

Advancing Health Equity Requires Examining the History of Genetics

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November 10, 2022



National Human Genome
Research Institute

—
The **Forefront**
of **Genomics**
—

Advancing Health Equity

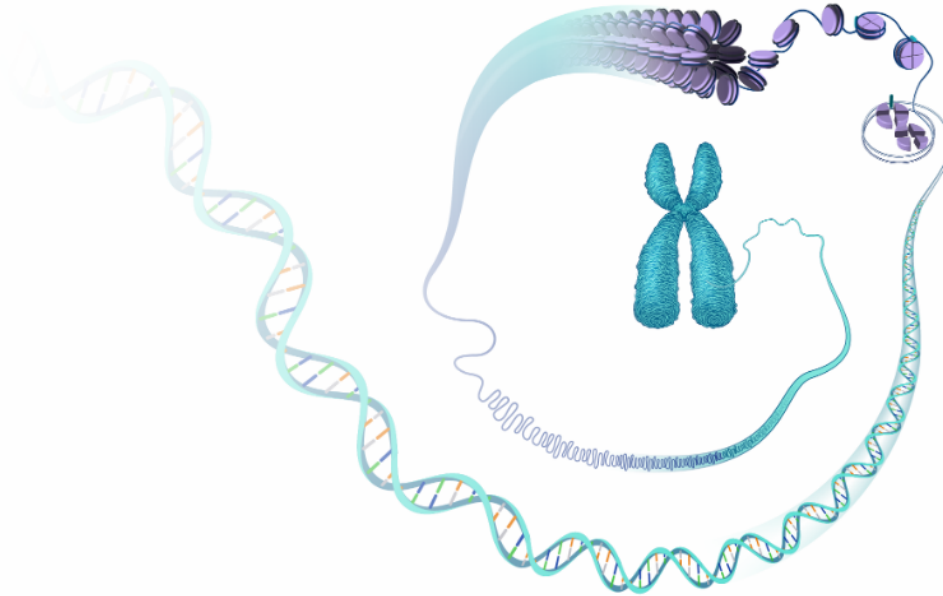
- When every person has the opportunity to attain their full health potential and no one is disadvantaged from achieving this potential because of social position or other socially determined circumstances
- Health equity through a genomics lens:
 - Access to high quality and comprehensive genomic information
 - Development of accessible technology and methods
 - Access to genetic and genomic testing
 - Quality of treatment and management of genetic and genomic findings
 - Acceptability of genetic and genomic approaches and interventions

Reckoning with the History of Genetics



EUGENICS

updated: May 10, 2022



Definition



Eugenics is a discredited belief that selective breeding for certain inherited human traits can improve the “fitness” of future generations. For eugenicists, “fitness” corresponded to a narrow view of humanity and society that developed directly from the ideologies and practices of scientific racism, colonialism, ableism and imperialism.

<https://www.genome.gov/genetics-glossary/Eugenics>

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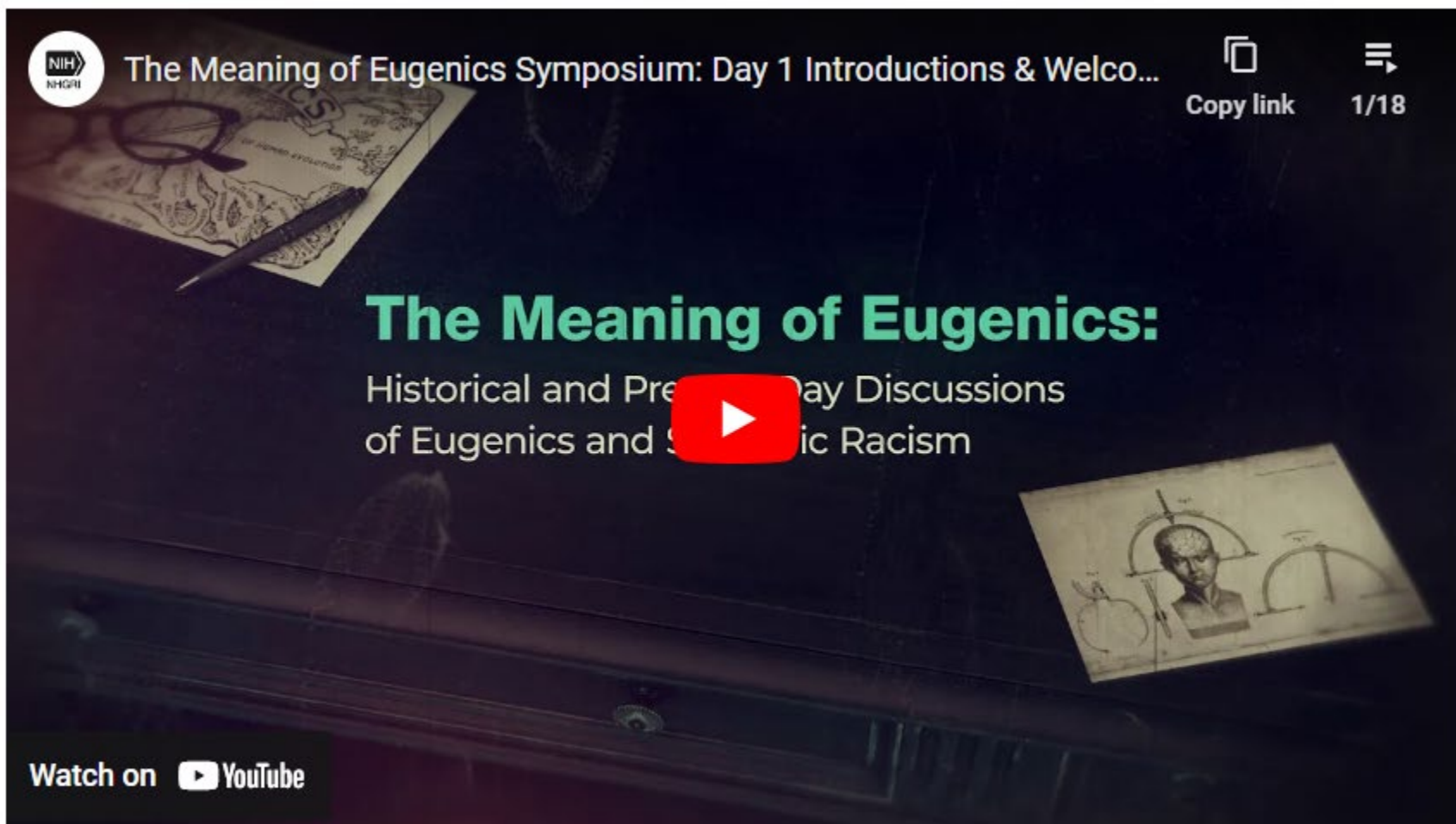
The Meaning of Eugenics: Historical and Present-Day Discussions of Eugenics and Scientific Racism



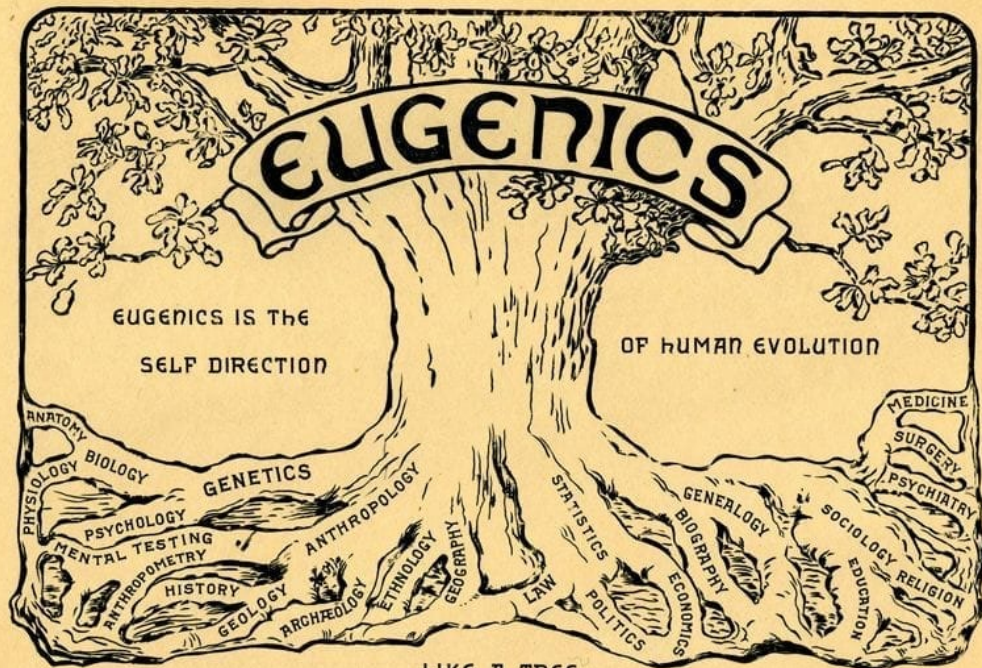
Event Details

Eugenics and scientific racism are widely misunderstood despite their long histories. Studying and sequencing the human genome were supposed to help eliminate common misconceptions about the biological differences between humans. After all, we are 99.9% the same according to our DNA.

And yet, why do these misconceptions continue to persist, resulting in modern day discrimination and bias? We look to the history of science and medicine to help explain.

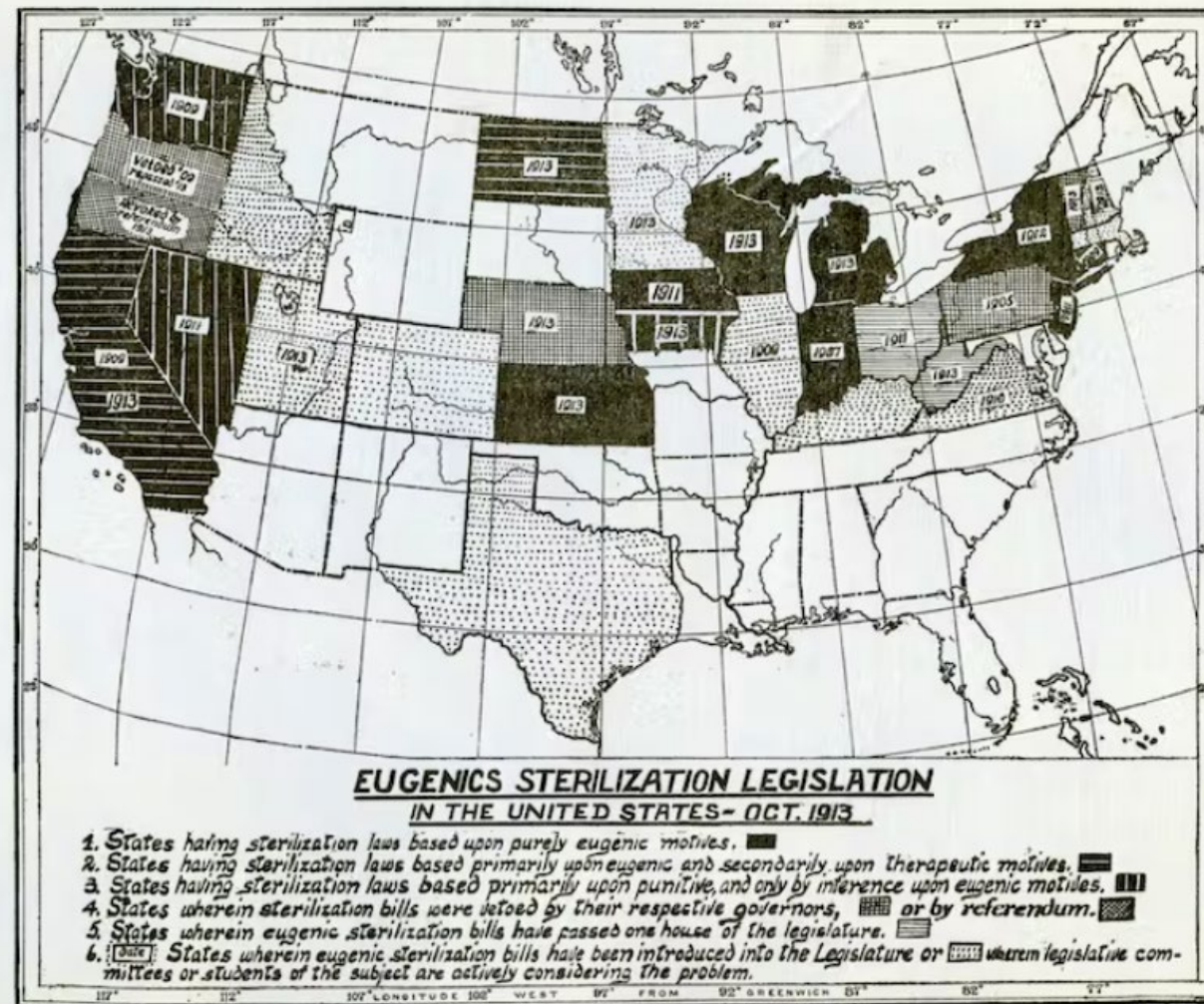


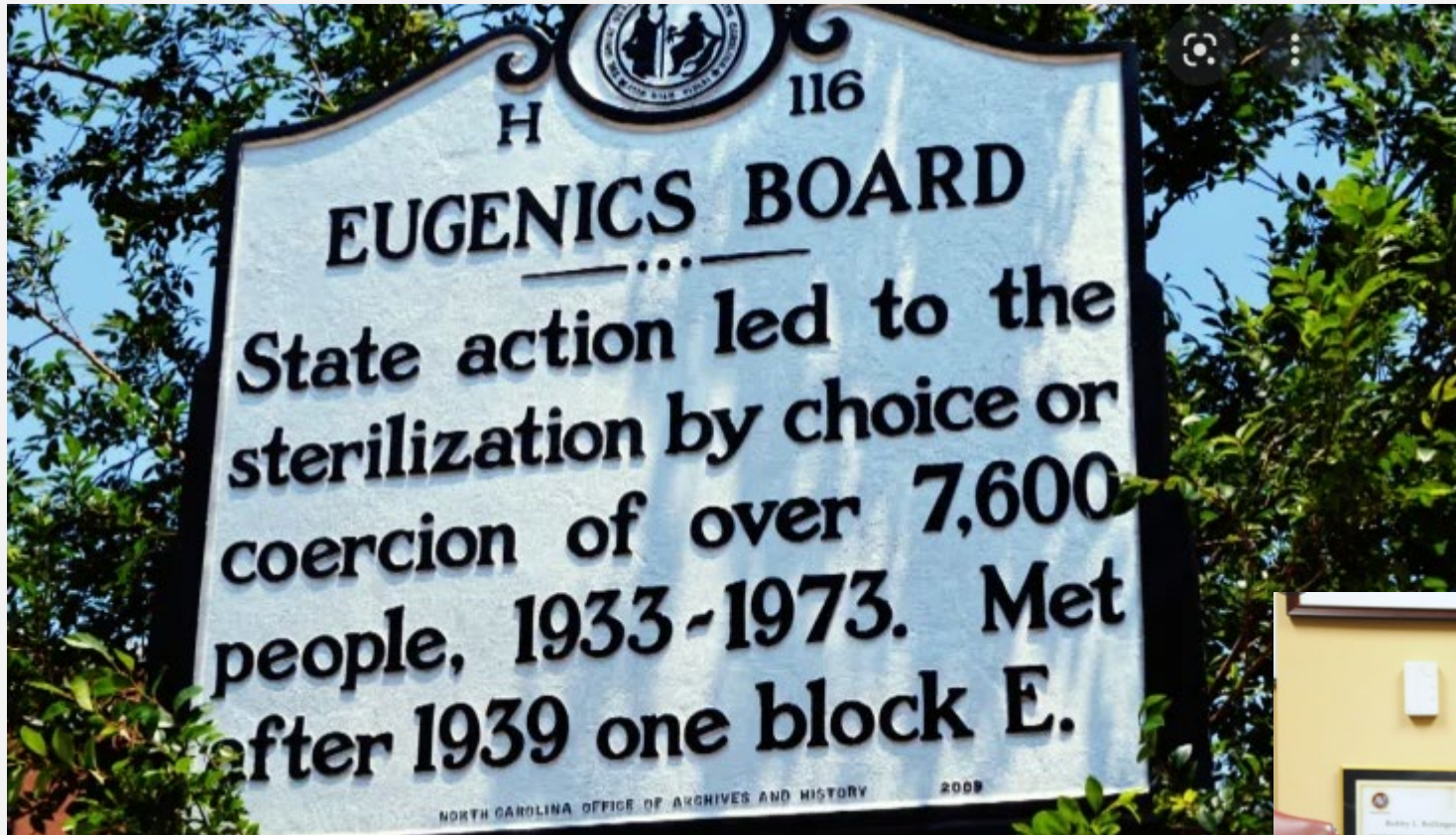
[View on YouTube](#)



LIKE A TREE

EUGENICS DRAWS ITS MATERIALS FROM MANY SOURCES AND ORGANIZES THEM INTO AN HARMONIOUS ENTITY.





North Carolina

<https://www.npr.org/sections/health-shots/2014/10/31/360355784/payments-start-for-n-c-eugenics-victims-but-many-wont-qualify>



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California

“California is committed to confronting this dark chapter in the state’s past and addressing the impacts of this shameful history still being felt by Californians today,” said Governor Newsom. “While we can never fully make amends for what they’ve endured, the state will do all it can to ensure survivors of wrongful sterilization receive compensation.”

California Launches Program to Compensate Survivors of State-Sponsored Sterilization

Published: Dec 31, 2021

New law creates Forced or Involuntary Sterilization Compensation Program

HISTORY

California Once Targeted Latinas for Forced Sterilization

In the 20th century, U.S. eugenics programs rendered tens of thousands of people infertile

Nicole L. Novak and Natalie Lira, *The Conversation*

March 22, 2018

Los Angeles Times

CALIFORNIA
California’s central role in the eugenics movement

Irreducible Subjects: Disability and Genomics in the Past, Present and Future

Event Details

The National Human Genome Research Institute (NHGRI) and The State University of New York at Buffalo Center for Disability Studies will hold a two-day symposium entitled, "Irreducible Subjects: Disability and Genomics in the Past, Present and Future."

<https://www.genome.gov/event-calendar/irreducible-subjects-disability-and-genomics-in-the-past-present-and-future#slides>



SCIENTIFIC RACISM

updated: May 10, 2022



Definition

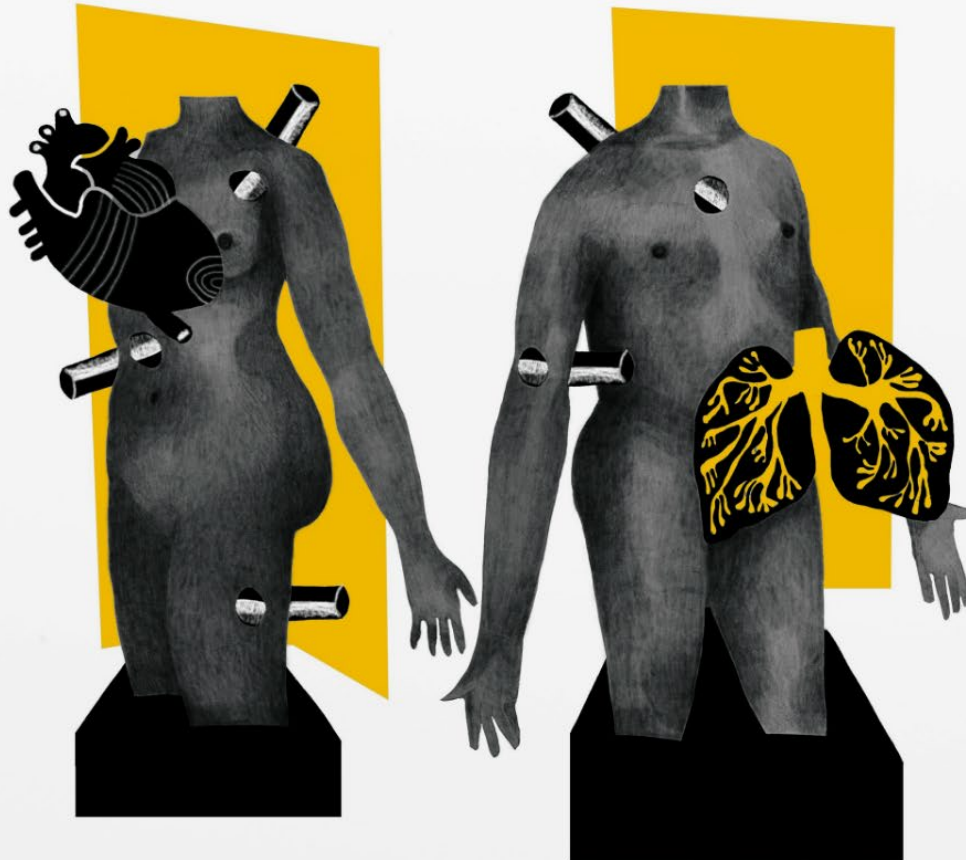


Scientific racism. Scientific racism is a historical pattern of ideologies that generate pseudo-scientific racist beliefs. That perpetually influences racial bias and discrimination in science and research. Leading scientists across scientific institutions in the 19th and early 20th centuries were proponents of such ideologies. By the mid-20th century, pseudo-scientific racist beliefs were widely disproven. However, evidence shows that scientific racism persists in science and research.

Scientific racism

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Myths about physical racial differences were used to justify slavery — and are still believed by doctors today.

By Linda Villarosa

AUG. 14, 2019

Using race and ethnicity requires historical awareness

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I think that such a study as you have contemplated would be of immense value. It will be necessary of course in the consideration of the results to evaluate the special factors introduced by a selection of the material from negro males. Syphilis in the negro is in many respects almost a different disease from syphilis in the white.³³

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Racialization of Disease



INCURABLE 'NEGRO DISEASE' STRIKES FIVE IN FAMILY

Problems linked to sickle cell anemia cloud life in upper New York home

MOST parents expect to encounter a fair share of problems as well as pleasure in the rearing of their children, but fate has weighted the scales heavily on the negative side for Hamp Johnson and his wife, Alice, of Saratoga Springs, N. Y.

Through ten years of hardship and deprivation, they have looked on helplessly while two of their six children have wasted away from the debilitating effects of sickle cell anemia, an incurable, hereditary blood disease predominant in Negro youths. Then, too, they are constantly plagued by fears that the malady will develop into its active state in three other younger children who bear the sickle cell trait. All this has had a tragic impact on the total pattern of life for the Johnson family. Medical bills accumulated during recurring periods when the children were ill have left them heavily in debt, and they are shunned by many neighbors who do not realize that the disease is not contagious.

Their only comfort is in knowing that they are far from being alone in this unfortunate situation, for sickle cell anemia is more common in some parts of the country than all other primary blood diseases put together, including leukemia, hemophilia and platelet diseases. It is believed to cause more paralysis than polio, can affect any organ of the body and is a major cause of maternal mortality. Though it has been



"House for Sale" sign marks home of Hamp Johnson family in Saratoga Springs, N. Y. It is built over shanty and dampness is bad for ailing children. Standing on stoop are (l. to r.), Jeffrey, Percy and Hamp Johnson Jr.

found occasionally in whites of the Mediterranean type, approximately eight to nine per cent of the Negro population carries the benign sickle cell trait, while the disease, fortunately, occurs much less frequently—in about one out of every 400 American Negroes. Caused by presence of the abnormal hemoglobin S in the red blood cells, which results in their collapsing and assuming a crescent or sickle shape, sickle cell anemia usually is manifest in its victims before they reach the age of four. Then they might become pale, listless, have a fever and complain of abdominal, thoracic and particularly skeletal pain. Crises, or periods of extreme illness, are followed by remissions when the victims appear to be fairly well. But they remain highly susceptible to infections and serious conditions like pneumonia and tuberculosis, which frequently result in early deaths.

These are the grim truths that haunt the Johnson family. There is a story that could be duplicated in hundreds of other homes throughout the country.

Marleen Ann Johnson, 12, was first stricken when she was three years old. Within a few days, Percy Lee Johnson, her older brother by one year, also fell ill. At first their conditions were diagnosed as leukemia and Marleen was given only months to live. But subsequently, doctors

*Ebony Magazine, May
1966*

Racialization of Sickle cell

“up to the present, the malady has been seen only in the Negro [sic], and so far as could be ascertained, it is the only disease peculiar to that race”

Mason VR. Landmark article Oct. 14, 1922: Sickle cell anemia. By V.R.

Mason. *Jama* 1985; 254(14):1955-7

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“The most significant feature of sickle cell anemia is the fact that it is apparently the only known disease that is completely confined to a single race”

Annon, Sickle Cell Anemia: A Race Specific Disease. JAMA 1947; 133:33

Race, Ethnicity, Ancestry in the Era of Genomics



MOISES VELASQUEZ-MANOFF

What Doctors Should Ignore

Science has revealed how arbitrary racial categories are. Perhaps medicine will abandon them, too.

How Genetics Is Changing Our Understanding of 'Race'



Is direct-to-consumer genetic testing reifying race?

How to Talk About 'Race' and Genetics

Opinion

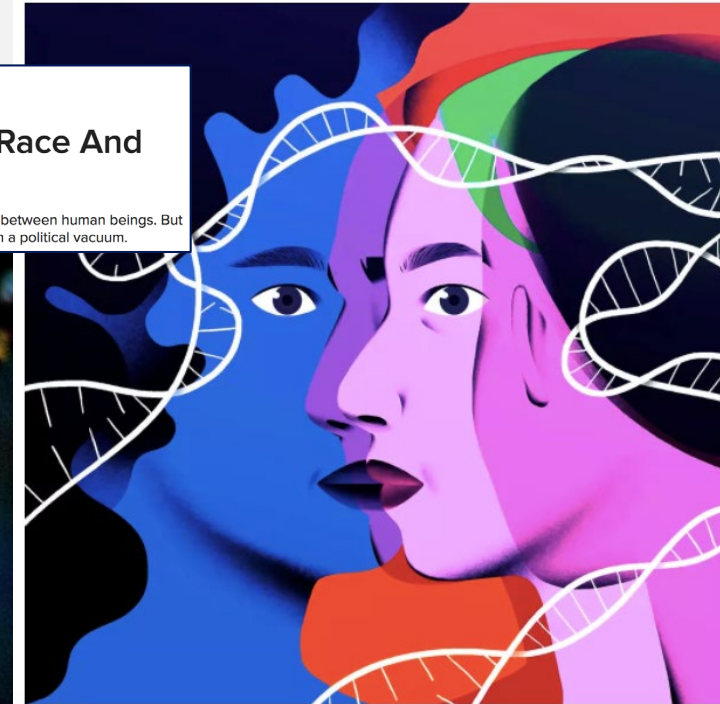
How Not To Talk About Race And Genetics

Race has long been a potent way of defining differences between human beings. But science and the categories it constructs do not operate in a political vacuum.

THE RACE ISSUE

There's No Scientific Basis for Race—It's a Made-Up Label

It's been used to define and separate people for millennia. But the concept of race is not grounded in genetics.

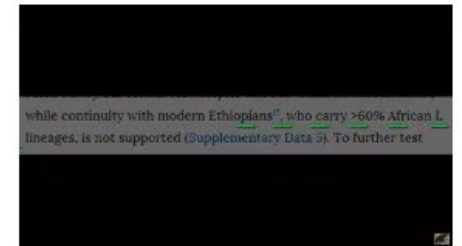
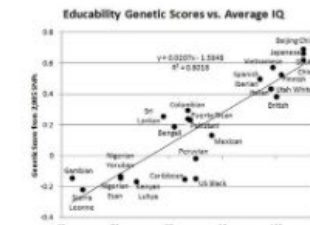


How Genetics Is Changing Our Understanding of 'Race'

David Reich, New York Times March 23, 2018



Why White Supremacists Are Chugging Milk (and Why Geneticists Are Alarmed)



"Race is a social construct"
LOL
Does anyone believe this nonsense?
The truth: Race is biologically real.

The Cultural Marxist War against Darwinism

Creationists: evolution is a social construct, not biologically real.
Liberal Creationists: race is a social construct not biologically real.

Charles Darwin: I'm not a creationist; I actually wrote: "There is, however, no doubt that various races, when carefully compared and measured, differ much from each other..."



No Credible Scientists Believe in Race

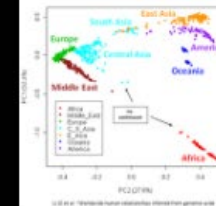
Some people feel that they do not have the needed expertise to judge the validity of race. So, they defer to the experts, and the experts tell them that race does not exist. The problem with this argument is, even though the most vocal anthropologists and biologists deny race, academic surveys show that there is no actual consensus on this topic.

Year	Race-neutral description of race	Race-based description of race
1952-1962	90%	0%
1963-1972	80%	18%
1973-1982	89%	9%
1983-1992	21%	40%
1993-2002	42%	32%

storing jobs

Journal Type	Articles That Group People Racially	Articles That Don't Group People Racially
American Physical Anthropology Journals	40%	60%

Myth #1: "Race has no biological basis"



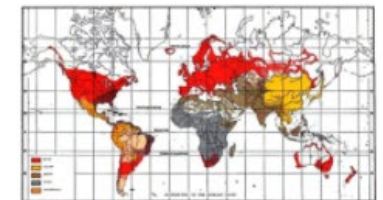
Fact: There are very accurate ways to determine someone's ancestry based on DNA such as principal component analysis.

When you sample many individuals across the globe and map them, you notice an overall clustering pattern where you can identify populations and races.

This clustering is a natural consequence of divergent evolution due to geographical isolation and differing environmental pressures that *Horrea* sapiens encountered since migrations took place.

Africans are consistently separated from the rest and this is due to the fact that they have been evolving separately for more than 40,000 years and unlike the rest, they don't have any Neanderthal admixture.

Lothrop Stoddard World Race Map (1920):



Today's proponents of racial hierarchy are making their case by misinterpreting research on the human genome itself.

By Amy Harmon

Oct. 17, 2018



What Happens When Geneticists Talk Sloppily About Race

The field widely agrees that race is a social construct, but gets into trouble when it ignores semantics.

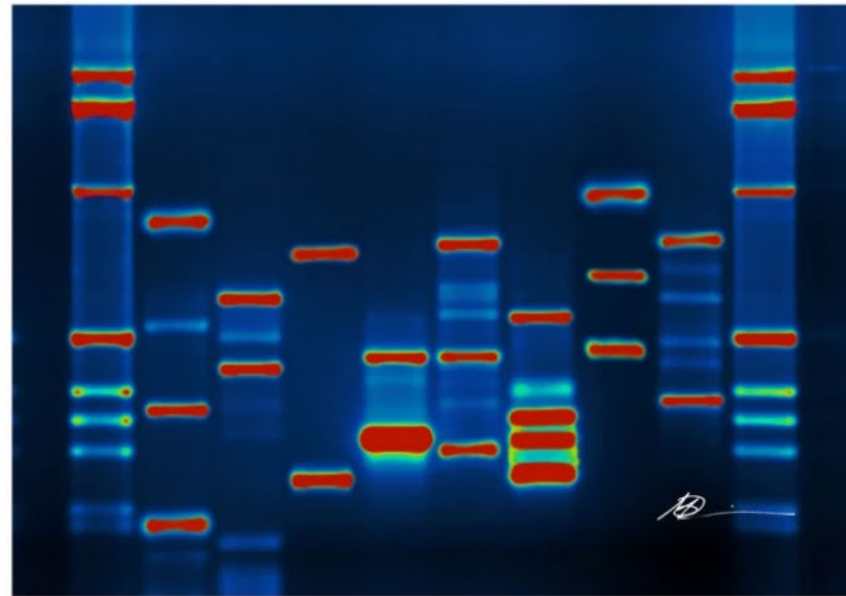
IAN HOLMES | APR 25, 2018 | **SCIENCE**

Opinion

How Not To Talk About Race And Genetics

Race has long been a potent way of defining differences between human beings. But science and the categories it constructs do not operate in a political vacuum.

Posted on March 30, 2018, at 5:29 p.m.



Micah Baldwin / Via Flickr: micahb37

This open letter was produced by a group of 67 scientists and researchers. The full list of signatories can be found below.

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“ Precisely because the problems of race are complex, scientists need to engage these issues with greater care and sophistication. Geneticists should work in collaboration with their social science and humanities colleagues to make certain that their biomedical discoveries make a positive difference in health care, including the care of those studied.”

NASEM Consensus Study

*The National
Academies of* SCIENCES
ENGINEERING
MEDICINE



- 14 NIH Institutes, Centers, Offices, & Programs co-sponsoring study (NHGRI as co-lead)
- Review existing methodologies, benefits, and challenges in the use of race and ethnicity and other population descriptors in genomics research
- Study committee announced on February 2
- Three public meetings planned
- Final report in February 2023

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Trust and Trustworthiness



Trust

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“Trust remains the cornerstone of the patient-provider relationship. The importance of this bond cannot be understated; it often predicts the willingness of a patient to seek care, share confidential information, comply with treatment regimens, and participate in biomedical research.”



- Persaud A, Bonham VL. The Role of the Health Care Provider in Building Trust Between Patients and Precision Medicine Research Programs. Am J Bioeth. 2018 Apr;18(4):26-28.

Trustworthiness

“Trustworthiness, a commonly recognized antecedent to trust, can be described as the perception of probabilities, or expectation, that a trusting relationship will result in gains and/or losses from engaging in an encounter that requires trust (i.e., risk/uncertainty and interdependence). Trustworthiness begets trust by giving patients a reason to trust.”

Trustworthiness

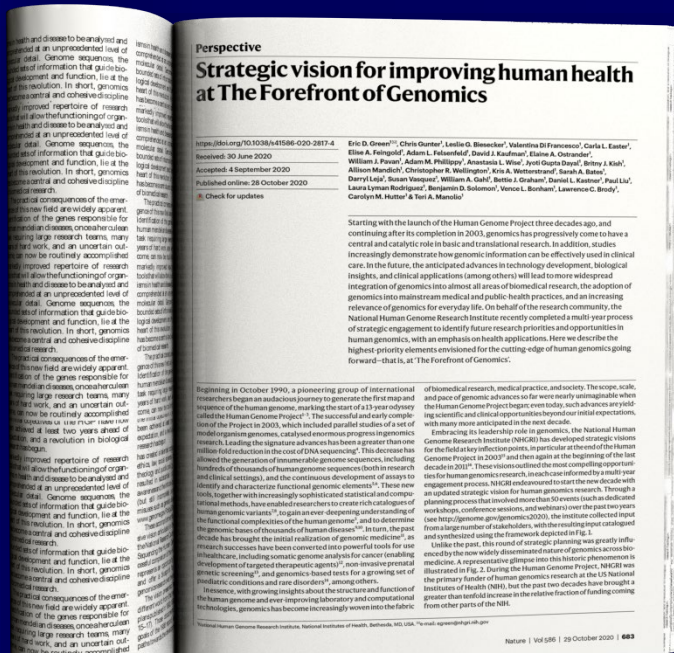
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“While physicians carry incredible potential to serve as agents and mediators of trust between patients and research institutions, there are potential impediments that may undermine this process if not considered carefully.”

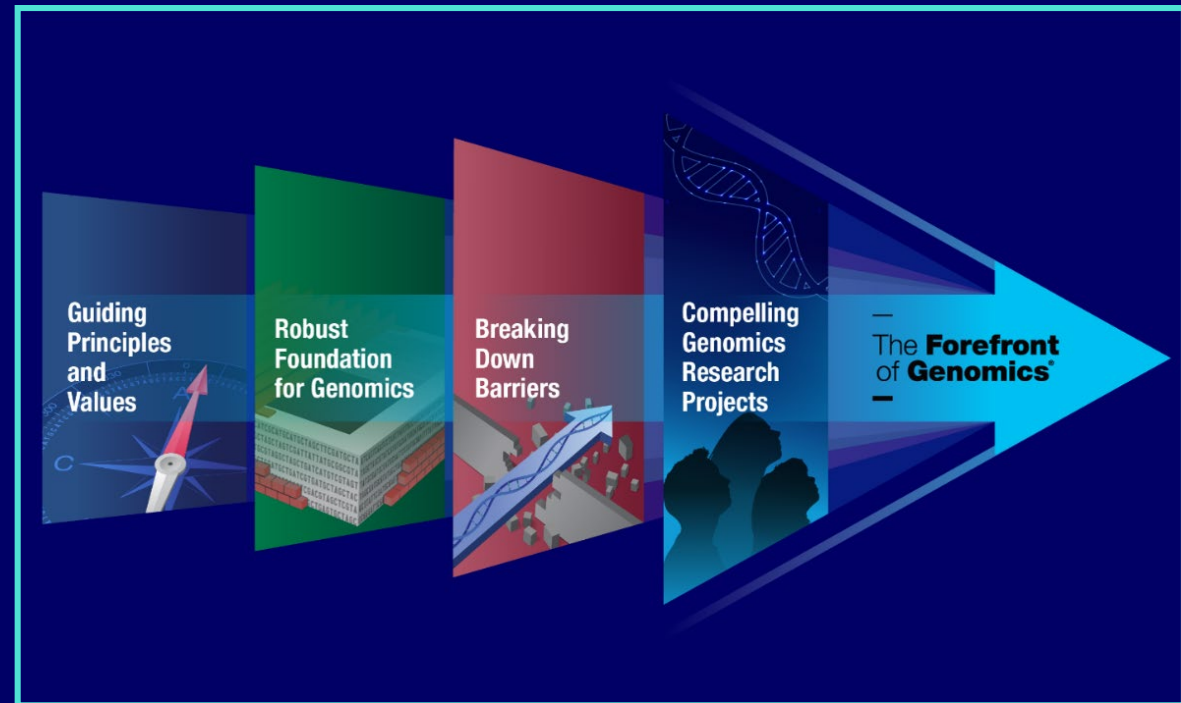
Persaud A, Bonham VL. The Role of the Health Care Provider in Building Trust Between Patients and Precision Medicine Research Programs. Am J Bioeth. 2018 Apr;18(4):26-28.

BIG QUESTION:

How do we enhance health equity in genetics and genomics?



Green et al. (2020)



Box 1

• **Champion a diverse genomics workforce** — the promise of genomics cannot be fully achieved without attracting, developing, and retaining a diverse workforce, which includes individuals from groups that are currently underrepresented in the genomics enterprise.

and underrepresented individuals in major genomic studies

— attention to diversity in genomics research is both socially just and scientifically essential, which includes meaningful, sustained partnerships with diverse communities in the design and implementation of research studies, the propagation of research findings, and the development and use of new technologies.

• **Maximize the usability of genomics for all members of the public, including the ability to access genomics in healthcare**

— engagement, inclusion, and understanding the needs of diverse and medically underserved groups are required to ensure that all members of society benefit equitably from genomic advances, with particular attention given to the equitable use of genomics in healthcare that avoids exacerbating and strives towards reducing health disparities.

• **Champion a diverse genomics workforce** — the promise of genomics cannot be fully achieved without attracting, developing, and retaining a diverse workforce, which includes individuals from groups that are currently underrepresented in the genomics enterprise.

• **Provide a conceptual research framing that consistently examines the role of both genomic and non-genomic contributors to health and disease** — routinely considering the

— the use of carefully defined standards (for example, those for generating, analysing, storing, and sharing data) has benefited genomics in numerous ways, and this must include appropriate privacy and data-security protections for those participating in genomics research.

• **Embrace the interdisciplinary and team-oriented nature of genomics research** — starting with the Human Genome Project, some of the most challenging genomics endeavours have benefited from the creation and management of large, interdisciplinary research collaborations.

• **Adhere to the highest expectations and requirements related to open science, responsible data sharing, and rigor and reproducibility in genomics research** — the genomics enterprise has a well-respected history of leading in these areas, and that commitment must be built upon and continually reaffirmed.

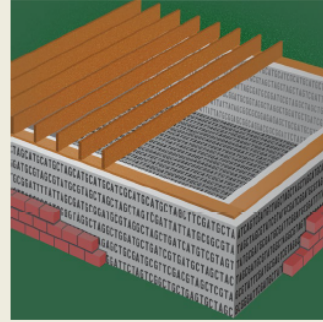
• **Pursue advances in genomics as part of a vibrant global community of genomics researchers and funders** — the challenges in genomics require the collective energies and creativity of a collaborative international ecosystem that includes partnerships among researchers, funders, and other stakeholders from academia, government, and the commercial sector.

Box 2

Sustaining and improving a robust foundation for genomics

Genome structure and function

- Enable the routine generation and analysis of increasingly complex genomic data
- Use evolutionary and comparative genomic data to maximize understanding of genome function



Genomic data science

- Develop new methods and build sustainable data resources for genomics research
- Ensure facile storing, sharing, and computing on large genomic datasets
- Develop integrated knowledgebases and informatics methods for genomic medicine

Genomics and society

- Understand the interrelationships between genomics and the social and environmental factors that influence human health
- Empower people to make well-informed decisions about genomic data and develop data-stewardship systems that reinforce their choices
- Increase the genomic literacy of all sectors of society

Training and genomics workforce development

- Ensure that the next generation of genomic scientists are sufficiently trained in data science
- Train healthcare providers to integrate genomics into the clinical workflow
- Foster a diverse genomics workforce

In both research and clinical settings, the global genomics workforce—as with the general biomedical research workforce—falls considerably short of reflecting the diversity of the world's population (a vivid example of this is seen in the United States⁷²), which limits the opportunity of those systematically excluded to bring their unique ideas to scientific and clinical research⁷³. To attain a diverse genomics workforce, new strategies and programs to reduce impediments to career opportunities in genomics are required, as are creative approaches to promote workforce diversity, leadership in the field, and inclusion practices. Efforts must intentionally include women, underrepresented racial and ethnic groups, disadvantaged populations, and individuals with disabilities. Initiatives should not focus exclusively on early-stage recruitment; instead, they must also include incentives to recruit and retain a diverse workforce at all career stages⁷⁴ as well as new approaches for cultivating the next generation of genomics practitioners.



Building a Diverse Genomics Workforce: An NHGRI Action Agenda

The **Forefront**
of **Genomics**

The genomics workforce must become more diverse: a strategic imperative

Vence L. Bonham^{1,*} and Eric D. Green^{1,*}

American Journal of Human Genetics 108(1):3-7 (2021)

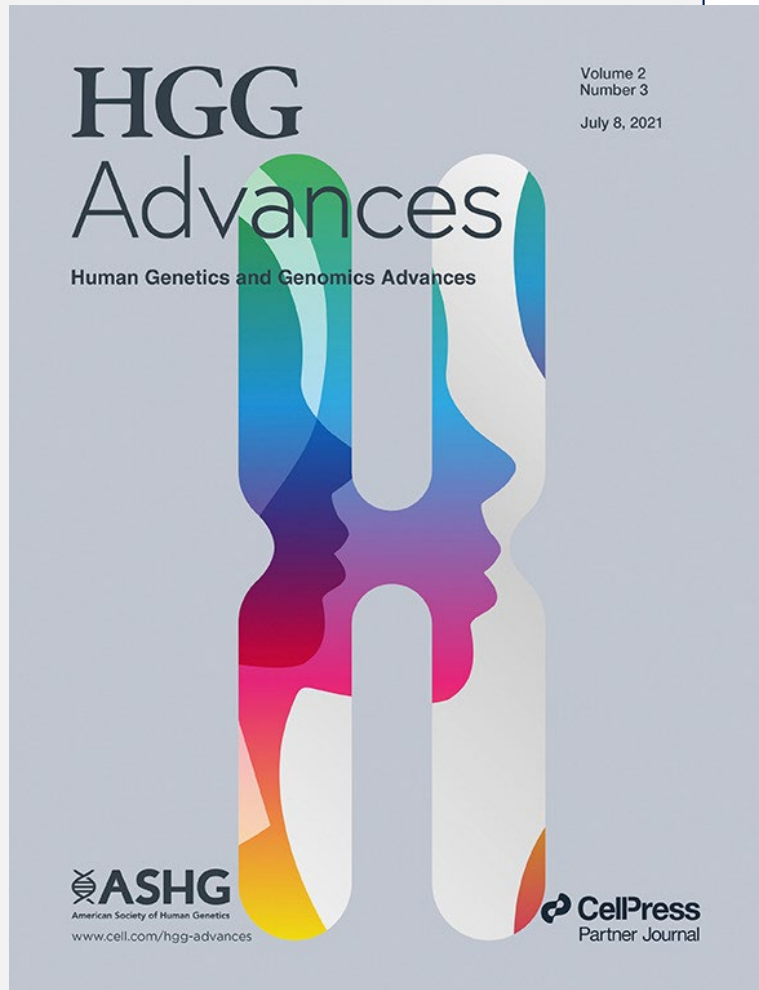


genome.gov/workforcediversity

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Cultivating Diversity as an Ethos



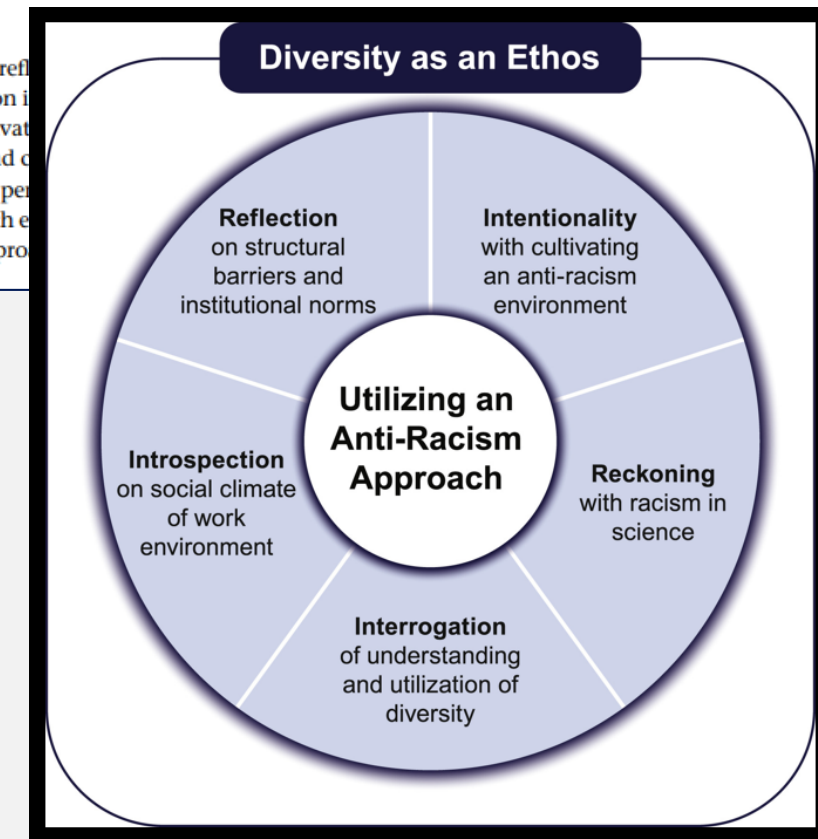


Cultivating diversity as an ethos with an anti-racism approach in the scientific enterprise

Shameka P. Thomas,^{1,*} Kiana Amini,¹ K. Jameson Floyd,¹ Rachele Willard,¹ Faeben Wossenseged,¹ Madison Keller,¹ Jamil B. Scott,² Khadijah E. Abdallah,¹ Ashley Buscetta,¹ and Vence L. Bonham^{1,*}

Summary

The diversity of the U.S. population is currently not reflected in the scientific fields. Although diversity and inclusion efforts have focused on individual scientists, structural racism remains. Thus, the cultivation of an intentional about an institution's character, culture, and climate is an anti-racism approach within the field. Adopting a new perspective for researchers as we build supportive, collaborative research environments in the research enterprise and propose an anti-racism approach.



Cultivating diversity as an ethos with an anti-racism approach in the scientific enterprise

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“The cultivation and adoption of diversity as an ethos requires shifting our focus to being intentional about an institution’s character, culture, and climate. One way for this ethos to be sustained is by facilitating an intentional anti-racism approach within the field.”

Thomas SP, Amini K, Floyd KJ,... Bonham VL. Cultivating diversity as an ethos with an anti-racism approach in the scientific enterprise. HGG Adv. 2021 Sep 21;2(4):100052.

CRISPR-Cas9



Thank You
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