"Advancing the Equitable Implementation of Genomics into Clinical Care"

February 16, 2022 ISCC-PEG Vence L Bonham, Jr. J.D. Acting Deputy Director National Human Genome Research Institute



2020 NHGRI Strategic Vision



Perspective

Strategic vision for improving human health at The Forefront of Genomics

https://dol.org/10.1038/s41586-020-2817-4	Eric D. Green ¹ [™] , Chris Gunter ¹ , Leslie G. Biesecker ¹ , Valentina Di Francesco ¹ , Carla L. Easter ¹ ,
Received: 30 June 2020	Elise A. Feingold ¹ , Adam L. Felsenfeld ¹ , David J. Kaufman ¹ , Elaine A. Ostrander ¹ , William J. Pavan ¹ , Adam M. Phillippy ¹ , Anastasia L. Wise ¹ , Jyoti Gupta Daval ¹ , Britny J. Kish ¹ .
Accepted: 4 September 2020	Allison Mandich ¹ , Christopher R. Wellington ¹ , Kris A. Wetterstrand ¹ , Sarah A. Bates ¹ ,
Published online: 28 October 2020	Darryl Leja', Susan Vasquez', William A. Gahl', Bettie J. Graham', Daniel L. Kastner', Paul Liu', Laura Lyman Rodriguez', Benjamin D. Solomon', Vence L. Bonham', Lawrence C. Brody'.
Check for updates	Carolyn M. Hutter' & Terl A. Manolio'
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Starting with the launch of the Human Genome Project three decades ago, and continuing after its completion in 2003, genomics has progressively come to have a central and catalytic role in basic and translational research. In addition, studies increasingly demonstrate how genomic information can be effectively used in clinical care. In the future, the anticipated advances in technology development, biological insights, and clinical applications (among others) will lead to more widespread integration of genomics into almost all areas of biomedical research, the adoption of genomics into mainstream medical and public-health practices, and an increasing relevance of genomics for everyday life. On behalf of the research community, the National Human Genome Research Institute recently completed a multi-year process of strategic engagement to identify future research priorities and opportunities in human genomics, with an emphasis on health applications. Here we describe the highest-priority elements envisioned for the cutting-edge of human genomics going forward-that is, at 'The Forefront of Genomics'.

Beginning in October 1990, a pioneering group of international of biomedical research, medical practice, and society. The scope, scale researchers began an audacious journey to generate the first map and and pace of genomic advances so far were nearly unimaginable when sequence of the human genome, marking the start of a 13-year odyssey the Human Genome Project began; even today, such advances are yieldcalled the Human Genome Project¹⁻³. The successful and early comple-ing scientific and clinical opportunities beyond our initial expectations, tion of the Project in 2003, which included parallel studies of a set of with many more anticipated in the next decade. model organism genomes, catalysed enormous progress in genomics research. Leading the signature advances has been a greater than one Genome Research Institute (NHGRI) has developed strategic visions million-fold reduction in the cost of DNA sequencing⁴. This decrease has for the field at key inflection points, in particular at the end of the Human allowed the generation of innumerable genome sequences, including hundreds of thousands of human genome sequences (both in research decade in 2011¹⁶. These visions outlined the most compelling opportuniand clinical settings), and the continuous development of assays to ties for human genomics research, in each case informed by a multi-year identify and characterize functional genomic elements^{5,6}. These new tools, together with increasingly sophisticated statistical and computational methods, have enabled researchers to create rich catalogues of human genomic variants^{7,8}, to gain an ever-deepening understanding of the functional complexities of the human genome⁵, and to determine (see http://genome.gov/genomics2020), the institute collected input the genomic bases of thousands of human diseases^{9,10}. In turn, the past decade has brought the initial realization of genomic medicineⁱⁱ, as research successes have been converted into powerful tools for use in healthcare, including somatic genome analysis for cancer (enabling development of targeted therapeutic agents)12, non-invasive prenatal genetic screening¹³, and genomics-based tests for a growing set of paediatric conditions and rare disorders14, among others. In essence, with growing insights about the structure and function of

the human genome and ever-improving laboratory and computational greater than tenfold increase in the relative fraction of funding coming

nal Human Genome Research Institute, National Institutes of Health, Bethesda, MD, USA. 🕮 - mail: egreen@nhgri.nih.go

Embracing its leadership role in genomics, the National Human

Genome Project in 200315 and then again at the beginning of the last engagement process. NHGRI endeavoured to start the new decade with planning process that involved more than 50 events (such as dedicated workshops, conference sessions, and webinars) over the past two years from a large number of stakeholders, with the resulting input catalogued and synthesized using the framework depicted in Fig. 1.

Unlike the past, this round of strategic planning was greatly influ enced by the now widely disseminated nature of genomics across biomedicine. A representative glimpse into this historic phenomenon is illustrated in Fig. 2. During the Human Genome Project, NHGRI was the primary funder of human genomics research at the US National Institutes of Health (NIH), but the past two decades have brought a technologies, genomics has become increasingly woven into the fabric from other parts of the NIH.

Nature | Vol 586 | 29 October 2020 | 683

The Forefront of **Genomics**®



Green ED., Strategic vision for improving human health at The Forefront of Genomics. Nature 586: 683–692(2020)

Box 1

NIH

NHGRI

Guiding principles and values for human genomics

• Maintain an overarching focus on using genomics to understand biology, to enhance knowledge about disease, and to improve human health — genomics is now foundational across the entire continuum of biomedical research, from deciphering fundamental principles of biology to translating that knowledge into disease prevention and medical advances.

• Strive for global diversity in all aspects of genomics research, committing to the systematic inclusion of ancestrally diverse 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 importance of social and environmental factors that influence human health (and the interactions among those components and genomics) will be important for the comprehensive understanding of most human diseases. • Promote robust and consistently

applied standards in genomics research — 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. 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.



Green ED., Strategic vision for improving human health at The Forefront of Genomics. Nature volume 586, pages683–692(2020)



Health Equity in Genomics

Health Equity

"Health equity work requires an acknowledgment and reconsideration of previously taken for granted beliefs about health (and how it is produced), the health care and public health systems (and how they work), and society (and how it is set up to advantage some and disadvantage others)."



Will Everyone Benefit?

Genomic and Precision Medicine

Diagnostics

Risk Assessment

Pharmacogenomics

New Drugs Genetic Therapies Diverse communities face barriers in accessing genomic medicine



CITIES To inform policy, urban scholarship must get organized and funded p.165	HISTORY A biography of Enrico Fermi, Italy's fallible atomic physicist J.160	championed offer a window on his time p.170	OBITUARY Roger Yonchien Tsien, fluorescent-biology pioneer, remembered 1.172
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Certain drugs may be less effective, or even unsafe, in some populations because of genetic differences.

Genomics is failing on diversity

An analysis by Alice B. Popejoy and Stephanie M. Fullerton indicates that some populations are still being left behind on the road to precision medicine.

2009 analysis revealed that 96% of participants in genome-wide asso-Ciation studies (GWAS) were of European descent¹. Such studies scan the genomes of thousands of people to find variants associated with disease traits. The finding prompted warnings that a much broader range of populations should be investigated² to avoid genomic medicine being of benefit merely to "a privileged few". Seven years on, we've updated that analysis. Our findings indicate that the US National Institutes of Health (NIH) proportion of individuals included in mandated the inclusion of diverse participants in the biomedical research it funds, GWAS who are not of European descent has increased to nearly 20%. Much of this GWAS funded by the NIH and other sources rise, however, is a result of more studies are continuing to miss a vast portion of the being done in Asia on populations of Asian world's genetic variation. ancestry. The degree to which people of African and Latin American ancestry, Hispanic people and indigenous peoples are represented in GWAS has barely shifted.

Over the past decade, GWAS have been the preferred tool for discovering the genetic factors involved in common diseases. Tens of thousands of significant associations between Thus, more than 20 years after the genetic variants and biological traits have 🕨

13 OCTOBER 2016 | VOL 538 | NATURE | 161 © 2016 Macmillan Publishers Limited, part of Springer Nature. All rights reserved.

PERSISTENT BIAS

Over the past seven years, the proportion of participants in genome-wide association studies (GWAS) that are of Asian ancestry has increased. Groups of other ancestries continue to be very poorly represented.



Popejoy, AB, and Fullerton, SM. Genomics is failing on diversity. Nature. 2016 Oct 12;538(7624):161-164



5X increase in non-European samples

78% of this increase from populations with Asian ancestry

All other ancestral populations make up **less than 4%**

Popejoy AB, Fullerton SM, Nature, 538:161-164, October 13,2016

The Missing Diversity in Human Genetics Studies



Sirugo G, Williams SM, Tishkoff SA. The Missing Diversity in Human Genetic Studies. Cell. 2019 May 2;177(4):1080.

Breaking Down Barriers that Impede Progress in Genomics

Leading the Way at NHGRI





PRIMED Polygenic Risk Methods in Diverse Populations





Race in the Clinic

Multiple dimensions of race

Race that others believe you to be be



Race you believe others assume you to be

Roth W.D., The Multiple Dimensions of Race. Ethnic and Racial Studies 39(8): 1310-1338.

Race and ethnicity are complex and fluid

"First, it is essential to point out that 'race' and 'ethnicity' are terms without generally agreed upon definitions. Both terms carry complex connotations that reflect culture, history, socioeconomics and political status, as well as a variably important connection to ancestral geographic origins."

Collins FS, What we do and don't know about 'race', 'ethnicity', genetics and health at the dawn of the genome era, Nature Genetics Supp. November 36;11:S13-S15 (2004)



"Both black and white physicians concluded that the race of the patient is medically relevant but did not agree upon why race is important in clinical decisions"

Bonham VL, Sellers SL, Gallagher TH, Frank D, Odunlami AO, Price EG, and Cooper LA, Genetics in Medicine 2009:11(4):279-286.

Characteristics of Physician Respondents (n=787)



The Meaning of 'Race' and 'Ethnicity'

'Race'





Biological difference between <u>racial</u> groups affect health outcome differences



Race is the <u>best proxy</u> clinicians have to identify genetic effects on health



Genetic Variation Knowledge Assessment Index (GKAI)

ITEM#	QUESTION	[†] ANSWER
GKAI1	The DNA sequences of two randomly selected healthy individuals of the same sex are 90-95% identical.	False (22%)**
GKAI2	Most common diseases, such as diabetes and heart disease, are caused by a single gene variant.	False (80%)
GKAI3*	Common structural genetic variation (changes in the human genome such as deletions, duplications and large-scale copy-number variants) is important in health and disease.	True (90%)
GKAI4	All the genetic variation in an individual can be attributed to either spontaneous (i.e., de novo) or inherited changes in the human genome.	True (60%)
GKAI5*	The variation in the human genome includes both disease-causing gene variants and variants that have no effect on health and disease.	True (92%)
GKAI6	Individual genetic variants are usually highly predictive of the manifestation of common disease.	False (60%)
GKAI7	Prevalence of many Mendelian diseases differs by racial groups.	True (69%)
GKAI8	Self-reported race is informative of a racial group's genetic ancestral background.	True (39%)

Bonham VL, Sellers SL, Woolford S, BMC Health Services Research. 2014; 14:456.

Self reported race is informative of a racial group's genetic ancestral background (True or False)



Racial Attributes in Clinical Evaluation (RACE) Measure

Table 3 Items and standardized factor loadings for the Racial Attributes in Clinical Evaluation (RACE) scale				
ITEM#	QUESTION	LOADINGS		
RACE1	I consider information from patients about their racial background.	.61		
RACE2	I consider my patients race to better understand their genetic predispositions.	.69		
RACE3	I consider my patients race when making decisions about which medications to prescribe.	.74		
RACE4	l consider my patients race in determining genetic risk for common, complex diseases (e.g. kidney disease or diabetes).	.77		
RACE5	I consider my patients race in making medication dosage decisions.	.64		
RACE6	I consider my patients race when determining age of initiation of screening for certain diseases.	.66		
RACE7	I consider my patients race in determining how aggressively to treat particular diseases.	.61		
RACE8*	l consider my patients race in determining genetic risk for single gene conditions (e.g. cystic fibrosis or sickle cell disease).			

*Item not included in final scoring.

RACE measure yield one factor (alpha=.86, 7 items)

Bonham VL, Sellers SL, Woolford S, BMC Health Services Research. 2014; 14:456.

Physicians' Anxiety Due to Clinical Uncertainty & Use of Race

We found that general internists with higher anxiety due to clinical uncertainty report using race in medical decision making at higher levels than those with lower anxiety due to understanding.

Cunningham et al. Medical Care, 2014;52:8728-733

2020-2021

The NEW ENGLAND JOURNAL of MEDICINE

MEDICINE AND SOCIETY

Debra Malina, Ph.D., Editor

Hidden in Plain Sight — Reconsidering the Use of Race Correction in Clinical Algorithms

Darshali A. Vyas, M.D., Leo G. Eisenstein, M.D., and David S. Jones, M.D., Ph.D.

August 27, 2020, Epub June 17, 2020

Table 1. Examples of Race Correction in Clinical Medicine.*					
Table 1. Exa Tool and Clinical Utility		Input Variables		Use of Race	Equity Concern
Tool and Cl Breast Cancer Surveillance Consortiu Cardiology Calculator ¹⁹ (https://tools.bcsc-sc.org/BCSyearRisk/calculator.htm		Age Race/ethnicity: white, black, A Native American, other/mu	Asian, Iltiple	The coefficients rank the race/ethnicity categories in the following descending order of risk: white, American Indian, black Hispanic Asian	Returns lower risk estimates for all nonwhite race/ethnicity categories, potentially reducing the likelihood of close surveillance in these patients
The Americ hroast concer in women with no	previous	RIRADS breast density core			
Estimated glomerular filtration rate (eGFR) MDRD and CKD-EPI equations ¹¹ (https://ukidney.com/nephrology- resources/egfr-calculator) Estimates glomerular filtration rate on the basis of a measurement of serum creatinine	Serum o Age and Race: bl	reatinine sex ack vs. white or other	The MD (by a f identi simila sex (0 The CKE larger study mode 1.159) This c correc	RD equation reports a higher eGFR factor of 1.210) if the patient is fied as black. This adjustment is ar in magnitude to the correction for 0.742 if female). D-EPI equation (which included a number of black patients in the population), proposes a more est race correction (by a factor of if the patient is identified as black. correction is larger than the ction for sex (1.018 if female).	Both equations report higher eGFR values (given the same creatinine measurement) for patients identified as black, suggesting better kidney function. These higher eGFR values may delay referral to specialist care or listing for kidney transplantation.
Organ Procurement and Transplantation Network: Kidney Donor Risk Index (KDRI (https://optn.transplant.hrsa.gov/resource /allocation-calculators/kdpi-calculator/) Estimates predicted risk of donor kidney gra failure, which is used to predict viability of potential kidney donor.†	Age ¹² Hyperte es Serum o Cause o ft accide Donatio Hepatiti Height a HLA ma Cold isc En bloc Double Race: Af	nsion, diabetes reatinine level f death (e.g., cerebrovascular ent) n after cardiac death s C and weight tching hemia transplantation kidney transplantation rican American	Increa graf ider (coo inte hyp (0.1 (0.2	ases the predicted risk of kidney ft failure if the potential donor is ntified as African American efficient, 0.179), a risk adjustment ermediate between those for ertension (0.126) and diabetes 30) and that for elevated creatinine 209–0.220).	Use of this tool may reduce the pool of African-American kidney donors in the United States. Since African-American patients are more likely to receive kidneys from African-American donors, by reducing the pool of available kidneys, the KDRI could exacerbate this racial inequity in access to kidneys for transplantation.
diagnose and monitor pulmona	y disease				racial/ethnic minorities (e.g., in asthma and COPD). ²³

Reconsidering the Use of Race Correction in Clinical Algorithms

"Our understanding of race has advanced considerably in the past decades. The clinical tools we use daily should reflect these new insights to remain scientifically rigorous. Equally important is the project of making medicine a more antiracist field. This involves revising how clinicians conceptualize race to begin with."

Vyas DA et al. NEJM 383;9:874-882 (2020)

Evolving use of ancestry, ethnicity, and race in genetics research

Evolving use of ancestry, ethnicity, and race in genetics research—A survey spanning seven decades

ARTICLE

Yen Ji Julia Byeon,^{1,3} Rezarta Islamaj,² Lana Yeganova,² W. John Wilbur,² Zhiyong Lu,² Lawrence C. Brody,^{4,*} and Vence L. Bonham^{3,*}



Byeon YJJ, Islamaj R, Yeganova L, Wilbur WJ, Lu Z, Brody LC, Bonham VL. Evolving use of ancestry, ethnicity, and race in genetics research-A survey spanning seven decades. Am J Hum Genet. 2021 Dec 2;108(12):2215-2223.

Perspective

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> Starting with the launch of the Human Genome Project three decades ago, and continuing after its completion in 2003, genomics has progressively come to have a central and catalytic role in basic and translational research. In addition, studies ncreasingly demonstrate how genomic information can be effectively used in clinic: care. In the future, the anticipated advances in technology development, biological insights, and clinical applications (among others) will lead to more widespread integration of genomics into almost all areas of biomedical research, the adoption of genomics into mainstream medical and public-health practices, and an increasing elevance of genomics for everyday life. On behalf of the research community, the National Human Genome Research Institute recently completed a multi-year process of strategic engagement to identify future research priorities and opportunities in human genomics, with an emphasis on health applications. Here we describe the highest-priority elements envisioned for the cutting-edge of human genomics going orward-that is, at 'The Forefront of Genomics'

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na zoos, material sous parmer issues on energy in the source of the sour Improving laboratory and computational greater than tenfold increase in the relative fraction of funding co become increasingly woven into the fabric from other parts of the NIH.

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Box 5

Bold predictions for human genomics by 2030

Some of the most impressive genomics achievements, when viewed in retrospect, could hardly have been imagined ten years earlier. Here are ten bold predictions for human genomics that might come true by 2030. Although most are unlikely to be fully attained, achieving one or more of these would require individuals to strive for something that currently seems out of reach. These predictions were crafted to be both inspirational and aspirational in nature, provoking discussions about what might be possible at The Forefront of Genomics in the coming decade.

- 1. Generating and analysing a complete human genome sequence will be routine for any research laboratory, becoming as straightforward as carrying out a DNA purification.
- 2. The biological function(s) of every human gene will be known; for non-coding elements in the human genome, such knowledge will be the rule rather than the exception.
- 3. The general features of the epigenetic landscape and transcriptional output will be routinely incorporated into predictive models of the effect of genotype on phenotyr
- 4. Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.

o. Studies that involve analyses of genome sequences and associated phenotypic information for millions of human participants will be regularly featured at school science fairs.

6. The regular use of genomic information will have transitioned from boutique to mainstream in all clinical settings, making



4. Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.

equitably from advances in numan genomics.

10. Breakthrough discoveries will lead to curative therapies involving genomic modifications for dozens of genetic diseases.

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

https://www.nationalacademies.org/our-work/use-of-race-ethnicity-and-ancestry-as-population-descriptors-in-genomics-research

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
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 just and scientifically essential, which includes meaningful,
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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.

COMMENTARY



Building a Diverse Genomics Workforce: An NHGRI Action Agenda



The genomics workforce must become more diverse: a strategic imperative

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



genome.gov/workforcediversity

Training Diversity and Health Equity Office (TiDHE)

 Office was developed in April 2021 in the Office of the Director and expanded to include the Extramural Research Program (ERP) training in September 2021

Mission:

- Coordinate, develop and support NHGRI training programs for genomics careers
- Develop and support initiatives to enhance genomic workforce diversity and genomic health equity
- Provide strategic programmatic leadership for training, diversity and health equity at NHGRI



Training Diversity and Health Equity Office (TiDHE)

- Work as a Team with NHGRI Leadership and Staff
- Build Partnerships
- Initiate Programs to Advance Health Equity
- Support NHGRI Training Programs
- Conduct Training, Diversity and Health Disparities Portfolio Analysis

- Advise and Incubate the Development of New Programs
- Engage NHGRI and NIH Community
- Facilitate NHGRI and Trans NIH
 Programs
- Represent NHGRI in NIH Diversity and Health Equity Efforts
- Convene the Scientific Community for the Advancement of Training, Diversity and Health Equity

TiDHE Team



Lucia Hindorff, PhD



Ebony Madden, PhD



Jamil Scott, PhD



Christina Daulton, MA



Faye Brown



Lorjetta Schools, MBA



UNITE

UNITE aims to establish an equitable and civil culture within the biomedical research enterprise and reduce barriers to racial equity in the biomedical research workforce

https://www.nih.gov/ending-structural-racism/unite



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

HGG

Advances

The diversity of the U.S. population is currently not ref Although diversity and inclusion efforts have focused on i entific fields, structural racism remains. Thus, the cultivat intentional about an institution's character, culture, and c anti-racism approach within the field. Adopting a new per researchers as we build supportive, collaborative research e the research enterprise and propose an anti-racism appro



COMMENTARY We're right here

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.





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,*}

"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.

The Promise of Genomics as a Field





CRISPR-Cash -

Thank You bonhamv@nih.gov