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Genes do not operate in a vacuum, and neither should our research

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Abstract

It's time for a paradigm shift in the scientific enterprise. Our social responsibilities, especially as stakeholders in a field such as genetics, are central to the responsible conduct of research.

The postgenomic era has ushered in a number of technological advancements, changing our understanding of ourselves and others. One of the most important lessons we have learned from the genetics revolution is that genes do not operate in silos—they act in concert with social and environmental factors. The 'nature' and 'nurture' dichotomy is over. We are now well aware that genes and the environment interact with each other. Even conditions that we might consider to be direct examples of genetic effects are influenced by the environment. For example, sickle-cell anemia is triggered by a mutation in the *HBB* (β-hemoglobin) gene on chromosome 11. Although having this mutation indicates that you will develop a severe form of sickle-cell disease (SCD), your experience as a person with sickle-cell anemia is shaped by your environment. SCD disproportionately affects African Americans in the United States, meaning that many people with SCD have to "face the consequences of a serious health condition … [and] navigate a society in which the color of their skin is often an unfair disadvantage"[1]. In other words, although a genetic mutation causes SCD, one's lifespan and quality of life are affected by the quality of healthcare that one receives. This increased focus on the interplay between genes and the environment signifies a larger and ever-present reality: the environment—including our upbringing, exposures and social structures—matters.

Yet, this reality has not always been recognized or studied, especially in genetics. This field represents the backbone of much of modern science but is plagued by inequitable and inappropriate applications and misconceptions. From the involuntary sterilization of Black and brown people, and women of lower socioeconomic status[2], to restrictions on immigration[3], the idea that some people are genetically superior to others has driven unethical practices and has been used to validate and further perpetuate social inequalities.

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Competing interests

The authors declare no competing interests.

As Black women, we are painfully aware that the conflation of race with ability remains today; to some in this world, we are deemed as being ‘less than’ and are scrutinized more harshly. Given the collective popular tendency to view genetics as separate from the environment, we as researchers in genetics and genomics need to address the social implications of our work as we conduct studies and communicate our findings. Genes do not operate in a vacuum, and neither should our research. Our social responsibilities are an essential component of responsible research conduct; these responsibilities require us to “address the moral, political, social and policy issues at stake”[4].

We believe there are two major ways to fulfill our social obligations to the public across disciplines and specifically within genetics. The first is to implement methods and practices that incentivize the scientific research enterprise to value social responsibility. A second equally important step is to increase the prevalence of, and the public’s accessibility to, professionals and researchers from historically marginalized groups in the field of genetics.

However, to incentivize social responsibility, we need to recognize its importance. Research is the process of discovery and learning. What we learn can lead us to innovate—to discover or create something new. To truly support the flourishing of all human beings, researchers have important social and ethical obligations that extend beyond our duties to one another and toward the many publics that compose our social world. It is our opinion that socially responsible research and research communication are important spaces for discovery and innovation that are as significant as our research findings.

Nevertheless, the primary responsibility of researchers has traditionally been to conduct research. What happens to that research, how it is interpreted and used, and the subsequent consequences can too often be left to policymakers and the public. In reality, however, science deals with values: the research questions we ask and the problems we try to solve stem from the questions and problems that individuals and collectives consider important according to our social structures. As many individuals of color know, the questions asked of science have not always proven to be equitably beneficial or in service to our needs and interests.

Recognizing and emphasizing our social responsibilities is challenging because the research enterprise does not prioritize it, feigns ignorance and/or implements perfunctory initiatives. Our research institutions are set up to protect human participants involved in research, but there are few incentives for community engagement and public outreach beyond mechanisms that reduce institutional and researcher liability. This needs to change. To practice social responsibility, we ought to consider how to increase the accessibility of our research and engage those who typically remain outside the decision-making process. Responsible innovation and research call for the genetics community to consider the roles of intragenerational and intergenerational justice[5]. In other words, when we think about how our work affects people and environments as they exist today (intragenerational justice) and as they might exist in the future (intergenerational justice), we can begin the ‘good trouble’ of dismantling systemic racism. Enacting these forms of justice means that we need to bring diverse voices into the research process from inception to finish. We can accomplish this by making our work accessible to multiple audiences and by identifying,

supporting and including people from diverse backgrounds in decision-making positions as researchers, journal editors, peer reviewers, grant-funding bodies, genetic counselors, members of community advisory boards and so on.

As an example for how to make our work more accessible, some researchers in the field of social genomics have taken to publishing frequently asked questions (FAQs) that accompany their genome-wide association studies on complex and socially fraught behavioral traits, such as educational attainment or income. These FAQs ask simple questions, such as: “Did you find ‘the gene for’ (or ‘the genes for’) risk tolerance?” (https://www.thessgac.org/faqs). These questions are answered in a similarly straightforward manner, for example: “No … the genetic factors we identified are involved in a long chain of biological processes that exert an influence on human behavior, and those processes are intricately entwined with the environment.” To further encourage these well-intentioned efforts, academic journals such as *Nature Genetics* could require authors to publish FAQs alongside academic research publications and make these FAQs open source. Journals serve as intellectual gatekeepers between the ‘ivory tower’ of academia and the public. Making materials open source, adding in easily accessible article summaries or providing public-health relevance statements up front will hold scholars and journals to greater levels of accountability, specifically in how research is conveyed to the wider community. Additionally, socially responsible communication and community engagement do not need to be text based; they can include videos, comics (https://botswanabaylor.org/genome_adventures.html) and other visual media.

Importantly, clinical-trial recruitment of African American participants in genomic studies is extremely low in comparison to that of other groups. Many studies have shown that African Americans are distrustful of genomic medicine because of a longstanding history of exploitation and continued medical racism[6,7]. Major concerns have been expressed by African American participants about genomic-medicine health practices and research based on past abuses in biomedical research. In a pilot study, Banda et al.[8] have shown that by using a culturally appropriate video, the authors were able to change the attitudes and intentions of African American patients with cancer to enroll in therapeutic clinical trials: 34% of patients showed positive changes in intention[8]. Thus, problems with accrual into genomic clinical trials can be ameliorated by culturally sensitive education.

Together with making research more accessible, researchers must also actively reflect on their work. It is not enough to state genomic findings. We ought to consider the impact and use appropriate approaches when stating conclusions and associations, especially when examining ancestral differences. A prime example is the use of European-ancestry populations as reference groups in most genomic studies[9], thus further perpetuating the notion that people of European descent are the standard, and everything else is a deviation. Additionally, using this method fails to acknowledge diversity within other ancestral groups and hinders research and policies aimed at reducing health disparities. Researchers and policymakers need to be better educated about the potential pitfalls associated with this approach and about alternative strategies that would be suited for their research goals. To accomplish this, better research and publishing guidelines are needed for disaggregating populations of different ancestries to alleviate methodological challenges[10]. We must

encourage researchers to consider the implications of using European-ancestry populations as the default reference group. The democratization of information on the Internet, along with historical and continued racism in genomics, has increased the distrust of experts and led to failures in distinguishing experts from non-experts. Thus, by increasing accessibility and establishing research guidelines for studying ancestral groups, we can create a more socially responsible research enterprise that historically marginalized people would be willing to participate in.

If our goal as researchers is to advance humanity and, when possible, contribute to the greater social good, then our institutions should be reformed accordingly. Often, we include or consult the internal voices of research and exclude the voices external to it. When researchers are taught about the responsible conduct of research, it is by examining past acts of violence or selfishness on the part of researchers. As the world begins a long and painful process of waking up to the realities of racism, colonialism and imperialism, we have an opportunity to create structural reforms that build societal responsibilities into the foundations of how research is conducted and disseminated. For instance, tenure committees should value socially responsible initiatives in tenure portfolios; doing so would make working with community-based organizations such as the Personal Genetics Education Project (PgEd) or Facing Hereditary Cancer Empowered (FORCE) enticing for researchers. These organizations are community-based initiatives that disseminate information on genetics and genomics and gather external perspectives; they are invaluable resources for giving voice to those who are often silenced or forgotten.

As Black women, we would be remiss not to talk about our own positions in genetics. We need more Persons Excluded because of Ethnicity or Race (PEER) as researchers, not just test participants. We look around our field and do not find ourselves reflected back. When Black researchers and participants are underrepresented in the field, this creates a form of invisibility that ultimately leads to worse science. It is only by bringing voices like ours to the table that new questions get asked and new answers are identified. Bringing in these diverse perspectives leads to better-thought-out conclusions and results in more robust findings. Finally, we must go beyond hiring PEER researchers; we need inclusive, antiracist supporting infrastructures at institutions and outside traditional research centers to ensure matriculation and retention. This process begins with engaging communities and local providers early on. For too long, the field of genetics has operated in a vacuum that does not consider the many gifts and contributions that the PEER community has to offer. Let the postgenomic era usher in more than technological progress. It’s time for a paradigm shift in the scientific enterprise.

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