

**NATIONAL HUMAN GENOME CENTER  
HOWARD UNIVERSITY**

**POSITION PAPER**

**STATE OF THE SCIENCE ON HUMAN GENOME VARIATION AND “RACE”**

Recent human genome research and studies of human genetic variation have exacerbated a long-standing debate about the biological meaning and importance of the concept of “race” and groups traditionally called “races” in science and medicine. The resultant conundrum regarding the ethics and science of research into “group-limited” or “group-specific” human genetic variation has substantial implications for the conduct of biomedical research -- health disparities research in particular, the practice of medicine, and the well-being of humanity as a whole.

The National Human Genome Center at Howard University maintains that attempts to assess and address the broader ethical and societal issues must be preceded by examination of some fundamental questions such as: Do any set of human groups merit the term “race” based on the criteria of modern molecular systematics? How does human genetic variation distribute across geographically, socioculturally or sociohistorically defined groups or populations? What are the implications of the distribution of this variation for understanding health and disease? What is the relationship between health disparities among groups and group differences in social and physical environments and human genetic variation?

In an effort to comprehensively evaluate the state of the science on human genome variation and “race”, we examined our own research, reviewed existing relevant literature, convened a meeting with experts in the field, and assessed the updated perspectives and commentaries in the November 2004 Nature Genetics special issue. We affirm that human genome variation research must proceed, albeit with greater precision, as it is a critical component of the holistic approach needed to prevent disease and improve health and well-being.

This position paper outlines pertinent scientific findings from the fields of anthropology, human genetics, sociology, and epidemiology; our conclusions based on those findings; implications for science, medicine, and society; and our recommendations for moving forward.

**Findings**

1. The biological variation of the human species exhibits gradients of differentiation, not subdivisions into homogeneous, discontinuous units.
2. The within- versus the between-group variation is greater in the so-called “races” of traditional anthropology (e.g.s. Mongoloid, Australoid, Caucasoid, Negroid). Therefore the majority of variation within the human species is found between individuals and not groups.

3. The ancestry of maternal (mtDNA) and paternal (Y chromosome DNA) lineages crosscut the group (“racial”) boundaries suggested by anatomical traits like skin color and hair form.
4. Combinations of physical traits used to define groups are not obligatorily linked to each other or to health, disease, or other unobservable traits.
5. The demographic units of human societies (and of the U.S. census) are the products of social, cultural, or political rules, not the forces of biological evolution.

## **Conclusions**

1. Modern extant humans do not fracture into races (subspecies) based on the modern phylogenetic criteria of molecular systematics. Thus,
  - a. The social and demographic units of human societies are not races, and
  - b. The racial taxa (groups) of classical anthropology are not races.
2. The biological “boundaries” between any human divisions (groups, populations, nationalities) are arbitrary and largely dependent on what traits are chosen for emphasis.
3. Group differences in health parameters have not been shown to be encoded in the human genome as part of an evolutionary pattern of divergence of humans.
4. Gene-environment interactions may be more important in explaining group differences in health and disease.

## **Implications**

1. The non-existence of human races (subspecies) does not mean the non-existence of racism - the structured systematic oppression of groups defined by ancestry or physical traits and/or conceptualized as being fundamentally different. Racism must be addressed, especially as it relates to health and disease.
2. Individuals cannot be treated as representative for all those who physically resemble them, or have some degree of shared continental ancestry, or are placed in the same group.
3. Group differences in health or disease cannot be understood as solely related to membership in genetically-defined groups.
4. There is a need for a paradigm shift in the conceptualization and application of human genetic variation relative to our understanding of group definition and health outcomes. This shift will require new approaches to methodology, training, policy, and priorities.

## Recommendations

1. Training of a new generation of biomedical scientists and reorientation of established scientists toward the development and implementation of better-designed, interdisciplinary studies that take into account knowledge of population history and migration, genetic variation, disease incidence and prevalence, as well as culture and various other social and psychosocial factors. Messages ought to include the following:
  - a. Research design should focus on local groups in their social and political environments.
  - b. Ancestries of individuals and groups should be incorporated into research to better evaluate inherited genetic effects.
  - c. Inquiry into gene-environment interactions and their impact on physiology should dominate research on disease causation, especially research on health disparities among groups.
  - d. Conclusions based on one community of a socially- or census-designated group should not be automatically extrapolated to other such communities.
2. Funding agencies should increase acceptance of, and allocate adequate funding for more nuanced studies that integrate multiple factors impacting human biology, including genetic variation, cultural practices, socioeconomic and physical environmental factors, and access to healthcare.
3. The development of drugs and other therapeutic strategies should be based on mechanism of disease and not on group membership. For example, the subset of individuals who will benefit from a drug should not be defined by labels such as “African American” or “Hispanic” or “Asian”, since these large human population groups do not consist of biologically identical individuals.
4. Similarly, clinicians must evaluate and treat patients as individuals with full consideration for their present and historic environment, ancestry, family medical history and other relevant health parameters.

Given the overall societal impact of research in this arena and the need for clarity, it is imperative that scientists communicate their findings to “community-based groups” to connect the “grass roots” of society to the mainstream press.

Scientists must communicate their findings outside of the scientific establishment with the cooperation of a responsible media and science press to accurately inform policy, ethics, attitudes, etc.