Open letter

The ethics of characterizing difference: guiding principles on using racial categories in human genetics

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Published: 15 July 2008

*Genome **Biology*** 2008, **9:**404 (doi:10.1186/gb-2008-9-7-404)

The electronic version of this article is the complete one and can be found online at http://genomebiology.com/2008/9/7/404

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Abstract

We are a multidisciplinary group of Stanford faculty who propose ten principles to guide the use of racial and ethnic categories when characterizing group differences in research into human genetic variation.

Since the completion of the Human Genome Project, research focused on human genetic variation, including differences among groups, has intensified. This focus has rekindled debates about the connection between genetic (DNA-level) traits and human 'racial' difference [1-5]. Scholars are divided on the question of whether racial categorization is an appropriate means of organizing potentially useful genetic data or a pernicious reification of historically destructive typologies [6,7]. To explore these issues, faculty from the humanities, social sciences, life sciences, law and medicine at Stanford University convened over the past few years to engage in an extended interdisciplinary dialog. The initial meeting consisted of a two-day workshop in 2003 that developed into an ongoing faculty research seminar sponsored by the Stanford Humanities Center, Affymetrix Corporation, the Mellon Foundation and the Research Institute of the Center for Comparative Studies on Race and Ethnicity. This seminar series, which continued for two years and culminated in a public conference, included invited experts and led to the publication of a collection of essays [8].

Our goal was to generate principles to guide the use of race and ethnicity categories in research in human genetic variation. Central questions included the following: Can we find areas of common ground? Do we agree about the implications and interpretation of emerging genetic data? Under what conditions might genetic data transform social understandings of racial and ethnic categories, possibly enhancing racist ideologies? From this discussion, we have endorsed ten statements discussed below. Although not an exhaustive consideration of the broad range of issues that deserve attention, this article is intended to promote interdisciplinary dialog on these important concerns and to encourage responsible practices.

Statement 1: We believe that there is no scientific basis for any claim that the pattern of human genetic variation supports hierarchically organized categories of race and ethnicity

The equality of rights of all human beings is an unquestionable, moral claim that cannot be challenged by

descriptive, scientific findings [9-11]. As a normative commitment, equality is fundamental to our conception of human rights, and is not open to debate. Classification by racial and ethnic categories has, at particular moments in history, been used to further racist ideology [12]. In view of concerns that linking of emerging genetic data and race/ethnicity categories may promote racist ideologies, we emphasize that there is no scientific basis for any claim that the pattern of human genetic variation supports hierarchically ranked categories of race or ethnicity. Furthermore, we abhor any use of genetic data to reinforce the idea of between-group difference in order to benefit one group to the detriment of another.

Statement 2: We recognize that individuals of two different geographically defined human populations are more likely to differ at any given site in the genome than are two individuals of the same geographically defined population

Research in human genetics has highlighted that there is more genetic variation within than between human groups, where those groups are defined in terms of linguistic, geographic, and cultural boundaries [3,5,13,14]. Patterns of variation, however, are far from random. We recognize that human population history, including major migrations from one continent to another as well as more short-range movements, has led to correlation between genetic variation and geographic distribution [14-17]. This finding is particularly true of indigenous peoples; populations characterized by a high degree of interaction with neighboring groups adhere less to these patterns.

Statement 3: We urge those who use genetic information to reconstruct an individual's geographic ancestry to present results within the broader context of an individual's overall ancestry

An individual's 'geographic ancestry' or 'biogeographical ancestry' can be taken to mean the sum of all the geographic locations inhabited by an individual's biological ancestors. Often, however, genetic data reflect just a small subset of these ancestors. For example, knowing a person's Y-chromosomal lineage is at best a partial view of an individual's ancestry. We note also that in some cases individuals' or groups' self-identification differs from their biogeographic ancestry, depending on a range of historical, cultural and sociopolitical factors. We see value in recognizing both biogeographical and cultural ancestry that underlies an individual's and group's identity, particularly in the context of addressing health disparities.

Statement 4: We recognize that racial and ethnic categories are created and maintained within sociopolitical contexts and have shifted in meaning over time

Human genetic variation within continents is, for the most part, geographically continuous and clinal, particularly in regions of the world that have not received many immigrants in recent centuries [18]. Genetic data cannot reveal an individual's full geographic ancestry precisely, although emerging research has been used to identify geographic ancestry at the continental and subcontinental levels [3,19]. Genetic clusters, however, are far from being equivalent to sociopolitical racial or ethnic categories. Diverse populations identified as 'Hispanic', for example, are heterogeneous and have distinct ancestries and social histories [20]. We recognize that social experiences and conditions inform racial identity, making such identity a poor proxy for genetic ancestry.

Statement 5: We caution against making the naive leap to a genetic explanation for group differences in complex traits, especially for human behavioral traits such as IQ scores, tendency towards violence, and degree of athleticism

Among the most pervasive and pernicious claims of genetically determined traits are theories on the racial ordering of intelligence [21,22]. Despite the weak scientific basis for such ordering, the consistent return to the rhetoric of racial hierarchies of IQ reflects the powerful role that science has historically played in promoting racist ideologies [23]. Current evidence suggests that for most complex behavioral traits, contribution of any one gene to normal variation is small and these traits may be more fully explained by variation in environmental factors. We therefore caution against making the naive leap to a genetic explanation for group differences in a complex behavioral trait, where environmental and social factors clearly can and do play major roles [24,25].

Statement 6: We encourage all researchers who use racial or ethnic categories to describe how individual samples are assigned category labels, to explain why samples with such labels were included in the study, and to state whether the racial or ethnic categories are research variables

A first step towards preventing the use of science for racial stereotyping is careful consideration of the use of racial and ethnic categories in the initial design of research. Researchers can assess the purpose and impact of using racial and ethnic categories in their research and investigate whether alternative approaches would be appropriate. The editorial boards of several flagship scientific journals have issued publication guidelines to their authors on the use of 'race' in reporting research findings [26,27].

Statement 7: We discourage the use of race as a proxy for biological similarity and support efforts to minimize the use of the categories of race and ethnicity in clinical medicine, maintaining focus on the individual rather than the group

Although a broad range of associations between genetic markers and human traits - including diseases - is emerging, any accompanying correspondence with race or ethnicity is statistical. Although certain relatively rare genetic

diseases, such as Tay-Sachs, are found in higher frequencies in some human populations, the result of population bottlenecks or environmental pressure, these diseases are also found in other populations. Overemphasizing the genetic contribution to complex human disease or behavioral traits can promote not only racism, but also a naive genetic essentialism - the notion that genes determine health status or behavior [28-30]. Such essentialism is particularly dangerous in clinical translation, where a focus should be maintained on the individual rather than the group [31].

Statement 8: We encourage the funding of interdisciplinary study of human genetic variation that includes a broad range of experts in the social sciences, humanities and natural sciences

Common human behaviors and diseases result from the interaction of genetic, cultural, linguistic, economic, social and behavioral factors; genetic differences underlying behavioral or health status differences between groups are especially difficult to identify [32]. Medical research is most likely to be successful when genetic studies proceed in tandem with studies of environmental and behavioral factors that include geneticists, epidemiologists and social scientists as members of the research team.

Statement 9: We urge researchers, those working in media, and others engaged in the translation of research results to collaborate on efforts to avoid overstatement of the contribution of genetic variation to phenotypic variation

Scientific data are often quickly politicized and incorporated into specific policy agendas without extensive explanation of the scientific research and its details [33-35]. Often lost in the announcement of scientific findings is discussion of the limitations of the research. Our hope is that scientific data about human genetic variation might undermine spurious popular beliefs about the existence of biologically distinct human races and beliefs that support racist ideologies.

Statement 10: We recommend that the teaching of genetics include historical and social scientific information on past uses of science to promote racism as well as the potential impact of future policies; we encourage increased funding for the development of such teaching materials and programs for secondary and undergraduate education

Education is critical in providing both the foundation - basic scientific literacy - and the historical context through which to understand human genetic variation as data from studies are released. We believe that expanded public education at all levels will enhance understanding of human genetic variation and interpretation of any correspondence with categories of race and ethnicity. We recommend that the teaching of genetics include what we recognize today as past uses of science in promoting racism. Finally, we encourage increased funding for the development of such teaching materials and educational programs that focus on the social impact of scientific discoveries as well as the impact of social values and beliefs on the conduct of science.

In conclusion

The 'gene' remains a powerful icon in the public imagination and is often misunderstood as deterministic and immutable. Furthermore, history reminds us that science may easily be used to justify racial stereotypes and racist policies. Our discussion at Stanford University resulted in part from a desire to try to minimize the chances that scientific research inadvertently contributes either to inequities between groups or to the abuse of human rights. Disagreements did arise during these discussions. For example, biomedical scientists tended to accept that such labels could be used as neutral descriptors of groups of individuals, whereas scholars in the social sciences and humanities tended to question whether such labels could be stripped of embedded sociohistorical meaning. However, dialog and the discovery of language that worked across disciplinary boundaries enabled us to clarify our perspectives and find many points of agreement. This workshop statement constitutes one step in an ongoing, open dialog that takes into account the potential for misinterpretation or misuse of research in human genetic variation. More specifically, this statement looks to shape future use of categories of race and ethnicity in biomedical research.

Acknowledgements

We would like to acknowledge the tremendous intellectual contribution of George Fredrickson, late Professor Emeritus of History at Stanford University, to the creation of this statement. His life-long commitment to the study of racism was critical to the workshop discussion and our understanding of processes of racialization over time. Joanna Mountain and Barbara A Koenig were faculty members at Stanford University during the course of this dialog and were co-organizers of the initial workshop. This work was supported by the National Institutes of Health (career development award K01 HL72465 to SL).

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