority and the "ideology of race."[34] Terms like "Asian" that combine numerous self-identified groups under a single umbrella risk not only conflating sociopolitical race and genetic ancestry, but also obscuring disparities in specific subgroups.[35] Disaggregating such categories by self-identified ethnicity or national origin can help address this challenge. The use of more granular terms when discussing genetic ancestry can similarly help prevent continental ancestry (e.g., "Asian ancestry") from becoming a proxy for social categories of race (e.g., "Asian-American race").[36,37] Publishers and journal editors can support these goals by incorporating guidance on these issues into a manual of style and by providing authors, editors, and reviewers with clear rubrics.[32,33]

Principle 4: Variables related to race as a sociopolitical category, as with all scientific variables, should be ascertained and described in a rigorous manner. Whenever possible, self-reported race and ethnicity should be used. Perceived race is still frequently used in health-care settings. However, this is not a scientifically rigorous method, especially given its dependence on the preconceptions of health-care providers and other staff.

Principle 5: Authors should explicitly name racism when it is an underlying factor leading to health disparities, and should further describe this racism in terms of its form, mechanisms, etc.[30] Too often, the sociopolitical factors leading to disparities in health outcomes are described in oblique terms intended to obscure uncomfortable truths, much in the way the term "Tuskegee effect" is used to conceal the history of racial transgressions in biomedical research. This often takes the form of a focus on mechanisms alone—discrepancies in access to care, environmental exposures, educational inequalities, etc.—rather than the underlying systemic racism that causes and perpetuates these mechanisms. Disparities are also sometimes described in ways that frame the disparity as a characteristic of the group, rather than highlighting the failures of the health-care system or other institutions in serving these groups.[38]

Principle 6: Authors examining the genetic contribution to health disparities should avoid framing health disparities in reductive terms. It is to be expected that research in genetics and genomics will include a focus on elucidating the genetic contributions to health disparities, such as differential drug response between genetic ancestry groups. However, emphasizing genetic factors risks de-emphasizing the role of social determinants of health, including those driven by systemic racism. Even when scientific work is designed to examine possible genetic underpinnings of health disparities, authors should contextualize those genetic factors within the broader set of factors that contribute to health disparities. For example, the discussion or limitations sections of journal articles focused on the role of genetic factors in health disparities should include a discussion of the social determinants of health, environmental exposures, and other factors that also influence the observed disparities. Even better, genetics and genomics researchers should engage in interdisciplinary work designed to characterize both the genetic and social determinants of health disparities.

Principle 7: Given the underrepresentation of Black, Latinx, Asian, and other non-White populations in genetics and genomics research, editors and reviewers should prioritize manuscripts with strong representation of these groups, even when findings replicate earlier findings in White populations. The "input–output problem" highlights that when research is conducted with a single ancestry group, other ancestry groups will tend not to experience the same level of benefit from that research.[39] This is especially the case for precision medicine applications, where treatments may be based on genetic variants that occur with different frequencies in different ancestry groups. It is thus important for biorepositories and research cohorts to actively work to increase representation of non-European ancestry groups, and for authors to discuss limitations that may arise in their work as a result of underrepresentation.

Principle 8: Authors should carefully avoid structuring data tables and other representations of data in such a way as to treat White populations or European ancestry groups as the "normal" in group comparisons. For example, tables reporting odds ratios should typically not utilize White populations as the reference category.

CALL TO ACTION

These principles are designed to address systemic racism in scientific publishing in the field of genetics and genomics. Without action by authors, reviewers, editors, and publishers, however,

these principles are merely good ideas. We call on publishers to develop and circulate template policies that will lower the "activation energy" for individual journals to undertake efforts to combat racism. Such policies, when combined with educational efforts and thoughtful peer review, would also have the salutary effect of raising awareness of the ways racism is embedded in research. For the editorial boards and other leaders of individual journals, the time is ripe to adopt explicit and actionable policies to begin disassembling systemic racism that influences journal practices and the broader scientific fields they serve.

For *Genetics in Medicine* in particular, we hope this commentary will serve as a foundation for the development of policies that address the unique needs of the field of genetics and genomics. We call on peer journals in this field to likewise implement policies operationalizing the principles laid out here. Journals must go beyond mere policies, however. Editors and reviewers will need training on these policies and principles, and will benefit from resources like rubrics that can be used for evaluating the adherence of submissions to these guidelines.

Given that racist ideology has influenced the field of genetics and genomics since its earliest days, it is clear that disassembling the influences of systemic racism in this field will be no easy task. As layers of racist structures are peeled back, we will discover additional practices whose racist assumptions were not previously discernible. In this way, we believe the effort to excise racism will be an iterative process. It is time to take the next difficult step in that process. We hope these principles will help chart a course forward for journals focused on human genetics and genomics.

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