Future Directions in Genomics and Health Equity

NHGRI TiDHE Office

April 6, 2022 | 10:00 AM – 2:15 PM ET

April 7, 2022 | 11:00 AM – 3:00 PM ET

National Human Genome Research Institute

The Forefront of Genomics®

Future Directions in Genomics and Health Equity Research

April 6, 2022 | 10:00 AM – 2:15 PM ET
April 7, 2022 | 11:00 AM – 3:00 PM ET

Virtual Zoom Meeting

Meeting page: https://www.genome.gov/event-calendar/future-directions-in-genomics-and-health-equity-research

TABLE OF CONTENTS

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AGENDA

Day 1: April 6

10:00 AM **Welcome and Introduction of Workshop Goals and Objectives**
Ebony Madden, Ph.D.
National Human Genome Research Institute

10:10 AM **Connecting NHGRI's Strategic Vision for Genomics with Health Equity**
Eric Green, M.D., Ph.D.
Director, National Human Genome Research Institute

10:20 AM **Talk: Setting the Stage: A Vision for Health Equity in Genomics**
Nancy Cox, Ph.D.
Vanderbilt University

11:00 AM **Panel: Moving Forward: From Health Disparities to Health Equity in Genomics**
Panelists:
Esteban Burchard, Ph.D., MPH, University of California, San Francisco
Vanessa Hiratsuka, Ph.D., MPH, University of Alaska Anchorage
Latrice Landry, M.S., Ph.D., M.Sc., Harvard University
Maya Sabatello, LLB, Ph.D., Columbia University

Moderator:
Eliseo Pérez-Stable, M.D., National Institute on Minority Health and Health Disparities

12:00 PM **BREAK**

12:15 PM **Talk: Current Research in Genomics and Health Equity**
John Carpten, Ph.D.
University of Southern California

12:55 PM **Panel: Identifying Research Gaps and Opportunities**
Panelists:
Rick Kittles, Ph.D., City of Hope
Loren Saulsberry, Ph.D., University of Chicago
Michael Inouye, Ph.D., University of Cambridge

Moderator:
Chanita Hughes-Halbert, Ph.D., University of Southern California

1:55 PM **Moderated Discussion: End of Day 1 Check-in / Preparing for Day 2**
Moderator:
Sandra Soo-Jin Lee, Ph.D.
Columbia University

2:15 PM **ADJOURN**

Day 2: April 7

11:00 AM **Building a Genomic Science Health Equity Research Agenda**
Vence Bonham, Jr., J.D.
Acting Deputy Director, National Human Genome Research Institute

11:10 AM **Talk: Current Challenges in Genomic Research and Genomic Medicine That Lead to Health Disparities**
James Hildreth, Ph.D., M.D.,
Meharry Medical College
&
Genevieve Wojcik, Ph.D., MHS,
John Hopkins University

12:00 PM **Panel: Addressing Structural Factors Needed to Support Health Equity Research in Genomics**
Panelists:
Kellan Baker, Ph.D., Whitman-Walker Institute
Rene Begay, M.S., C.P.H., University of Colorado Anschutz Medical Campus
Faith Fletcher, Ph.D, M.A., Baylor College of Medicine
Neil Risch, Ph.D., University of California, San Francisco
Moderator:
Carol Horowitz, M.D., MPH, Mount Sinai School of Medicine

12:30 PM **Breakout Groups (Participants will join one of five breakout groups)**

1. *Social Determinants of Health and Genomic Equity*
   Leaders:
   Tabia Henry Akintobi, Ph.D., M.P.H., Morehouse School of Medicine
   Nanibaa' Garrison, Ph.D., University of California, Los Angeles

2. *Structural Factors*
   Leaders:
   Nita A.Limdi, Pharm.D., Ph.D., MSPH, University of Alabama at Birmingham
   Stephanie Malia Fullerton, D.Phil., University of Washington

3. *From Bench to Bedside: The Implementation Science of Genomics and Health Equity*
   Leaders:
   Denise Dillard, Ph.D., Southcentral Foundation
   Elizabeth Ofili, M.D., M.P.H., FACC, Morehouse School of Medicine

4. *Data Science Genomics Equity*
   Leaders:
   Valentina Di Francesco, M.S., National Human Genome Research Institute
   Jeff Leek, Ph.D., M.S., John Hopkins School of Public Health

5. *Health Equity Research in ELSI*
   Leaders:
   Catherine Hammack-Aviran, M.A., J.D., Vanderbilt University School of Medicine
   Benjamin Wilfond, M.D., Seattle Children's Hospital

1:30 PM **Breakout Groups Report Back**

1:45 PM **BREAK**

2:00 PM **UNITE Presentation**
Speaker: Leia Butler, J.D., National Institutes of Health
Moderator: Lawrence Brody, Ph.D., National Human Genome Research Institute

2:15 PM ***All of Us* Presentation**
Speaker: Karriem Watson, DHSc, M.S., M.P.H., National Institutes of Health
Moderator: Lawrence Brody, Ph.D., National Human Genome Research Institute

2:30 PM **Polling and Prioritization of Recommendations; Measuring Success; Summary and Wrap-up Discussion**
Moderators:
Judy Cho, M.D., Mount Sinai School of Medicine
Lucia Hindorff, Ph.D, National Human Genome Research Institute

3:00 PM **ADJOURN**

GOALS AND OBJECTIVES

The goal of the workshop is to identify research gaps and opportunities that will help to decrease health disparities and improve health equity in genomics. Participants will emphasize the scientific value of diversity, inclusion, and health equity research; identify and prioritize future research needs in genomics and health equity research; and explore best practices for making genomic data, technology, and genomically-informed healthcare ethical, representative, accessible, and beneficial to all.

OBJECTIVES:

- Delineate the meaning of moving forward from health disparities to health equity in genomics.
- Go beyond health disparities research to identify areas of genomic research that are important to advance health equity.
- Identify research and partnerships needed to understand and address structural factors that impact health equity in genomics.
- Define how success is measured within genomics and health equity.

Working Definitions

- *Health Disparities*: Health disparities populations are race and ethnic minorities as defined in the US Census, persons of low socioeconomic status (low-SES), underserved rural residents, and sexual and gender minorities. A health disparity is defined as a condition that has worse outcomes in the disparity populations. (Source NIH Research, Condition, and Disease Categorization)

- *Health Equity*: The attainment of the highest level of health for all people. Achieving health equity requires valuing everyone equally with focused and ongoing societal efforts to address avoidable inequalities, historical and contemporary injustices, and the elimination of health and health care disparities. (Health People 2020

BIOGRAPHIES

Co-Chairs

Judy Cho, M.D.
Judy H. Cho, MD, is the Director of the Charles Bronfman Institute for Personalized Medicine (CBIPM), and the Ward-Coleman Chair for Translational Genetics at the Icahn School of Medicine at Mount Sinai. Her research focuses on inflammatory bowel disease (IBD) genetics and disease mechanisms and her laboratory is applying single cell RNASeq and CITE-Seq toward developing novel therapeutic insights. Dr. Cho has served as Principal Investigator of the Data Coordinating Center of the NIH-funded NIDDK IBD Genetics Consortium since 2003. In this capacity, she has led efforts in the identification of over 200 genetic regions associated to IBD. Since 2015, Dr. Cho has led the CBIPM, which includes the school's major biobank, BioMe, which represents one of the most diverse biobanks in the world and sequencing results underscore the enormous potential of a genetics first strategy in clinical care. These initiatives reflect the school's major commitment to Personalized Medicine to improve the care of patients on an individualized basis. Most recently, Dr. Cho received the prestigious 2021 Sherman Prize for Excellence in Crohn's and Colitis. Her cutting edge research in IBD has placed her in great demand on an international level as a guest speaker, collaborator, and leading authority in this particular field of research.

Sandra Soo-Jin Lee, Ph.D.
Sandra Soo-Jin Lee, PhD, is Professor of Medical Humanities and Ethics and Chief of the Division of Ethics at Columbia University. Trained as a medical anthropologist, Dr. Lee leads multi-disciplinary bioethics research on race, ancestry and equity in genomics, precision medicine and artificial intelligence, governance of biorepositories and commercialization of biotechnology. A current R01 funded study that she co-leads with Dr. Janet Shim at UCSF is entitled *Ethics of Inclusion: Diversity in Precision Medicine Research (*1R01HG010330). Dr. Lee publishes broadly in the genomics, medical, bioethics, and social science literatures, and co-edited *Revisiting Race in a Genomic Age* (2008). Dr. Lee is Co-Director of the NIH/NHGRI funded Center for ELSI Resources and Analysis (CERA) and the Co-Director of biennial International ELSI Congress. Dr. Lee currently serves as President-elect of the Association of Bioethics Program Directors, and on the US Health and Human Services Secretary's Advisory Committee on Human Research Protections and the National Academies of Science, Engineering and Medicine's Committee on the Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research, and on the Scientific Advisory Boards of the Kaiser Permanente National Research Biobank and the Human Pangenome Reference Consortium. She is a Hastings Center Fellow and serves on the editorial boards of the *American Journal of Bioethics* and *Narrative Inquiry in Bioethics*. Dr. Lee received her doctorate from the UC Berkeley/UCSF joint program in Medical Anthropology and her undergraduate degree in Human Biology from Stanford University.

Speakers

Leia Butler, J.D.

Leia Butler is on a detail assignment with the Chief Officer for Scientific Workforce Diversity as a Program Manager. In this role, Leia is responsible for the daily operations and all programming for UNITE and serves as a liaison among the UNITE committees. She also serves as an Anti-Racism Steering Committee (ARSC) Program Manager. Prior to assuming these roles, Leia served as a Supervisory Human Resources Specialist with the NIH Office of Human Resources' Workforce Relations Division, Employee and Labor Relations Branch. Leia also served as a Co-Chair of the ARSC Recruitment Recommendation (Non-Scientific) Subcommittee. She has over 14 years of Federal government experience, including the United States Patent and Trademark Office and the Federal Aviation Administration. Leia earned a Bachelor of Arts Degree, Psychology from the University of Maryland, Baltimore County and a Juris Doctorate Degree from American University's Washington College of Law.

John Carpten, Ph.D.

Dr. John Carpten is an internationally recognized leader in cancer genomics and precision oncology and the founding chair of translational genomics at the University of Southern California. His current work focuses on the entire DNA and RNA sequences of tumors to identify biochemical vulnerabilities that can be targeted with new or existing therapies. With over 190 peer-reviewed publications and more than a dozen patents to his credit, Dr. Carpten has generated landmark findings. He was a lead author on the first study to probe the entire genome for inherited prostate cancer genes and on a study that identified a novel mutation in a gene that plays a role in the development of breast, colorectal and ovarian cancers. His lab profiled common mutations in genes leading to multiple myeloma — a form of cancer that disproportionately affects African Americans. He hopes that his work will one day lead to improvements in knowledge-based therapeutics to improve outcomes for cancer patients.

Nancy Cox, Ph.D.

Nancy J. Cox, PhD, is a quantitative human geneticist with a long-standing research program in understanding the genetic basis of human disease. She earned a BS in Biology from the University of Notre Dame in 1978, a PhD in Human Genetics from Yale University in 1982 and did post-doctoral work at Washington University in St. Louis and the University of Pennsylvania before joining the University of Chicago in 1987, where she spent 28 very happy years. She joined Vanderbilt in 2015 as the inaugural Director the Vanderbilt Genetics Institute, the Division Director for Genetic Medicine, and the Mary Phillips Edmonds Gray Professor. Current research includes a focus on integrating genome variation with genome function in the context of electronic health records research in BioVU, All of Us, the Global Biobanking Consortium, and related efforts. Dr. Cox has also worked since 2015 with Consuelo Wilkins at VUMC in research on how genetics can be used to improve health equity.

James Hildreth, Ph.D., M.D.

Dr. James Hildreth is a renowned immunologist and the 12th president and chief executive officer of Meharry Medical College. He is known for his groundbreaking work with AIDS and HIV. He was the first African American to hold a full tenured professorship in basic research at Johns Hopkins School of Medicine. Dr. Hildreth has led Meharry's efforts to ensure that disadvantaged communities have access to COVID-19 testing and vaccines. He graduated from Harvard University as a Rhodes Scholar, from Oxford University with a PhD in immunology, and obtained an MD from Johns Hopkins School of Medicine. He was the first associate dean for graduate studies at Johns Hopkins University School of Medicine for several years, where he created a summer research program for underrepresented minorities and was active in recruiting undergraduate students for graduate programs. He was also previously director of the Center for AIDS Health Disparities Research at Meharry Medical College in Nashville, Tenn.

Karriem Watson, DHSc, M.S., M.P.H.
Dr. Karriem S. Watson, DHS, MS, MPH, is the Chief Engagement Officer of the National Institutes of Health's *All of Us* Research (AoU) Program. Karriem leads the All of Us Research Program's efforts to foster relationships with participants, communities, researchers, and providers across the U.S. to help build one of the largest, most diverse health databases of its kind to study health and illness. Prior to joining the NIH, Karriem spent over 15 years as a community engaged research scientists with prior research funding from the NIH addressing cancer prevention and control. Karriem also held administrative roles in leading research and engagement in Federally Qualified Health Centers (FQHCs) in the Chicagoland area.

Genevieve Wojick, Ph.D., MHS
Genevieve L. Wojcik, Ph.D. is an Assistant Professor of Epidemiology at the Johns Hopkins Bloomberg School of Public Health in Baltimore, Maryland. As a statistical geneticist and genetic epidemiologist, her research focuses on method development for diverse populations, specifically understanding the role of genetic ancestry and environment in genetic risk in admixed populations. Dr. Wojcik integrates epidemiology, sociology, and population genetics to better understand existing health disparities in minority populations, as well as underserved populations globally. She is a long-standing member of multiple NHGRI consortia focused on diverse populations, most notably the Population Architecture using Genomics and Epidemiology (PAGE) Study. Prior to her faculty appointment, Dr. Wojcik was a postdoctoral research scholar at Stanford University in the Departments of Genetics and Biomedical Data Science. She received her Ph.D. in Epidemiology and M.H.S. in Human Genetics/Genetic Epidemiology from the JHSPH and her B.A. in Biology from Cornell University.

Moderators

Carol Horowitz, M.D., MPH
Carol R. Horowitz, MD, MPH, is Professor of Population Health Science and Policy, Professor of Medicine and a practicing general internist at the Icahn School of Medicine at Mount Sinai. She is the founding Dean for Gender Equity in Science and Medicine and the Director of the new Institute for Health Equity Research. Her research focuses on using Stakeholder-Engaged and Community-Based Participatory Research to address health disparities. As Principal and Co-investigator for federally-funded studies, her special interests are in chronic disease prevention and control, and how social, clinical, biological and behavioral determinants impact health disparities and health. She mentors diverse trainees and faculty interested in addressing disparities and inequities faced by individuals from underrepresented racial, ethnic, gender and sexual minority groups, as well as women. She partners with community and clinical stakeholders to use lessons learned to inform health, policies, systems, and environments.

Chanita Hughes-Halbert, Ph.D.
Chanita Hughes-Halbert, Ph.D., is the associate Director for Cancer Equity at the USC Norris Comprehensive Cancer Center and a Professor and vice chair of research in the Department of Population and Public Health Sciences at the Keck School of Medicine of USC. She is a nationally recognized leader in cancer prevention and minority health research. She has dedicated her career to reducing the disparities in cancer outcomes that affect patients from underrepresented communities, with a primary focus on African American communities. Among her many achievements, she has identified sociocultural, psychological, genetic, and environmental determinants of cancer health disparities and translates this information into interventions to improve health equity among racially and ethnically diverse populations, as well as other medically underserved groups. For her many contributions, Hughes-Halbert was elected to the National Academy of Medicine in 2017. In addition to her election to the National Academy of Medicine, Hughes-Halbert received the American Cancer Society's Cancer Control

Award in 2010. President Barack Obama appointed her to the National Cancer Institute's Board of Scientific Advisors in 2012, and in 2014 she joined the National Advisory Council of the National Human Genome Research Institute.

Eliseo Pérez-Stable, M.D.
Eliseo J. Pérez-Stable, M.D. is Director of the National Institute on Minority Health and Health Disparities (NIMHD) at the National Institutes of Health (NIH), which advances the science of minority health and health disparities research. Prior to becoming NIMHD Director, Dr. Pérez-Stable was a professor of medicine and chief of the Division of General Internal Medicine, at the University of California, San Francisco (UCSF). Dr. Pérez-Stable's research interests have centered on improving the health of racial and ethnic minorities through effective prevention interventions, understanding underlying causes of health disparities, and advancing patient-centered care for underserved populations. Recognized as a leader in Latino health care and disparities research, Dr. Pérez-Stable spent 32 years leading research on smoking cessation and tobacco control in Latino populations in the United States and Latin America. He has published more than 300 peer-reviewed papers and a career mentor for many students, residents, and faculty, and a research mentor for over 70 minority investigators.

Panelists

Kellan Baker, Ph.D.
Kellan Baker is the Executive Director of the Whitman-Walker Institute, which is the research, policy, and education arm of Whitman-Walker, a federally qualified community health center in Washington, DC. Kellan has been a member of the Community Engagement in Genomics Working Group at NHGRI since 2016 and is the co-chair of the LGBTQI+ Issues in Genomics Project Group through the Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG). He is a frequent advisor on health equity policy and research for entities across the federal government and for the National Academies of Sciences, Engineering, and Medicine. He holds a PhD in health policy and management from the Johns Hopkins School of Public Health, an MPH and MA from the George Washington University, and a BA with high honors from Swarthmore College.

Rene Begay, M.S.,C.P.H.
Rene Begay, M.S., is (Diné/Navajo) from Arizona. She is an Indigenous geneticist and public health researcher. She obtained her Bachelors of Science degree in Biology from the University of Arizona and a Masters in Clinical Science from the University of Colorado Anschutz Medical Campus. Currently, she works as a Professional Research Assistant at the Centers for American Indian and Alaska Native Health at the University of Colorado School of Public Health while studying as a Masters of Public Health and Bloomberg Scholar with the Johns Hopkins School of Public Health focusing on the topic of childhood obesity. Her research is informed by her cultural background and lived experiences in order to inform her work in genomics and health for Indigenous communities.

Esteban Gonzalez Burchard, M.D., M.P.H.
Dr. Burchard is a Mexican American physician-scientist and Professor in the UCSF Schools of Medicine and Pharmacy. Dr. Burchard initiated and now directs the largest gene-environment study of asthma in minority children in the U.S. Dr. Burchard was the first in the world to use genetic ancestry to improve the accuracy of lung function measures among Latino and African Americans. Dr. Burchard was an Advisor to the NIH Director for the Precision Medicine Initiative (2015); an Advisor to the National Academy of Sciences Board on Environmental Studies and Toxicology (2015); a member of the RWJ's Amos Medical Faculty Development Program National Advisory Committee (2017-2021); and a current member of the National Institute on Minority Health and Health Disparities Council (2022-2025).

Faith Fletcher, Ph.D, M.A.
Faith Fletcher, PhD, MA, is an Assistant Professor in the Center for Medical Ethics and Health Policy at Baylor College of Medicine and a senior advisor to the Hastings Center, a leading bioethics research institute. Nationally, Dr. Fletcher is contributing to critical conversations around health equity, structural racism, medical mistrust, and anti-racism in bioethics. She has been featured in Huffington Post, National Public Radio, Science Magazine, and Rolling Stone. In 2017, Dr. Fletcher was named one of the National Minority Quality Forum's 40 under 40 Leaders in Health for her commitment to improving access to scientific research and quality health care for medically underserved populations. This prestigious award acknowledges the next generation of leaders primed to reduce health disparities.

Vanessa Hiratsuka, Ph.D., MPH
Dr. Hiratsuka (Diné/Winnemem Wintu; she/her) is an assistant professor of clinical and translational research and co-director of research and evaluation at the University of Alaska Anchorage Center for Human Development. Her research interests include ethical, social, and legal implications of genomic research and precision medicine among Indigenous populations; cultural adaptation of chronic disease and behavioral health interventions; and community-engaged evaluation of health and training programs serving individuals experiencing intellectual and developmental disabilities.

Michael Inouye, Ph.D.
Professor Inouye is a computational biologist who has been analysing human genome data for more than 20 years. After training in the US, UK and Australia, he is now a Director of Research (i.e. Research Professor) in the Department of Public Health and Primary Care at the University of Cambridge (UK), Munz Chair of Cardiovascular Prediction and Prevention at the Baker Heart and Diabetes Institute (Australia) and Director of the Cambridge Baker Systems Genomics Initiative.

Rick Kittles, Ph.D.
Rick Kittles, Ph.D., is professor and founding director of the Division of Health Equities within the Department of Population Sciences at City of Hope. He is also associate director of health equities in the comprehensive cancer center. Dr. Kittles is well known for his research of prostate cancer and health disparities among African-Americans. His research has focused on understanding the complex issues surrounding race, genetic ancestry and health disparities. He received a Ph.D. in biological sciences from George Washington University in 1998. His first faculty appointment was at Howard University where he helped establish the National Human Genome Center at Howard University. Over the last 20 years, he has been at the forefront of the development of ancestry-informative genetic markers, and how genetic ancestry can be quantified and utilized in genomic studies on disease risk and outcomes. His work has shown the impact of genetic variation across populations in pharmacogenomics, biomarker discovery and disease gene mapping.

Latrice Landry, M.S.,Ph.D.,M.Sc.
As a clinical geneticist, epidemiologist and nutritionist, Dr. Landry is focused on the engineering of equity-based systems for clinical integration of biomarkers with a keen focus on genomic and nutrition related biomarkers, in the translation, evaluation, optimization, and implementation of technologies in diverse populations and is helping to lead equity and disparities research in the field of precision medicine and public health. She received both her master's degree in Policy and her PhD in Nutrition from Tufts University. Her doctoral research focused on the interactions between diet and genetics as determinants for dyslipidemia in African Americans in the Jackson Heart Study. As a doctoral student, Dr. Landry was awarded the Albert Schweitzer fellowship, nominated as a finalist in the American Society for Nutrition's Clinical Emerging Leaders Award, and was given the Presidential Award for Citizenship and Public Service at Tufts University. In

2015, following her doctoral research she joined Harvard Medical School's Biomedical Informatics Fellowship Program to study biomedical information systems (clinical informatics and bioinformatics) as tools for biomarker translation. In 2020, she joined the National Minority Quality Forum, Quest Diagnostics, and the Centene Corporation for the launch of the Minority and Rural Coronavirus Insights Study- a study aimed at understanding the role of COVID in minority communities. In 2021, she received the Dana Farber Cancer InstitutesCURE mentoring award.

Neil Risch, Ph.D.
Neil Risch is the Lamond Family Foundation Distinguished Professor in Human Genetics, Founding Director of the Institute for Human Genetics, and Professor and former chair of the Department of Epidemiology and Biostatistics at the University of California San Francisco. Dr. Risch received his undergraduate training at the California Institute of Technology in mathematics and his Ph.D. from the University of California Los Angeles in Biomathematics. He has previously held professorships at Columbia, Yale, and Stanford Universities. His research interests are in human population genetics, genetic epidemiology and statistical genetics. He is the recipient of the Curt Stern Award from the American Society of Human Genetics (ASHG), a fellow of the AAAS, the California Academy of Sciences and the National Academy of Medicine, and is past president of the ASHG. He is recognized for his novel statistical approaches in human genetics and in particular the introduction of genome-wide association studies.

Maya Sabatello, LLB,Ph.D.
Maya Sabatello, LLB, PhD is an Associate Professor of Medical Sciences (in Medicine) at the Center for Precision Medicine and Genomics, Department of Medicine; Associate Professor (in Medical Humanities and Ethics), at the Division of Ethics, Department of Ethics and the Humanities; and Co-Director of the Precision Medicine: Ethics, Politics, and Culture Project at Columbia University. She is a former litigator with trans-disciplinary background and has extensive experience in national and international policy-making relating to human and disability rights. Sabatello studies how biomedical technologies and genomic information impact social structures, marginalized communities, and individual rights and health outcomes. Her scholarship focuses on law, society, medicine, and disability; regulations of reproductive technologies; and the ethical, legal, and social implications of genetics and precision medicine.

Loren Saulsberry, Ph.D.
Dr. Loren Saulsberry is an Assistant Professor in Health Policy and Health Services Research in the Department of Public Health Sciences at The University of Chicago. Dr. Saulsberry's research evaluates the diffusion and uptake of emerging medical technologies to treat and manage chronic diseases with a particular focus on how health innovations impact health disparities. Her research studies pharmacogenomics and how to guide its implementation in a manner that advances health equity within genomic medicine, and she is currently pursuing this work as a part of a NHGRI Career Development Award. Dr. Saulsberry received her Ph.D. in Health Policy from Harvard University and is an alumna of the Dana Farber/Harvard Cancer Center's Training in Oncology Population Sciences Program. She is the Assistant Program Leader of the Cancer Prevention & Control Program within the UChicago Medicine Comprehensive Cancer Center and the Assistant Director of Diversity Studies within the UChicago Center for Personalized Therapeutics. Before entering academia, her prior experiences include cancer genetics research and working with the Kaiser Family Foundation.

Break Out Group Leaders

Tabia Henry Akintobi, Ph.D., MPH
Tabia Henry Akintobi, PhD, MPH is Professor and Chair of Community Health and Preventive Medicine at Morehouse School of Medicine. She is Principal Investigator of the MSM Prevention Research Center- advancing community-based participatory research and related approaches for over 20 years. As Associate Dean for Community Engagement, Dr. Henry Akintobi has led collaborations with leaders, across the institution to demonstrate MSM's preeminence in community health strategies towards successful acquisition of the Carnegie Designation for the Advancement of Teaching in Community Engagement and the Josiah Macy Inaugural Award for Excellence in Social Mission. She recently collaborated with the National Human Genome Research Institute to engage community groups to understand perceptions and recommendations associated with optimal community-engagement. These efforts are guided by training in public health, social marketing, community-based participatory research, and a mission to not only understand and address, but eradicate health disparities.

Denise Dillard, Ph.D.
Denise Dillard is Inupiaq Eskimo and was born in Fairbanks and raised in Anchorage. She is a licensed psychologist and has conducted quantitative and qualitative research with American Indian and Alaska Native peoples since 1998. She is currently the Director of Research for Southcentral Foundation (SCF), a tribal health organization in Anchorage, Alaska. She oversees the direction of a diverse portfolio of research studies addressing the wide-ranging needs of American Indian and Alaska Native community served by the organization. She works directly with tribal leadership at SCF as they consider approval of research proposals, abstracts, and manuscripts and is a member of the Alaska Area Institutional Review Board. At a national level, she serves as the Alaska Delegate of the National Institutes of Health Tribal Advisory Committee.

Stephanie Malia Fullerton, D.Phil
Stephanie Malia Fullerton, DPhil, is Professor of Bioethics and Humanities at the University of Washington School of Medicine. She is also Adjunct Professor in the UW Departments of Epidemiology, Genome Sciences, and Medicine (Medical Genetics), as well as an affiliate investigator with the Public Health Sciences division of the Fred Hutchinson Cancer Research Center. She received a PhD in Human Population Genetics from the University of Oxford and later re-trained in Ethical, Legal, and Social Implications (ELSI) research with a fellowship from the NIH National Human Genome Research Institute. Dr. Fullerton's work focuses on the ethical and social implications of genomic research and its equitable and safe translation for clinical and public health benefit. She serves as the ELSI lead for the Clinical Sequencing Evidence-Generating Research (CSER2) Consortium coordinating center, co-chairs the TOPMed Consortium ELSI Committee, and chairs the Bioethics Advisory Board of the Kaiser Permanente national Research Bank. She contributes to a range of empirical projects focused on clinical genomics translation and precision medicine approaches to the treatment and prevention of cancer and kidney disease in diverse patient populations.

Nanibbaa' Garrison, Ph.D.
Nanibaa' Garrison (Navajo), Ph.D. is an Associate Professor at the University of California, Los Angeles. She has appointments in the Institute for Society and Genetics, the Institute for Precision Health, and the Division of General Internal Medicine & Health Services Research. Her research focuses on the ethical, social, and cultural implications of genetic and genomic research in Indigenous communities. Using community-based research approaches, she engages with tribal communities to develop policies and guidance regarding genetic and genomic research. She is also a member of the US Indigenous Data Sovereignty Network, the

Navajo Nation Human Research Review Board, and co-director of the Summer internship for INdigenous peoples in Genomics (SING).

Catherine M. Hammack-Aviran, M.A., J.D.
Catherine M. Hammack-Aviran, MA, JD, is an Associate in Health Policy in the Department of Health Policy at Vanderbilt University's School of Medicine and is a member of the core faculty in the Center for Biomedical Ethics and Society at Vanderbilt University Medical Center. She received her Doctor of Jurisprudence from Wake Forest University School of Law and her Master of Arts in Bioethics from Wake Forest University's Center for Bioethics, Health, & Society. Prof. Hammack-Aviran has over ten years of experience in empirical bioethics, legal, and social science research using a variety of methodologies to study a myriad of topics. She is integrally involved in the design, development, conduct, analysis, and dissemination of myriad empirical bioethics investigations, drawing upon her expertise in bioethics and law/regulation/policy and experience in qualitative research to collaborate on more than 20 research projects (including 15 NIH-funded studies).

Jeff Leek, Ph.D., M.S.
Jeff is a professor of Biostatistics and Oncology at the Johns Hopkins Bloomberg School of Public Health and co-director of the Johns Hopkins Data Science Lab. His group develops statistical methods, software, data resources, and data analyses that help people make sense of massive-scale genomic and biomedical data. As the co-director of the Johns Hopkins Data Science Lab he has helped to develop massive online open programs that have enrolled more than 8 million individuals and partnered with community-based non-profits to use data science education for economic and public health development. He is a Fellow of the American Statistical Association and a recipient of the Mortimer Spiegelman Award and Committee of Presidents of Statistical Societies Presidential Award.

Nita Limdi, Pharm.D., Ph.D., MSPH
Nita Limdi, Pharm.D, PhD, MSPH is Professor of Neurology and Epidemiology. She started her career as a hospital pharmacist after graduating from Samford University with a Pharm.D (1994), continuing her training obtaining her MSPH (2005) and PhD in Epidemiology (2007). As a clinical pharmacist and chronic disease epidemiologist with 20 years of experience, she brings her breath of expertise in clinical pharmacy, chronic disease epidemiology, and pharmacogenomics to lead research and implementation of genomics in clinical practice. Her efforts to recruit and engage African Americans (AA) and medically underserved patients has been vital to her contributions to understanding racial differences in drug response, identifying race-specific variants, reporting on the differential impact of gene variants and comorbidities by race. Through her work, Dr. Limdi has collaborated extensively with national/ international consortia including the Pharmacogenomics Research Network (PGRN), the Pharmacogenomics Knowledge Base (PharmGKB), the Clinical Pharmacogenetics Implementation Committee (CPIC), the Personalized Medicine Coalition (PMC), and Standardizing Laboratory Practices in Pharmacogenomics (STRIPE).

Elizabeth Ofili, M.D., M.P.H., FACC
Dr. Elizabeth Ofili is a Professor of Medicine, at Morehouse School of Medicine, and a practicing cardiologist with Morehouse Healthcare in Atlanta, Georgia. She serves as Chief Medical Officer for the Morehouse Choice Accountable Care Organization, a Center for Medicare and Medicaid Services (CMS) Shared Savings Program, which includes Morehouse Healthcare, and Federally Qualified Health Centers across the state of Georgia. Dr. Ofili is a national and internationally recognized clinician scientist with particular focus on cardiovascular disparities and women's health. Dr. Ofili helped to raise over $175 million to fund pioneering research and training infrastructure at Morehouse School of Medicine. She leads multi-institutional regional and national networks funded by the National Institutes of Health, to address diversity and inclusion in the biomedical workforce, as well as community

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NHGRI

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Dr. Eric Green is the director of the National Human Genome Research Institute (NHGRI) at the U.S. National Institutes of Health (NIH). He is the third NHGRI director, having been appointed by NIH director Dr. Francis Collins in 2009. Dr. Green has been at the Institute for more than 25 years, during which he has had multiple key leadership roles. He served as the Institute's scientific director for 7 years, chief of the NHGRI Genome Technology Branch for 13 years and founding director of the NIH Intramural Sequencing Center for 12 years. For just over two decades, Dr. Green directed an independent research program that included integral start-to-finish roles in the Human Genome Project and groundbreaking work on mapping, sequencing, and characterizing mammalian genomes. Dr. Green earned his M.D. and Ph.D. degrees in 1987 from Washington University in St. Louis; coincidentally, the word "genomics" was coined in that same year. During his career, Dr. Green has authored and co-authored over 375 scientific publications.

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Strategic vision for improving human health at The Forefront of Genomics

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Abstract

Starting with the launch of the Human Genome Project three decades ago, genomics has become progressively entrenched within the bedrock of the biomedical research enterprise. Capitalizing on the momentum of the project's successful completion in 2003, genomics now regularly plays a central and catalytic role in basic and translational research, and studies increasingly demonstrate the vital role that genomic information can play in clinical care. Looking ahead, the anticipated advances in technology development, biological insights, and clinical applications (among others) will lead to more widespread integration of genomics into virtually all areas of biomedical research, the adoption of genomics into mainstream medical and public-health practices, and an increasing relevance of genomics in everyday life. On behalf of the research community, the National Human Genome Research Institute recently completed a multi-year process of strategic engagement to capture input about the future research priorities and opportunities in human genomics, with an emphasis on health applications. Here we articulate the highest-priority elements envisioned for the cutting-edge of human genomics going forward – that is, at "The Forefront of Genomics."

Introduction

Three decades ago this month, a pioneering group of international researchers began an audacious journey to generate the first map and sequence of the human genome, marking the start of a 13-year odyssey called the Human Genome Project[1–3]. The project's successful and early completion in 2003, which included parallel studies of a set of model organism genomes, catalyzed enormous progress in genomics research. Leading the signature

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advances has been a greater than one million-fold reduction in the cost of DNA sequencing[4]. This decrease has allowed the generation of innumerable genome sequences, including hundreds of thousands of human genome sequences (both in research and clinical settings), and the continuous development of new assays for identifying and characterizing functional genomic elements[5,6]. With these new tools, coupled with increasingly sophisticated statistical and computational methods, researchers have been enabled to create rich catalogs of human genomic variants[7,8], to gain an ever-deepening understanding of the functional complexities of the human genome[5], and to elucidate the genomic bases of thousands of human diseases[9,10]. In turn, the last decade has brought the initial realization of genomic medicine[11], as research successes have been converted into powerful tools for use in healthcare, including somatic genome analysis for cancer (enabling development of targeted therapeutics)[12], noninvasive prenatal genetic screening[13], and genomics-based tests for a growing set of pediatric conditions and rare disorders[14], among others.

In essence, with growing insights about the structure and function of the human genome and ever-improving laboratory and computational technologies, genomics has become increasingly woven into the fabric of biomedical research, medical practice, and society. The scope, scale, and pace of genomic advances to date were nearly unimaginable when the Human Genome Project began; even today, such advances are yielding scientific and clinical opportunities beyond our initial expectations, with many more anticipated in the decade ahead.

Embracing its leadership role in genomics, the National Human Genome Research Institute (NHGRI) has developed strategic visions for the field at key inflection points, in particular at the end of the Human Genome Project in 2003[15] and then again at the beginning of the last decade in 2011[16]. These visions outlined the most compelling opportunities for human genomics research, in each case informed by a multi-year engagement process. NHGRI endeavored to start the new decade with an updated strategic vision for human genomics research. Through a planning process that involved over 50 events (e.g., dedicated workshops, conference sessions, and webinars) over the last two years (see http://genome.gov/genomics2020), the institute collected input 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 significantly influenced 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 U.S. National Institutes of Health (NIH), but the past two decades have brought a greater than ten-fold increase in the relative fraction of funding coming from other parts of NIH.

The planning process continually encountered the realities associated with the broad and extensive use of genomics and the impracticality of being comprehensive, which together served to focus attention on the most cutting-edge opportunities in human genomics. This experience affirmed NHGRI's recently rearticulated role in providing genomics leadership at NIH, embodied by our newly conceived organizational mantra: "The Forefront of

Genomics." We ultimately linked this mantra to the strategic planning process to help guide the formulation of input. From the ensuing discussions, it became apparent that responsible stewardship is a central aspect of being at (and pushing forward) The Forefront of Genomics, specifically in the four major areas detailed in Fig. 1, Boxes 1, 2, 3, and 4, and below.

Principles and values for human genomics

As genomics has matured as a discipline, the field has embraced a growing set of fundamental principles and values that together serve as a guiding compass for the research efforts – some of these emerged organically within the field, whereas others have been adopted from the broader scientific community. The growing complexities of human genomics and its many applications (especially in medicine) at The Forefront of Genomics make it imperative to reaffirm, sharpen, and even extend these tenets, such as those highlighted in Box 1.

Many of these principles and values have been informed by the recognized area of genomics that focuses on ethical, legal, and social implications (ELSI) research[17], which was established at the beginning of the Human Genome Project to help guarantee that the eugenics movement and other misuses of genetics are not repeated. ELSI research has since grown to encompass a broad portfolio of studies examining issues at the interface of genomics and society, the results of which have informed policies and laws related to genetic discrimination, intellectual property, data sharing, and informed consent[18]. Similar efforts seek to ensure that the benefits of genomics are available to all members of society[19]. Genomics, like all fields of science, must reckon with systematic injustices and biases, fully cognizant of their criticality for health equity. Looking ahead, ELSI research needs to focus on aspects of genomic medicine implementation that present challenging questions about legal boundaries, study governance, data control, privacy, and consent. Complex societal issues must also be studied, including the expanded application of genomics in non-medical realms (e.g., ancestry testing, law enforcement, and genetics-based marketing of consumer goods)[20]. Finally, ELSI research could also examine the implications of studying genetic associations with bio-behavioral traits (e.g., intelligence, sexual behavior, social status, and educational attainment)[21] and of a future where machine learning and artificial intelligence are used to tailor risk communication and clinical decisions based on analyzing an individual's genome sequence[22].

Robust foundation for genomics

Genomics is now routinely and broadly utilized throughout biomedical research, with widespread reliance on a robust foundation for facilitating genomic advances. The foundation's integrity depends on a number of key elements, including infrastructure, resources, and dynamic areas of technology development and research. Sustaining and improving that foundation are key responsibilities at The Forefront of Genomics, the major elements of which are highlighted in Box 2 and detailed in corresponding paragraphs below.

Genome structure and function

The last two decades have brought a greater than million-fold reduction in the cost of DNA sequencing[23] along with an explosion in technologies for functional genomics[6,24,25] (i.e., the study of how elements in the genome contribute to biological processes). Additional opportunities are poised to be unlocked as the generation and analysis of genomic data become even faster, cheaper, and more accurate. Near-term expectations include enhanced capabilities for generating high-quality and complete (e.g., telomere-to-telomere and phased) genome sequences[26,27] and continued refinement and enhanced utility of a human genome reference sequence(s) that increasingly reflects human variation and diversity on a global scale[28] and that serves as a substrate for genome annotation[29]. Technologies for generating DNA sequence and other data types (e.g., transcriptomic data, epigenomic data, and functional readouts of DNA sequences) need to be enabled at orders-of-magnitude lower costs, at single-cell resolution, at distinct spatial locations within tissues, and longitudinally over time[30–32]. These genomic data should be integrated with other multi-omic data (e.g., proteomes, metabolomes, lipidomes, and/or microbiomes) in sophisticated ways, including novel methods that collect multiple data types from a single sample[32]. Transformative approaches will become increasingly vital for assimilating, sharing, and analyzing these complex and heterogeneous data types[33] and must expand to include the integration of environmental, lifestyle, clinical, and other phenotypic data. These capabilities should be incorporated into browsers, portals, and visualization tools for use by a broadening community of researchers and clinicians.

Genome sequences have now been generated for over 1,000 vertebrate species and are increasingly accompanied by multi-species annotations[34]. Understanding natural genomic variation, conservation of genomic elements, and the rapid evolutionary changes in genomic regions associated with specific traits is critical for attaining a comprehensive view of genome structure and function. The study of a wide range of organisms continues to be instrumental for elucidating the impact of genomic variation on biological processes and phenotypes, providing insights about the interplay of genomic variants and environmental pressures[35] and the relevance of putative pathogenic variants identified in clinical studies[36]. It is essential that the generation of high-quality multi-species genomic data be accompanied by community-accepted standards for data, metadata, and data interoperability. New groundbreaking methods would allow for integrating functional data from diverse species with human data and visualizing increasingly complex comparative genomic datasets. Continued progress in this area would move the field closer to the long-term aspirational goal of understanding the evolutionary history of every base in the human genome.

Genomic data science

All major genomics breakthroughs to date have been accompanied by the development of groundbreaking statistical and computational methods. Accordingly, continued innovations in both traditional and advanced methods (including machine learning and artificial intelligence) should be prioritized[37]. These approaches must be considered from the early stages of study planning and data collection in ways that complement and enhance, rather than inhibit, technical progress. Further, the biomedical research community requires accurate, curated, accessible, secure, and interoperable genomic data repositories and

informatics platforms that benefit all populations. Approaches for improving the efficiency of such resources include the use of shared storage and computing infrastructure, the adoption of common data-management processes, and the development of increasingly automated data-curation methods[38]. Carefully considered funding strategies must be designed to support these methods and resources, including a global, multi-funder model that ensures their development, enhancements, and long-term sustainability[39].

Recent progress has brought substantial transformations in how the petabytes of genomic data being generated each year are assimilated and analyzed, including the emergence of cloud-based and federated approaches. Effective and efficient management of increasingly complex genomic datasets requires addressing challenges with these emerging approaches as well as innovations in the use of hardware, algorithms, software, standards, and platforms[40]. Current barriers include the lack of interoperable genomic data resources (which limits downstream access, integration, and analyses) and the absence of controlled and consistently adopted data and metadata vocabularies and ontologies[41,42]. User-friendly systems that capture metadata in a scalable, intelligent, and cost-effective fashion and that allow for intuitive data visualizations are essential. Ever-improving routines and guidelines should be formulated to continue and even enhance responsible data sharing, requiring the collective efforts of researchers, funders, and publishers alike; similar attention should focus on ensuring the use of FAIR (findable, accessible, interoperable, and reusable) data standards and the reproducibility of data analyses[38]. Innovations in technology and policy must be integrated to develop data-stewardship models that ensure open science and reduce data-access burdens to advance research, including the use of optimally balanced and ethically sound approaches for respecting participant preferences and consent as well as engaging communities. Such developments should be done in an open-source culture to build consensus and enable the development, maintenance, and utilization of best-in-class tools, pipelines, and platforms that can be applied to all datasets.

Fully integrating genomics into medical practice will require informatics and data-science advances that effectively connect the growing body of genomic knowledge to clinical decision-making. To make genomic information readily accessible and broadly useful to clinicians, user-friendly electronic health record-based clinical decision support tools must be created to interact with a variety of clinical data from electronic health record and other data systems (e.g., laboratory, pharmacy, and radiology) as well as non-computable reports, such as those provided as portable document format (PDF) files[43,44]. These efforts require well-curated, highly integrated, and up-to-date knowledgebases that connect genomic information to clinical characteristics, other phenotypic data, and information on family health history[45]. Also needed are reliable risk-stratification and prevention algorithms, including polygenic risk scores (PRSs)[46], that incorporate both common and rare genomic variants from a broad range of population subgroups, phenotypic data, and environmental information into the risk modeling[47]. Such algorithms should be evaluated both for their validity across multiple populations and for their impact on patient outcomes and subsequent healthcare utilization. Finally, it will be important to evaluate new genomics-oriented clinical decision support tools to ensure that they are acceptable to practitioners across the spectrum of clinical disciplines.

Genomics and society

Understanding the role of genomics in human health requires knowledge and insights about how social, environmental, and genomic risk factors interact to produce health outcomes[48,49] (Box 1). Given that such interactions are, in general, poorly understood, it is critical that studies of genomic risk (particularly of common, complex diseases) account for the social and environmental factors that influence health and disease[50]. These factors must be properly described, measured, and incorporated in genomics studies[51]. Optimal implementation of genomic medicine will require understanding how the intersectional aspects of people's social and political identities influence the ways in which populations are described in research. Such knowledge will, in turn, provide clarity about the interrelationships among these multiple influences on health and disease.

People want to be able to make well-informed decisions about their genomic data, leading to the engagement efforts in initiatives such as the UK Biobank[52] and the *All of Us* Research Program[53]. Partnering with communities and individuals is fundamental to engaging participants in such large-scale research. Genomic researchers must incorporate models and methods of community engagement in their experimental design. Such studies must be appropriately tailored for different cultures and designed to reduce inequities and healthcare disparities; they must also be accompanied by effective information dissemination[54]. An unrelenting focus on the optimal ways to conduct research in partnership with data stakeholders and communities would ensure the identification of the key issues and values influencing peoples' choices about the provision of personal data for research[55,56]. Data-stewardship infrastructures that integrate appropriate policies, technologies[57], and governance and legal frameworks must be developed and assessed to ensure alignment between communities' and individuals' decisions about their data and the practices of researchers and clinicians.

To fully realize the fruits of genomic advances, a working understanding of the basic concepts of genomics will be important for science educators[58], healthcare professionals[59], policymakers, and the public[60]. Multiple educational strategies will inevitably be required for enhancing the genomic literacy of these heterogeneous groups, which points to the need for innovative approaches that are shared, assessed, and improved over time[58]. A growing evidence base shows that increasing the understanding of key genomics concepts and applications attracts students to careers in genomics[61], assists with the use of genomics for addressing health disparities[62], and facilitates the uptake of genomic medicine[63]. Curricula for enhancing genomic literacy must be designed to be accessible, effective, and scalable for use in the full range of settings where genomics education is provided – including primary and secondary schools, science museums, and informal science-education venues. Researchers and educators must also disseminate information about both the science of genomics as well as the key ethical and societal implications of genomics[64].

Training and genomics workforce development

Appropriate skills in data science and data stewardship are now prerequisites for becoming a genomics researcher[65]. Furthermore, given the ever-expanding use of genomics in basic, translational, social, behavioral, and clinical research, a greater number of scientists will

require fundamental data-science skills appropriate for the genomic applications being utilized[66]. Establishing and maintaining data-science competencies for conducting genomics research requires a series of interrelated educational and training efforts[67], including the recruitment of a cadre of data scientists into genomics and the reciprocal exchange of expertise between genomics researchers and data scientists.

Moving into healthcare, providers must be poised to manage questions from patients who receive genomic information, including that from direct-to-consumer testing, and this applies to the full spectrum of medical professionals (including nurses, pharmacists, physicians, and other clinicians)[68]. Education modules tailored to specific user groups should be designed to adapt rapidly to advances in genomics and data-science technologies; these should be available on demand and, where appropriate, integrated into existing clinical systems[69]. Research on the methodologies for train-the-trainer approaches, implementation of standards and competency-based education, and strategies for enhancing genomic literacy among all healthcare providers at all career stages[70] should also be pursued. The involvement of patients, caregivers, educators, professional organizations[71], and accreditation boards will be critical to ensure success. Importantly, cross-training in relevant aspects of genomics must also be available for specialists working in or around healthcare systems, including (but not limited to) those involved in health services research, health economics, law, bioethics, and social and behavioral sciences.

Both in 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 U.S.[72]), which limits the opportunity of those systematically excluded to bring their unique ideas to scientific and clinical research[73]. 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; rather, they must also include incentives to recruit and retain a diverse workforce at all career stages[74] as well as novel approaches for cultivating the next generation of genomics practitioners.

Breaking down barriers in genomics

Genomics has benefited enormously from the proactive identification of major obstacles impeding progress and the subsequent focused efforts to break down those barriers. Prototypic successes include the call for a "$1,000 human genome sequence" following completion of the Human Genome Project[15] and proposed actions to facilitate genomic medicine implementation in 2011[16]; in these cases, both the risks of failure and the benefits of success were high. Once again, breaking down barriers, as highlighted in Box 3 and detailed below, would accelerate progress and create new research and clinical opportunities at The Forefront of Genomics.

Laboratory and computational technologies

Advances in DNA synthesis and genome editing allow the field of genomics to progress from largely observational ("reading DNA") to more experimental ("writing" and "editing" DNA) approaches. Enabling true "synthetic genomics" (i.e., the synthesis, modification, and perturbation of nucleic acid sequences at any scale) will allow for more powerful experimental testing of hypotheses about genome variation and function and improve opportunities for linking genotypes to phenotypes[75]. Genome editing is increasingly being used for practical applications in medicine (e.g., in gene therapy[76]), biotechnology, agriculture, and other areas. Despite recent triumphs, however, the current approaches are limited in their ability to interrogate genome function at the pathway or network level and to study gene regulation, chromosome organization and mechanics, and other important phenomena that involve factors acting across large chromosomal (or genomic) distances. Furthermore, radically new capabilities for understanding how the full complement of genomic variation within any individual genome contributes to phenotype should be pursued. Innovative approaches for generating nucleic acid molecules with defined sequences and of any size, coupled with technologies that allow for the concurrent and large-scale perturbation of multiple genes or simultaneous examination of multiple genomic variants, would be transformative. These advances would benefit from the development of methods for introducing large synthetic constructs into mammalian cells.

In recent years, large human genomics projects have often relied on data generated as part of existing research studies, and emerging approaches involve developing biobanks and organized cohorts[77–79]. Meanwhile, direct-to-consumer (DTC) companies are generating substantial amounts of genomic data, and those efforts are rapidly being eclipsed by that being generated in the clinical care setting[80]. Properly leveraged, these DTC and clinical data offer opportunities for genomics-based studies at unprecedented scales; however, these data are often heavily fragmented, siloed, and mostly outside the purview of genomics researchers and their typical funders[81]. Eliminating the barriers to accessing these sources of data for conducting research is essential, but this will require resolving issues related to governance, policy infrastructure, and informatics and workflow solutions. Approaches are needed to mitigate the resulting gaps, limitations, and biases within this highly distributed data environment (e.g., with regards to population diversity, data-collection strategies, data standards, and data privacy), all while addressing concerns of the patients, participants, and groups. These challenges must be addressed globally[81] (Box 1), so as to accommodate differences in healthcare systems and views about data privacy. In addition, the healthcare stakeholders should take advantage of opportunities offered by genomics, thereby enabling virtuous-cycle routes between genomic learning healthcare systems and basic genomics research[82] (Fig. 3).

Biological insights

Despite progress in identifying genomic variants that cause monogenic traits or are statistically associated with complex phenotypes, connecting specific variants to phenotypes remains challenging[83]. Systematic approaches, including new tactics that connect high-throughput molecular readouts of functional genomic assays to organismal phenotypes, are required for establishing the phenotypic consequences of all genomic variants – individually

and in combinations – in a cell-type context across the life span[84]. Progress in this area requires global collaboration[85], advances in integrating multiple data types and performing perturbation assays, protein localization/interaction experiments, and animal models, as well as resources cataloging information about the fitness consequences of de novo mutations and the clinical relevance of genomic variants[83]. Because it is not possible to directly test every variant in all cell types and states, developmental stages, and disease processes, new data-collection strategies and analytical approaches are needed that can generalize and adapt predictions to new contexts, handle sparse data, and prioritize variants for experimental follow-up.

Recent advances have led to a greater appreciation of the extent of mosaicism – i.e., genomic variation among cells (both somatic and germline) within an individual. While there have been remarkable advances in understanding the somatic genomic changes encountered in cancer[86], there is a paucity of detailed knowledge about other impacts of mosaicism beyond a few well-studied examples[87]. Important areas of future research include investigating the prevalence and extent of different forms of mosaic variation in both nuclear and mitochondrial DNA, the mechanisms that generate mosaicism, and the roles of mosaicism in physiology and human disease. Such efforts might reveal if this form of genomic variation contributes to variable penetrance and expressivity, comprises a form of genetic epistasis, explains any currently undiagnosed diseases or sporadic cases (or apparent phenocopies) of known inherited diseases[9], and can inform the design of therapies for genetic diseases. Single-cell genomic technologies have extended knowledge about the functional impact of mosaicism in multiple experimental systems[88,89], with the next challenge being to translate such single-cell understanding to in vivo settings. The development of new laboratory and clinical approaches for readily detecting genomic mosaicism at high spatial and temporal resolution, especially in ways that are relatively non-invasive (e.g., requiring minimal amounts of tissue), would be catalytic.

Implementation science

A critical barrier to using genomics for improving health and preventing disease is the lack of clinical uptake of proven genomic interventions. Implementation science approaches are needed to identify the most effective methods and strategies for facilitating the use of evidence-based genomic applications, most notably pharmacogenomics-based selection of medications[90], in routine clinical care. Novel experimental designs, such as genotype-specific participant recruitment[91] or integration of patient-provided genomic data[92] (captured during previous healthcare encounters or from direct-to-consumer sources), should be explored for their potential to speed adoption and limit costs. The effectiveness of centralized resources for genomic referrals [e.g., genomic medicine specialists, consult services[93,94], centers of excellence in undiagnosed diseases (akin to transplantation centers or cancer centers)] should be explored as potential steppingstones to the more generalized uptake of genomics in clinical care. Strategies for deploying the limited workforce of highly trained genetics/genomics specialists (e.g., systematic referral networks or telemedicine/telecounseling) should also be evaluated for their effectiveness at increasing the availability of services broadly – as opposed to being limited to select, highly specialized centers.

Universal newborn genetic screening may represent the most visible and successful approach to population-based identification of serious and treatable inherited conditions, but population screening across the lifespan for other genetic conditions is less widely accepted. Standard public health screening approaches for the U.S. Centers for Disease Control and Prevention Tier 1 conditions[95,96] (e.g., Lynch syndrome, hereditary breast and ovarian cancer, and familial hypercholesterolemia) identify people at risk through blood relatives of affected individuals (referred to as "cascade testing" by geneticists[97]). Implementation research methods, coupled with effective science communication, are primed for optimizing approaches for engaging individuals in genetic testing for these disorders and also other emerging indications, such as genetic predisposition to adverse drug effects (pharmacogenomics), carrier testing of prospective parents, use of PRSs in disease detection and prevention[46], and genomic indicators (e.g., gene-expression and epigenetic patterns) of exposure to infectious pathogens[98] and other environmental agents.

Compelling genomics research projects

The field of genomics has routinely benefited from a willingness to articulate ambitious – often audacious – research efforts that aim to address questions and acquire knowledge that (at the time) may seem out of reach. Such boldness has served to stimulate interest in emerging opportunities, recruit new expertise, galvanize international collaborations involving multiple funders, and propel the field forward. While by no means comprehensive, the areas highlighted in Box 4 and detailed below illustrate the broadening range of compelling research projects that are ripe for pursuit at The Forefront of Genomics.

Advances in understanding gene regulation[5,24], the myriad functional roles of RNA[99], and the multi-dimensional nature of the nucleome[100] – coupled with the utility of single-cell genomic approaches[30,31] and anticipated new technological and computational capabilities for analyzing genomic datasets and variants – provide an unprecedented opportunity for deciphering the individual and combined roles of each gene and regulatory element. This must start with establishing the function of each human gene, including the phenotypic impact of human gene knockouts. Because genes and regulatory elements do not function in isolation, it is imperative to build robust experimental and computational models that deduce causal relationships and accurately predict cellular and organismal phenotypes using pathway and network models[101,102]. Novel analysis methods must address functional redundancy as well as the nearly boundless experimental space and complexity, including cell states and fates, temporal relationships, environmental conditions, and individual genetic background.

Building on the recent successes in unraveling the genetic underpinnings of rare and undiagnosed diseases[9], the field is poised to gain a more comprehensive understanding of the genetic architecture of all human diseases and traits[10,85]. However, myriad complexities can be anticipated. For example, any given genomic variant(s) may affect more than one disease or trait (i.e., pleiotropy); can confer disease risk or reduce it; and can act additively, synergistically, and/or through intermediates. New methods for analyzing data that account for human diversity[103], coupled with a growing clarity about genotype-phenotype relationships, must be developed to deduce associations and interactions among genomic

variants and environmental factors, improve estimates of penetrance and expressivity, and enhance the clinical utility of genomic information for predicting risk, prognosis, treatment response, and, ultimately, clinical outcomes.

Prioritizing the generation of genomic and corresponding phenotypic data from ancestrally diverse participants is a scientific imperative[104] and essential for achieving equitable benefits from genomic advances[105] (Box 1). However, this is an area in which genomics has repeatedly fallen short[19], leading to missed opportunities for understanding genome structure and function, identifying variants conferring risk for common diseases[106], and implementing genomic medicine for the benefit of all[107–109]. Ideally, studies should be designed for different groups, tailored for local sensibilities and situations, and consistent in capturing key information beyond participants' ancestry (e.g., the physical and social environments in which they live and receive healthcare[110]). Leveraging new insights from studies of diverse populations will require the development of new and robust methods for identifying novel signatures of natural selection, performing genotype imputation, mapping disease loci, characterizing genomic variant pathogenicity, and calculating PRSs[103,109]. Success in these efforts will yield a more-complete understanding of how the human genome functions in different environments and offer benefit to those participating in genomics research. Attaining the level of population diversity that will truly benefit all people requires bold scientific and community-based leadership, dedicated resources from funders, highly committed researchers, and effective partnerships that earn the trust of diverse groups of participants and their communities.

As genomics has grown in medicine and society, its potential to influence people's actions has also expanded. Increasingly, genomics has affected concepts of health, disease, responsibility, family, identity, and community, raising a number of important and changing questions. When and how is genomic information shared and communicated within families[111]? Will the identification of a strong genetic risk for a disease change a person's perception of their health or others' perception of that person? With some genetic risks being more common in certain identifiable populations, what role does group affiliation play in how risk is communicated and perceived, including potential group stigmatization? Research that catalogs, analyzes, and measures the impact of genomics on individuals, families, and communities is important for providing a more informed context to avoid future misrepresentations, misunderstandings, and misuses of genomics[54]. Finally, researchers must appreciate how their own backgrounds and experiences shape their interpretations of genomic data[112].

Extending genomics research in clinical settings beyond DNA sequence to include other multi-omic data, together with clinical variables and outcomes, would advance understanding of disease onset and progression and may also prove important for drug-discovery efforts[113,114]. This would require tissue- and cell-specific analyses that integrate these data, providing real-time snapshots of biological and disease processes. For clinical applicability and adoption, these high-dimensional, multi-omic data should be integrated with clinical decision support tools and electronic health records. Ultimately, such efforts could reveal important relationships among genomic, environmental, and behavioral

variation and facilitate a transition of the use of genomics in medicine from diagnosing and treating disease to maintaining health.

Sharp barriers between research and clinical care obstruct the virtuous cycle of moving scientific discoveries rapidly into clinical care and bringing clinical observations back to the research setting[82] (Fig. 3). Learning healthcare systems – in which real-time data on outcomes of healthcare delivery are accessed and used to enhance clinical practice – can lead to continuous care improvement, but only if the barriers between research and clinical care are reduced[115]. For example, offering genome sequencing to all members of a healthcare system, performed in conjunction with research and participant engagement and provided in real time[81], could help to assess the clinical utility of genomic information and may allow providers to improve disease diagnosis and management. System-wide implementation of such an experiment requires not only extensive patient and provider education, sophisticated informatics capabilities, and genomics-based clinical decision support, but also the development and evaluation of data security and privacy protections to ensure patient confidentiality[116]. Patients should be engaged in the design of such systems and informed at entry to them (and periodically thereafter), so as to be fully aware of the nature of the ongoing research with their clinical data and the goals and potential risks of their participation[117]. Extending such studies across multiple healthcare systems should reveal common challenges and solutions[118,119], thereby enhancing the learning healthcare model for genomic medicine more broadly (Fig. 3).

Concluding thoughts

The dawn of genomics featured the launch of the Human Genome Project in October 1990[1]. Three decades later, the field's journey has included stunning technological advances and high-profile programmatic successes, which in turn have led to the widespread infusion of genomic methods and approaches across the life sciences and, increasingly, into medicine and society.

NHGRI has for the third time[15,16] since the Human Genome Project undergone an extensive horizon-scanning process to capture, synthesize, and articulate the most compelling strategic opportunities for the next phase of genomics – with particular attention to those elements most relevant to human health. The now near-ubiquitous nature of genomics (including in the complex healthcare ecosystem) presented practical challenges for attaining a holistic assessment of the field. Another reality was that NHGRI's investment in genomics has now been multiplied many-fold by the seeding of human genomics throughout the broader research community. These changes reflect a continued maturation of both the field (in general) and NHGRI (more specifically), nicely aligning with the institute's evolving leadership role at The Forefront of Genomics.

Embracing that role, NHGRI formulated the strategic vision described here, which provides an optimistic outlook that the successes in human genomics over the past three decades will be amplified in the coming decade. Many of the details about what is needed to fulfill the promise of genomics have now come into focus. Major unsolved problems remain – among them determining the role for the vast majority of functional elements in the human genome

(especially those outside of protein-coding regions), understanding the full spectrum of genomic variation (especially that implicated in human disease), developing data-science capabilities (especially those that keep pace with data generation), and improving healthcare through the deployment of genomic medicine (especially in the areas of prevention, diagnosis, and therapeutic development). The new decade also brings new research questions related to the societal implications of genomics, including those related to social inequities, pointing to the continued importance of investigating the ethical, legal, and social issues related to genomics. But now more than ever, solutions to these problems seem to be within striking distance. Towards that end (and with the characteristic spirit of genomics audacity), we offer ten bold predictions of what might be realized in human genomics by 2030 (Box 5).

The strategic vision articulated here was crafted on behalf of the field of human genomics and emphasized broad strategic goals opposed to implementation tactics. The realization of these goals will require additional planning in conjunction with the collective creativity, energies, and resources of the global community of scientists, funders, and research participants. NHGRI has taken some initial steps for implementing this vision, although these will inevitably need to be adapted as advances occur and circumstances change. Indeed, the final words of this strategic vision were formulated as the world moved urgently to deal with the COVID-19 pandemic (see Epilogue), providing a vivid reminder of the need to be nimble and the importance of nurturing all parts of the research continuum – from basic to translational to clinical – for protecting public health and advancing medical science.

Despite the seismic changes seen in genomics since the inception of the field, the fundamental sense of curiosity, marvel, and purpose associated with genome science seems to be timeless. In concluding NHGRI's previous strategic vision[16] – published just under a decade ago – the then-envisioned opportunities and challenges were provided with "… a continuing sense of wonder, a continuing need for urgency, a continuing desire to balance ambition with reality, and a continuing responsibility to protect individuals while maximizing the societal benefits of genomics…." With the 2020 strategic vision described here providing a thoughtful guide and with enduring feelings of wonder, urgency, ambition, and social consciousness providing unfettered momentum, we are ready to embark on the next exciting phase of the human genomics journey.

Epilogue: COVID-19 and genomics

SARS-CoV-2 emerged as global threat to public health at the end of the multi-year process that generated the above strategic vision. Nonetheless, the pandemic provides a potent lesson about how a tiny string of nucleic acids can wreak global havoc on humankind. Understanding the mechanisms involved in the virus's transmission, invasion, and clearance, as well as the highly variable and at times disastrous physiologic responses to it, are fertile grounds for genomics research. Indeed, genomics rapidly assumed critical roles in COVID-19 research and clinical care in areas such as the (1) deployment of DNA- and RNA-sequencing technologies for diagnostics, viral isolate tracking, and environmental monitoring; (2) use of synthetic nucleic acid technologies for studying SARS-CoV-2

virulence and facilitating vaccine development; (3) examination of how human genomic variation influences infectivity, disease severity, vaccine efficacy, and treatment response; (4) adherence to principles and values related to open science, data sharing, and consortia-based collaborations; and (5) provision of genomic data science tools for studying COVID-19 pathophysiology. The growing adoption of genomic approaches and technologies into myriad aspects of the global response to the COVID-19 pandemic serves as another important and highly visible example of the integral and vital nature of genomics in modern research and medicine.

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Box 1

Guiding principles and values for human genomics

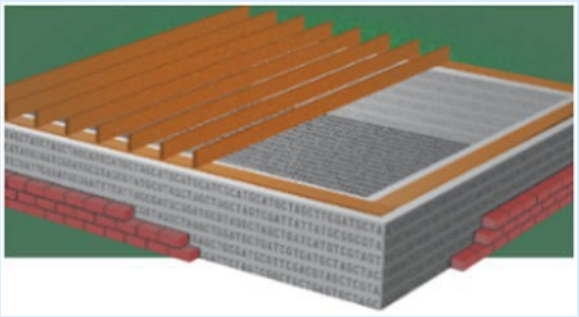
- **Maintain an overarching focus on using genomics to understand biology, to enhance knowledge about disease, and to improve human health** – *genomics is now foundational across the entire continuum of biomedical research, from deciphering fundamental principles of biology to translating that knowledge 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, including 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 utility 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, including individuals from groups 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 influencing 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 (e.g., those for generating, analyzing,*

storing, and sharing data) has benefited genomics in significant 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 endeavors 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 novel 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

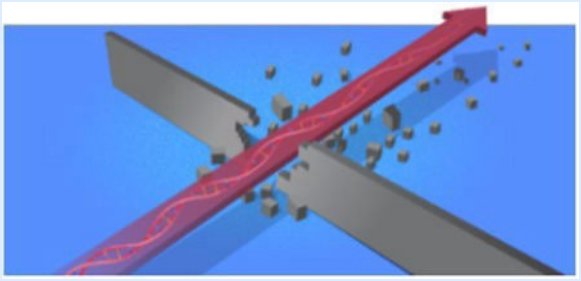
- Understand the interrelationships between genomics and the social and environmental factors influencing human health
- Empower people to make well-informed decisions about genomic data and develop data-stewardship systems that reinforce their choices
- Increase the genomic literacy of all sectors of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

Box 3

Breaking down barriers that impede progress in genomics



Laboratory and computational technologies

- Transform the study of the functional consequences of genomic variation by enhancing the scale of DNA synthesis and editing
- Maximally leverage the usability and utility of emerging datasets for genomic studies of human health and disease

Biological insights

- Establish the means to determine the functional consequences of genomic variants affecting human health and disease
- Characterize intraindividual genomic variation and understand its role in human disease

Implementation science

- Develop and assess strategies for implementing the use of genomic information in clinical care
- Test public health approaches for implementing population-wide genomic screening

Box 4

Compelling genomics research projects in biomedicine

- Acquire an increasingly comprehensive view of the roles and relationships of genes and regulatory elements in pathways and networks
- Elucidate the genetic architecture of the majority of human diseases and traits
- Design studies that include diverse ancestral populations to enable scientific discoveries and genomic medicine for all
- Understand how the use of genomics can influence concepts of health, disease, responsibility, identity, family, and community
- Extend multi-omic studies of human disease and health into clinical settings
- Design and utilize genomic learning healthcare systems for knowledge generation and clinical care improvement

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. While 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 analyzing 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 impact of genotype on phenotype.
4. Research in human genomics will have moved beyond population descriptors based on historic social constructs such as race.
5. Studies involving 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 genomic testing as routine as complete blood counts (CBCs).
7. The clinical relevance of all encountered genomic variants will be readily predictable, rendering the diagnostic designation “variant of uncertain significance (VUS)” obsolete.
8. A person’s complete genome sequence along with informative annotations can be securely and readily accessible on their smartphone.
9. Individuals from ancestrally diverse backgrounds will benefit equitably from advances in human genomics.
10. Genomic discoveries will lead to curative therapies involving genomic modifications for dozens of genetic diseases.

Fig. 1 |. Four-area strategic framework at The Forefront of Genomics.
Together, the indicated progressive and interrelated areas serve to organize the major elements in the strategic vision described here.

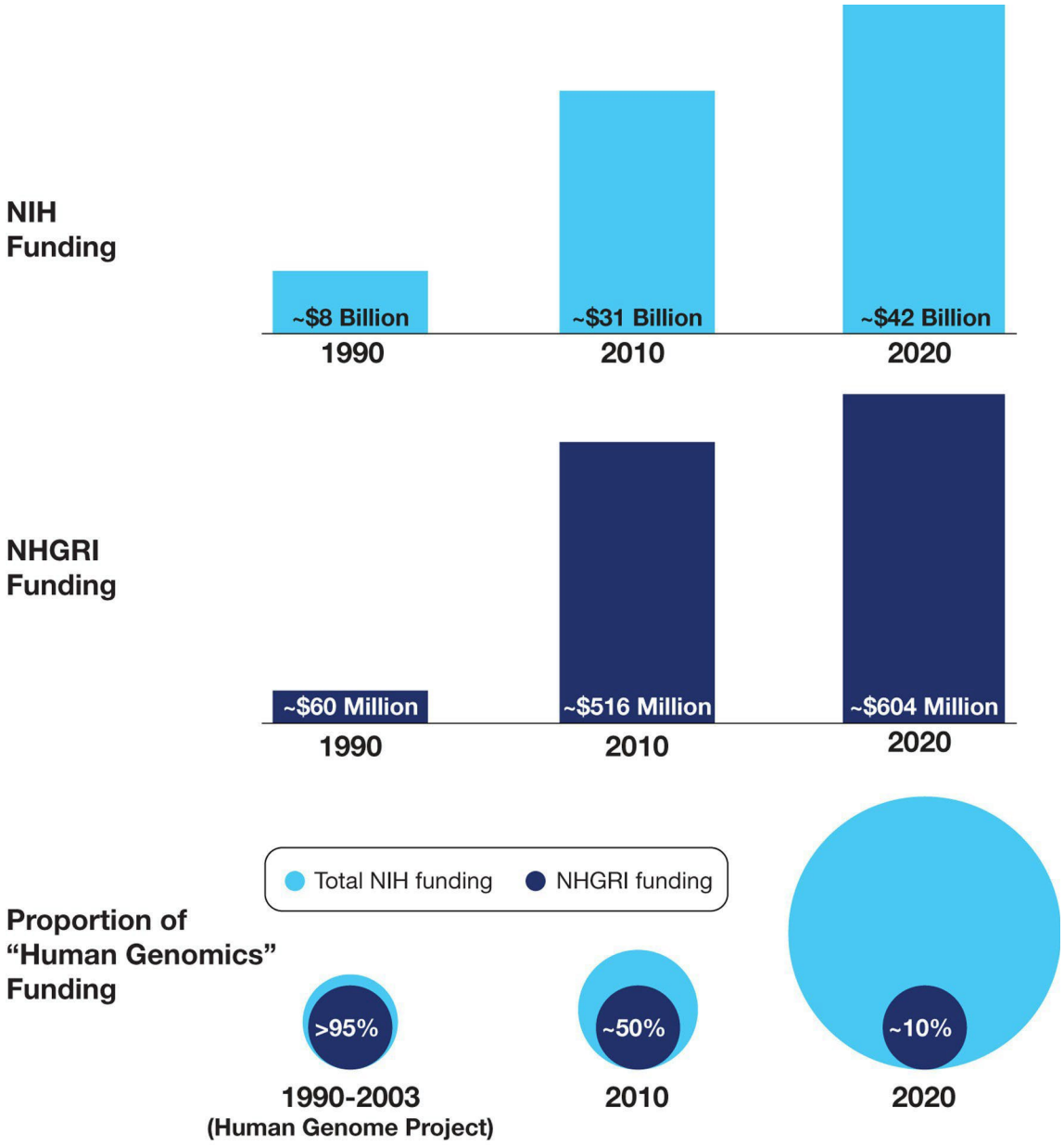


Fig. 2 |. Funding trends of NIH and NHGRI over the past 30 years.

The total funding levels for NIH (top panel) and NHGRI (middle panel) are indicated for 1990, 2010, and 2020, respectively. Also shown (bottom panel) is the relative proportion of funds supporting human genomics research provided by NHGRI versus all of NIH for the three corresponding time intervals (as derived from queries of the internal NIH Research, Condition and Disease Categorization database for funds assigned to the "human genome" category). During the 30-year period when the NHGRI budget increased roughly ten-fold (middle panel), the proportion of total NIH funding for human genomics research actually increased more dramatically, from <5% during the Human Genome Project to ~90% at the beginning of the current decade (bottom panel). In essence, these trends reflect a leveraging

of NHGRI's funds that increased NIH's overall human genomics research funding by greater than ten-fold.

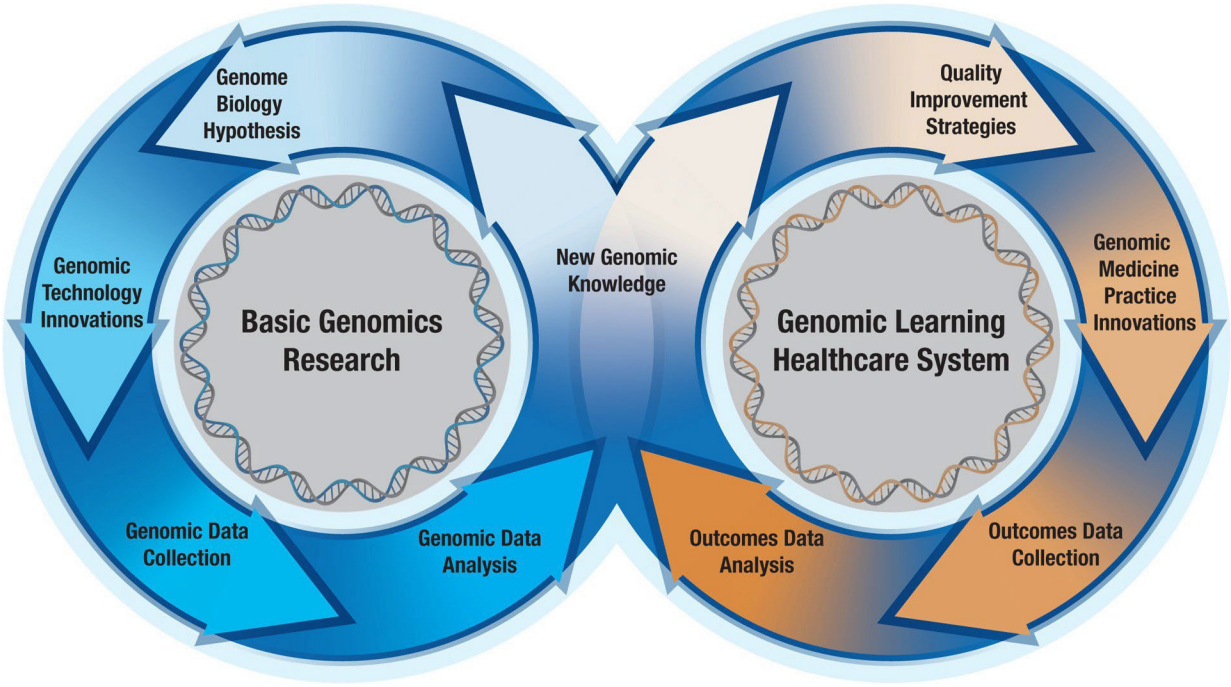


Fig. 3 |. Virtuous cycles in human genomics research and clinical care.

As human genomics has matured as a discipline, productive and connected virtuous cycles of activity have emerged, each self-improving with successive rounds of new advances. The cycle on the left reflects basic genomics research, in which technology innovations spur the collection and analysis of genomics research data, often yielding new knowledge and additional hypotheses for testing. The cycle on the right reflects a genomic learning healthcare system, in which the implementation of new genomic medicine practice innovations allows for the collection and analysis of outcomes data, often yielding new genomic knowledge and additional genomics-based strategies for improving the quality of clinical care. Note that the new knowledge emerging from either the left or right cycle has the potential to feed into the other, creating opportunities for "bench to bedside" and "bedside back to bench" progressions[82] – both of which are expected to grow in the coming decade.

National Human Genome Research Institute

Building a Diverse Genomics Workforce: An NHGRI Action Agenda

The Forefront of Genomics®

Letter from the Director

Data show that enhancing the diversity of scientific teams produces better ideas and more creativity. Developing teams of individuals from diverse backgrounds, however, means recognizing that some groups are underrepresented in the biomedical workforce. These groups include individuals from underrepresented racial and ethnic groups, individuals with disabilities, individuals from disadvantaged backgrounds, and women in staff, faculty, and leadership positions in the biomedical, clinical, behavioral, and social sciences research enterprises as well as in healthcare professions.

At present, the genomics workforce lacks diversity and, in fact, poorly reflects the make-up of the U.S. population. To address this in a meaningful way, a serious commitment of attention and resources is needed. The National Human Genome Research Institute (NHGRI) is prepared to make and sustain such a commitment. Towards that end, in late 2019, I charged an internal working group to develop an NHGRI plan aimed at improving the diversity of the genomics workforce. Chaired by Vence Bonham, J.D., my senior advisor on genomics and health disparities, the working group developed this document: *Building a Diverse Genomics Workforce: An NHGRI Action Agenda*.

This "action agenda" commits NHGRI to meaningfully enhance the diversity of the genomics workforce by 2030. This plan articulates goals and objectives to increase the number of individuals from diverse backgrounds, including underrepresented groups, in genomics. Specific programs and strategies will increase the number of individuals from diverse backgrounds, including underrepresented groups, who have the necessary training to pursue careers in genomics. New support will help early stage scientists from diverse backgrounds achieve independent research careers and foster pathways to leadership positions in genomics. Enhanced attention will also be given to genomics training for those in clinical and healthcare career tracks. The Institute's existing training programs that focus on enhancing the diversity of the genomics workforce will also be expanded. Our Institute promises to work with our community to change the culture where needed and create environments that will sufficiently support this new and more inclusive vision of a genomics workforce.

The genomics enterprise is at an exciting juncture with extraordinary career opportunities to improve human health through basic, translational, and clinical research. The realization of genomic medicine will require increasing genomics expertise among different healthcare professionals. NHGRI has the responsibility to facilitate the well-being of the genomics workforce, and that includes attracting a workforce that is diverse.

I endorse this action agenda and am eager to see it implemented across the Institute. As pointed out in the 2020 NHGRI Strategic Vision ("Strategic vision for improving human health at The Forefront of Genomics," *Nature* 586:683-692, 2020), the Institute sees the development of a diverse genomics workforce as a guiding value — something fundamental to the Institute and to the field.

I am grateful to the working group for their efforts and to those who contributed ideas and information that fed into the process of establishing this action agenda.

Eric Green, M.D., Ph.D.
Director, NHGRI

Background

> "The promise of genomics cannot be fully achieved without successfully attracting, developing, and retaining a diverse workforce that includes people from groups currently underrepresented in the genomics enterprise."
>
> – Eric Green, M.D., Ph.D.

Today, the genomics workforce does not reflect the diversity of the U.S. population. Research has documented that having an inclusive scientific workforce is necessary to increase innovation and creativity and to enhance performance in solving scientific problems.[1] When the scientific workforce is diverse, the variety of research topics that are explored increases substantially, which leads to more discoveries that benefit the biomedical community overall.[2]

This document reflects a new NHGRI-formulated action agenda that establishes goals, objectives, and implementation strategies to enhance the diversity of the genomics workforce. For this action agenda, the word "diversity" is used to mean individuals from diverse backgrounds, including individuals from groups identified as underrepresented in biomedical, clinical, behavioral, and social sciences. In the 2019 National Institutes of Health (NIH) announcement of the agency's interest in diversity, NIH identified underrepresented groups to include individuals from underrepresented racial and ethnic groups, individuals from disadvantaged backgrounds, and individuals with disabilities, as well as women at senior faculty level.

Barriers to diversity in the biomedical workforce occur at various stages of career progression, but additional studies are needed to fully understand those challenges. For example, while the number of doctoral degrees earned by women and individuals from other underrepresented groups in science has increased, this has not led to an increase in the diversity of the biomedical research workforce at later career stages, including those associated with faculty positions and the acquisition of grant funding.[3-5] Moreover, data on individuals with disabilities are lacking.[6] The major challenges associated with enhancing the diversity of the biomedical workforce have been documented.[7-16] A recent study found that Ph.D. recipients from underrepresented groups in the U.S. produce novel contributions in their dissertations, but their novel contributions are too-often devalued and discounted.[17]

Reducing such barriers to career opportunities in biomedical research for underrepresented groups requires new strategies, programs, and creative approaches that promote workforce diversity, inclusion practices, and leadership in the field.

The NIH identified its interest in workforce diversity in a 2019 statement:

> Every facet of the United States scientific research enterprise – from basic laboratory research to clinical and translational research to policy formation – requires superior intellect, creativity and a wide range of skill sets and viewpoints. NIH's ability to help ensure that the nation remains a global leader in scientific discovery and innovation is dependent upon a pool of highly talented scientists from diverse backgrounds who will help to further NIH's mission.
>
> Research shows that diverse teams working together and capitalizing on innovative ideas and distinct perspectives outperform homogenous teams. Scientists and trainees from diverse backgrounds and life experiences bring different perspectives, creativity, and individual enterprise to address complex scientific problems. There are many benefits that flow from a diverse NIH-supported scientific workforce, including: fostering scientific innovation, enhancing global competitiveness, contributing to robust learning environments, improving the quality of the research, advancing the likelihood that underserved or health disparity populations participate in, and benefit from health research, and enhancing public trust.

NHGRI commits to NIH's vision and calls on the broader genomics community — including academic institutions, industry, healthcare systems, and professional societies — to join the Institute in committing to substantially enhancing the diversity of the genomics workforce in the coming decade. This action agenda establishes NHGRI goals to develop innovative new programs, support current successful programs, and partner with others to substantially enhance the diversity of the genomics workforce. Overall, the objectives include both reducing barriers to training opportunities in the field and supporting the development and career progression of researchers from diverse backgrounds, including underrepresented groups.

In October 2020, NHGRI published a new Strategic Vision for the field of genomics aimed at accelerating scientific and medical breakthroughs.[18] That Strategic Vision maintains that the field needs new strategies and programs to enhance career opportunities in genomics, and these must include new and creative approaches to promote workforce diversity, leadership in the field, and inclusion practices. As part of the process that led to the development of the 2020 NHGRI Strategic Vision, the Institute established an internal Genomic Workforce Diversity Working Group (see Appendix for roster of members). The working group was charged with developing a 10-year action agenda for *Building a Diverse Genomics Workforce* (hereafter referred to as the *NHGRI Action Agenda for a Diverse Genomics Workforce*) to guide the Institute's efforts. To inform the development of this action agenda, the working group gathered stakeholder feedback by seeking public comments and interviewing leaders from research universities and professional societies as well as early career genomics professionals. NHGRI is committed to funding programs that are designed to enhance workforce diversity, and this will include establishing tangible metrics; partnering with academic institutions, industry, and professional societies; and preparing the next generation to join the research and clinical genomics workforce.

NHGRI has a history of funding diversity training programs. The NHGRI Diversity Action Plan (DAP) program was established in 2002 and has been helpful in diversifying the pool of scientists who are prepared to pursue genomics-related careers. The program was originally associated with NHGRI's large extramural research initiatives, including the Centers of Excellence in Genomic Science, Genome Sequencing Centers, and Model Organism Databases (MODs). Since its inception, the DAP program has included over 1,400 trainees across 20 projects. In 2014, NHGRI expanded the DAP program in several ways in an effort to encourage applicants to propose innovative educational programs that help enhance the diversity of genomics trainees. While DAP proposals are no longer limited to being associated with certain NHGRI programs, they must still fit within the NHGRI's scientific mission.

At an NIH-wide level, NHGRI participates in several extramural programs to promote diversity in the research workforce, including the Maximizing Opportunities for Scientific and Academic Independent Careers Transition Award to Promote Diversity (MOSAIC) program, the Research Supplements to Promote Diversity in Health-Related Research program, and the Ruth L. Kirschstein National Research Service Award Individual Predoctoral Fellowship to Promote Diversity in Health-Related Research. A listing of all funding opportunities can be found on NHGRI's Funding to Promote Diversity in the Genomics Workforce website. Additionally, in 2019, NHGRI was pleased to partner with the American Society of Human Genetics to support the Human Genetics Scholars Initiative.

NHGRI has also established informal science and education programs to facilitate the training of secondary school teachers, community college staff, and tribal college and university faculty in genomics.[19] Developing a pathway that begins with pre-college education programs sets a precedent and illustrates that NHGRI is committed to preparing students to enter the genomics workforce and fostering a genomically literate public.

The promise of genomics cannot be fully achieved without successfully attracting, developing, and retaining a diverse researchworkforce that includes people from groups that are underrepresented in the genomics enterprise. NHGRI recognizes that this will require the Institute to make a long-term commitment to accelerating the diversity of the genomics workforce.

Overview

To be at the forefront of efforts to enhance the diversity of the genomics workforce, the *NHGRI Action Agenda for a Diverse Genomics Workforce* has the following four major goals:

NHGRI is committed to both short- and long-term strategies for implementing the goals of the *NHGRI Action Agenda for a Diverse Genomics Workforce*, which includes evaluating progress using defined metrics. The Institute will use this agenda's goals, objectives, and implementation strategies to develop appropriate programs that, if successful, will substantially enhance the diversity of the genomics workforce by the end of this decade.

GOAL 1:

Develop and support initiatives that provide early exposure and access to careers in genomics.

As the population of students in the U.S. education system becomes more diverse, we must invest in steps to ensure they have the opportunity to become part of the genomic workforce. Pursuing a career in genomics usually entails early exposure to and interest in science, technology, engineering, and mathematics (STEM). To achieve meaningful preparation, students need foundational courses to enter undergraduate programs and pursue genomics-related majors. Waiting until students reach post-secondary education is often too late, especially for students with limited access to educational resources. Thus, NHGRI and the field of genomics must invest in programming for these pre-college students and the educators who teach them.

Objectives

1.1 : Identify best practices in programming designed to provide early exposure to genomics, including barriers and recommendations to eliminate those barriers

1.2 : Support and participate in programs that are designed to encourage individuals of diverse backgrounds to pursue genomics careers, especially for persons from groups who are historically underrepresented in science

Implementation Strategies

- Identify best practices that encourage the pursuit of and early exposure to genomics careers through the support of outreach and engagement programming, resources, and partnerships (1.1)
- Support and design outreach and engagement programs that enlist professional societies, academic institutions, industry, and community organizations to develop new approaches that bring awareness to opportunities and careers in genomics (1.1 & 1.2)
- Create resources and educational campaigns that are designed to bring awareness to opportunities for careers in genomics (1.2)
- Support STEM education, training, and professional development programs for diverse communities that are designed to demonstrate the applications of and breadth of professional opportunities in genomics (1.1 & 1.2)

GOAL 1:
Develop and support initiatives that provide early exposure and access to careers in genomics.

Indicators of Success

- Create best practices that can be incorporated into future programs designed to encourage the pursuit of genomics careers (1.1)
- Identify new approaches that bring awareness to opportunities and careers in genomics (1.1)
- Participate in and implement outreach programs using best practices (1.2)
- Use and distribute resources from, and participation in, campaigns that are designed to bring awareness to opportunities and careers in genomics (1.2)
- Increase the participation of individuals from diverse backgrounds (or those who teach individuals from diverse backgrounds) in STEM education, training, and professional development programs that are designed to demonstrate the applications of and breadth of opportunities in genomics (1.1 & 1.2)

Since 2003, NHGRI's Education and Community Involvement Branch has sponsored the annual Short Course in Genomics. The course is designed for pre-college and college educators from across the United States who teach genetics, genomics, biology, or other STEM courses. The course supports NHGRI's efforts to enhance the diversity of the genomics workforce through the integration of genomics into secondary and collegiate classrooms, including those classrooms with significant numbers of students from communities that have been historically underserved and underrepresented in genomics. Since its inception, there have been over 250 course participants; these individuals teach at middle and high schools, tribal colleges and universities, and two-year and four-year institutions.[19]

GOAL 2:

Develop and support training programs and networks that connect undergraduate and graduate education to careers in genomics.

The pathway that leads from early STEM education through graduate-level degrees in genomics has several key transition points. For those community college students and undergraduates from diverse backgrounds, including those from underrepresented groups who are interested in science, attention in the form of guidance and resources must go to where the students are concentrated, which will lead them to and through graduate genomics training programs. NHGRI is dedicated to supporting the development of graduate-level genomics training programs that mentor and support diverse cohorts of students.

Objectives

2.1 : Create a systematic network of support for students from diverse backgrounds, including those from underrepresented groups, as they move to and through graduate training programs in genomics

2.2 : Ensure that undergraduate minority-serving institutions (MSI) and community colleges are aware of and tightly connected to this network

2.3 : Encourage inclusive climates at all leading graduate-level genomics training programs so as to mentor and promote cohorts of individuals from diverse backgrounds, including underrepresented groups

Implementation Strategies

- Ensure that NHGRI's undergraduate and graduate-level training programs work together to provide continuous support to individuals as they move to and through graduate school (2.1)
- Proactively connect STEM programs at MSIs and community colleges to the network of training programs sponsored by NHGRI (2.1 & 2.2)
- Support cohort models of students within and across programs (2.1 & 2.3)
- Ensure that all graduate training programs in genomics that are supported by NHGRI include structured mentorship and career development plans for all participating trainees (2.3)
- Ensure that all graduate training programs in genomics that are supported by NHGRI establish or participate in integrated systems to address potential bias and faculty equity and reduce potential isolation of trainees, including those from underserved and underrepresented groups (2.1 & 2.3)
- Collect baseline data and support ongoing mixed-methods assessments on the inclusion of diverse trainees and research environments at major NHGRI-funded institutions with and without training programs (2.3)

GOAL 2:
Develop and support training programs and networks that connect undergraduate and graduate education to careers in genomics.

Indicators of Success

- Increase advancement of students from NHGRI-supported undergraduate and graduate training programs into postgraduate genomics careers
- At institutions where NHGRI develops new undergraduate training programs, measure whether those institutions consistently place students into graduate training programs in genomics or careers in genomics (including those supported by NHGRI)

"Perhaps the most important reason we have fewer students of color, African Americans and Latinos, in the scientific workforce is that most don't succeed at the undergrad level. If you look at our report from 2011 on expanding the scientific workforce, science and technology at the crossroads, what we found is that everybody wants to say, well it's K-12, well it's grad, it's whatever. We say no, all those are true but the lowest hanging fruit to make a big difference would be the undergraduate experience."

– Freeman Hrabowski, President of University of Maryland, Baltimore County

GOAL 3:

Develop and support training, career development, and research transition programs that lead to independent research and clinical careers in genomics.

The transition from formal education to research and clinical careers in genomics often requires overcoming barriers to become an established professional in the field. The retention of trained professionals who specialize in genetics and genomics is a major challenge despite the exciting scientific and health-related possibilities. Identifying key transition and retention barriers and developing intervention programs are first steps in achieving a more diverse genomics workforce.

Objectives

3.1 : Identify and reduce barriers for individuals from diverse backgrounds who want to enter research and clinical careers in genomics

3.2 : Facilitate the inclusion and retention of individuals from diverse backgrounds in research and clinical careers in genomics

Implementation Strategies

- Provide research funding to understand the unique barriers that underrepresented groups encounter when entering genomics research and clinical careers, and test interventions to eliminate such barriers (3.1 & 3.2)
- Build collaborations with academic institutions who train diverse student populations (3.1)
- Collaborate with professional societies, academic health centers, and clinical genetic programs that support the professional development of diverse genomics professionals (3.2)
- Support programs that expand the inclusion of diverse genomics professionals in the clinical and research workforce (3.2)
- Ensure that NHGRI staff members serve on trans-NIH workforce diversity committees/workgroups to enhance collaborations among NIH workforce diversity programs (3.1 & 3.2)
- Provide guidance to institutions and grantees to maintain inclusive climates to mentor and promote cohorts of genomics professionals from diverse backgrounds, including underrepresented groups (3.2)

Indicators of Success

- Increase the number of new or augmented research and clinical professional programs in genomics at MSIs (3.1 & 3.2)
- Enhance the diversity of the genomics research and clinical genomics workforce (3.1 & 3.2)
- Produce evidence-based interventions to reduce or eliminate barriers to entering and remaining in genomics-related positions (3.1)

"Although the dearth of underrepresented minority (URM) faculty members in medical schools typically has been framed as a 'pipeline' problem — i.e. a lack of available URM talent — our analysis shows that the rate of Ph.D. production for scientists from URM backgrounds has increased significantly over the past 33 years, and at a faster rate than that of well-represented (WR) scientists. Despite this progress, there was no statistical linkage between the size of the pool of URM talent, and the number of URM assistant professors hired in basic science departments of medical schools."

– Gibbs KD et al., *eLife*, 2016 [5]

GOAL 4:

Evaluate progress towards achieving greater diversity in the genomics workforce.

NHGRI has a long-standing interest in enhancing the diversity of the genomics workforce. Going forward, it will be important to evaluate the Institute's investments in this area to determine their effectiveness and, in turn, to guide changes and improvements that maximize their impact.

Objectives

4.1 : Establish a relevant set of metrics for evaluating NHGRI diversity training and career development programs

4.2 : Use these metrics to develop tracking protocols for all individuals supported by these training programs

4.3 : Assess all NHGRI training and career development programs, including diversity-targeted programs, with periodic reports to leadership

Implementation Strategies

- Use lessons learned from NHGRI's extramural training Data Analysis and Coordinating Center (DACC) and guidance from NIH leaders in training evaluations to help establish success metrics (4.1)
- Develop and implement plans to longitudinally monitor and track all trainees by program, both institutional and individual; use existing NIH reporting and tracking tools to the extent possible (4.2)
- Consider how to create comparison groups to track participating programs and individuals (4.2)
- Support assessments conducted by the staff who oversee the Institute's training programs (4.3)
- Seek advice from NHGRI's external advisory groups on metrics, tracking, and evaluation plans (4.1 & 4.2)
- Provide annual reports to NHGRI leadership and provide updates to relevant external advisory groups when warranted (4.3)

Indicators of Success

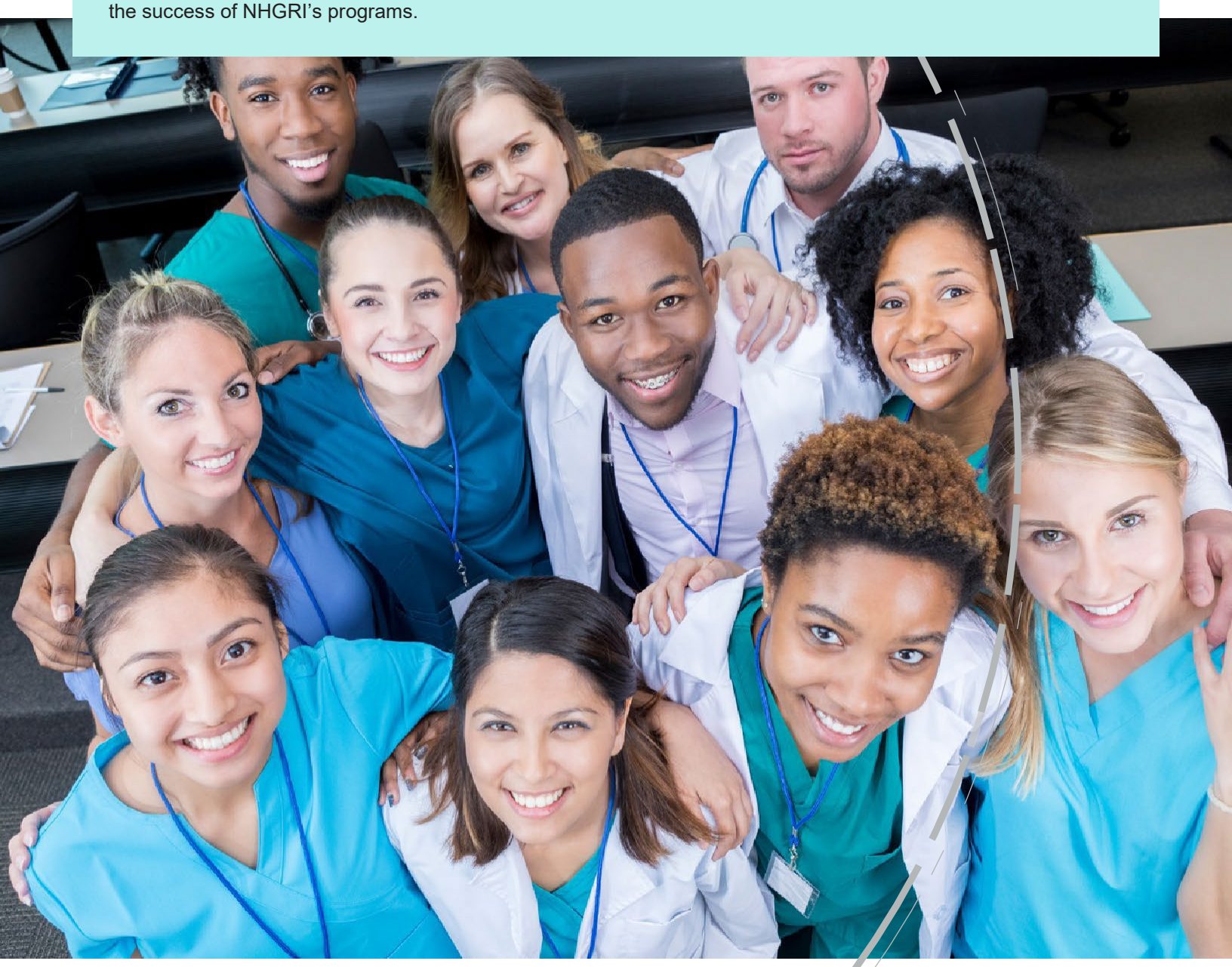
- Establish procedures to collect and evaluate data (4.1, 4.2, & 4.3)
- Conduct evaluations that guide changes/improvements to programs to further achieve greater diversity of the genomics workforce (4.3)

"NIH should develop quantitative metrics, evaluation details, and time frames to assess NIH's efforts to diversify its scientific workforce against its diversity strategic plan goals, and take action as needed."

– Government Accountability Office (GAO) Report (GAO-18-545)

Conclusion

NHGRI is prepared to work in partnership with the genomics community to enhance the diversity of the genomics workforce. This will require a long-term commitment, attention to the goals and objectives discussed in this action agenda, and continuously working with the broader community to develop new implementation strategies and evaluate the success of NHGRI's programs.



Appendix

Funding to Promote Diversity in the Genomics Workforce

Genomic Workforce Diversity Working Group Members

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The genomics workforce must become more diverse: a strategic imperative

Vence L. Bonham[1,*] and Eric D. Green[1,*]

Summary

The National Human Genome Research Institute (NHGRI) recently published a new strategic vision for the future of human genomics, the product of an extensive, multi-year engagement with numerous research, medical, educational, and public communities. The theme of this 2020 vision—The Forefront of Genomics—reflects NHGRI's critical role in providing responsible stewardship of the field of human genomics, especially as genomic methods and approaches become increasingly disseminated throughout biomedicine. Embracing that role, the new NHGRI strategic vision features a set of guiding principles and values that provide an ethical and moral framework for the field. One principle emphasizes the need to champion a diverse genomics workforce because "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." To build on the remarkable metamorphosis of the field over the last three decades, enhancing the diversity of the genomics workforce must be embraced as an urgent priority. Toward that end, NHGRI recently developed an "action agenda" for training, employing, and retaining a genomics workforce that reflects the diversity of the US population.

Introduction

The US biomedical research workforce, including the genomics workforce, falls dramatically short of reflecting the diversity of the nation's population.[1] As highlighted in a 2019 notice issued by the US National Institutes of Health (NIH), the lack of diversity reflects an underrepresentation of individuals from racial and ethnic groups, those with disabilities, and those from disadvantaged backgrounds as well as women in faculty and leadership positions. This problem exists in the biomedical, clinical, behavioral, and social sciences research arenas. The lack of diversity in the scientific and clinical workforce arises from a larger set of societal problems and has direct negative consequences on the conduct of research.

Studies have shown that enhancing the diversity of the research workforce fosters innovation and creativity, which arise from the variety of perspectives that emerge when not everyone is thinking in the same way. Groups with diverse life experiences provide different contexts and approaches for tackling challenging problems.[2–4] Hofstra and colleagues recently described the "diversity-innovation paradox in science" in a study that examined the career trajectories of nearly all US doctoral recipients from 1977 to 2015.[5] These researchers found that innovations and novel contributions to science from underrepresented individuals (gender and racial minorities) are more likely to be discounted.

NIH's efforts to increase the diversity of the biomedical research workforce have been ongoing for years, which led to multiple targeted approaches to increase the funding of underrepresented minority investigators.[6] In 2015, Valantine and Collins stated that "[d]espite longstanding efforts, diversifying the biomedical research workforce remains an elusive goal, and large sectors of the US population remain underrepresented."[1] Such an honest appraisal of the lack of significant improvement has reenergized NIH, and their efforts include a focus on strategies that "create institutional cultures where individuals with diverse perspectives can thrive and create new knowledge to improve human health."[7] New NIH initiatives are operationalizing this strategic focus.

The US medical field is also confronted with an underrepresentation of racial and ethnic minority groups[8] in general as well as women in leadership positions.[9] For example, research has documented that, in nearly all specialties, Black and Hispanic members of the academic physician workforce were more underrepresented in 2016 compared to 1990.[10] In 2015, the Association of American Medical Colleges issued a report entitled "Altering the Course: Black Males in Medicine," which found that there had been no increase in Black or African American men in the medical profession for nearly 40 years. Similarly, a 2014 NIH Physician-Scientist Workforce Report documented the lack of diversity within the physician-scientist workforce, describing this deficit as "a source of very serious concern to the NIH and to the professions." A diverse clinical workforce is essential for decreasing health inequities and improving the quality of care for all members of society.

Bringing change to the genomics workforce

The field of genomics is affected by the same problematic lack of diversity that plagues and hampers the US research and clinical workforces. Since 2001, NHGRI has prioritized funding

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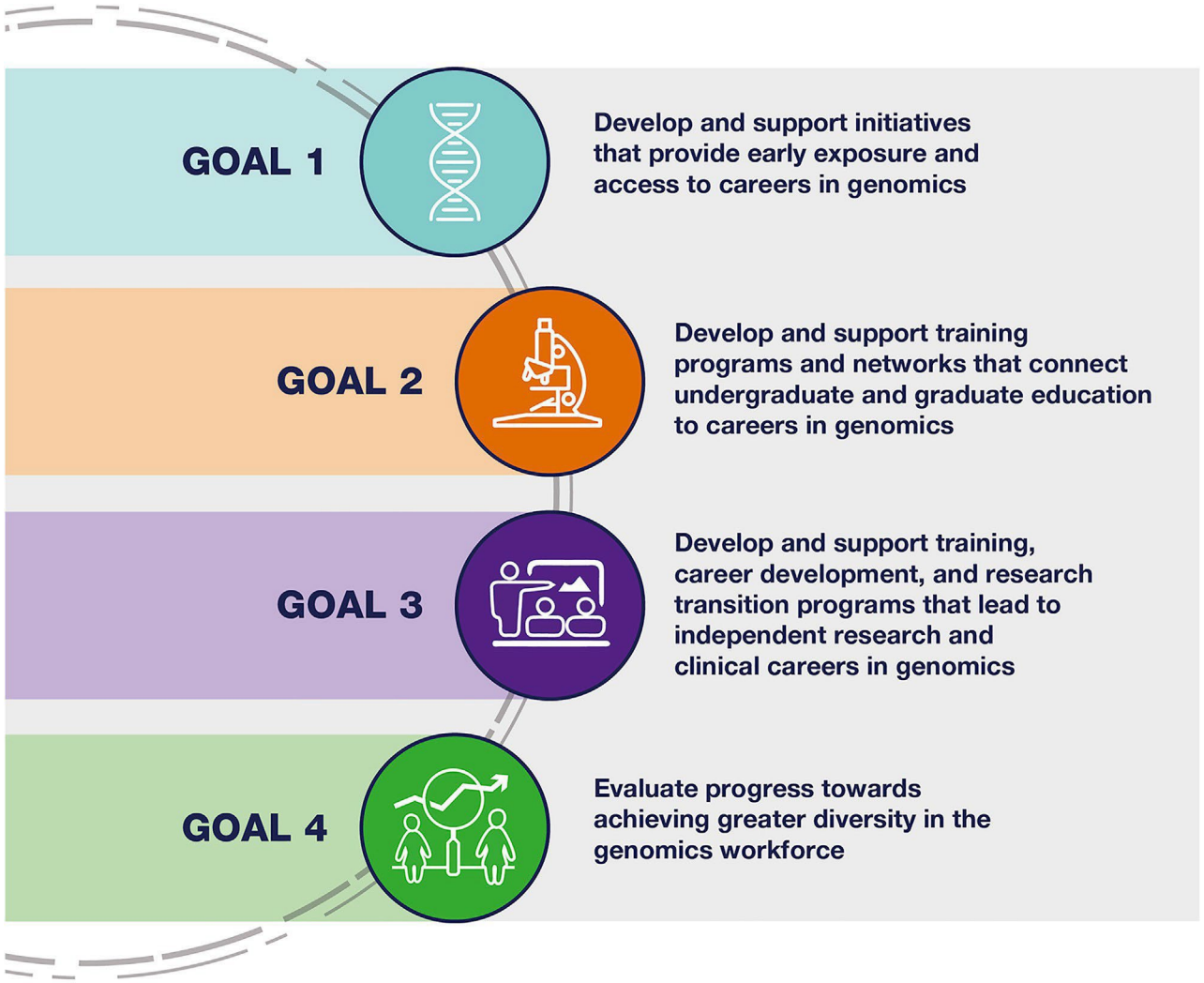

Figure 1. The Four Goals of NHGRI's Action Agenda for Building a Diverse Genomics Workforce

programs that increase the number of genomics-trained researchers from diverse groups.[11] Despite these efforts, the genomics research workforce has seen limited growth in the number of independent researchers from underrepresented backgrounds. From a clinical standpoint, in 2020, the US Government Accountability Office (GAO) reported a growing need for more trained medical geneticists and genetic counselors to support the growth of genetics services and genomic medicine in the US, with a dire lack of a clinical genomics workforce in certain regions of the country.[12] While the GAO report did not analyze the demographic composition of the current clinical genomics workforce, surveys and interviews conducted by genetics and genomics professional societies confirm that a profound lack of diversity exists in both the genetic counseling and medical genetics professions.[13] The successful implementation of genomics into clinical care requires the growth of a diverse clinical genomics workforce.

On behalf of NIH, NHGRI embraces the general call to action about workforce diversity, as recently articulated in the institute's new strategic vision.[14] Recognizing its leadership role at The Forefront of Genomics, NHGRI accepts the responsibility to provide stewardship of the field in numerous areas, including articulating and enhancing the fundamental principles and values that undergird the entire research and clinical ecosystem in which genomics now plays a prominent role. Among these principles and values, one in need of urgent attention is championing a more diverse genomics workforce.

Aiming to be bold in tackling a problem that connects with many broader societal issues related to inequities, health disparities, and injustices, NHGRI seeks to bring change to the composition of the workforce that conducts genomics research and brings genomics into healthcare. Toward that end, the institute recently released "Building a Diverse Genomics Workforce: An NHGRI Action Agenda," which provides a ten-year roadmap to increase the diversity of the US genomics workforce.

Box 1. "Building a Diverse Genomics Workforce: An NHGRI Action Agenda" Major Goals and Underlying Actions

1. Develop and support initiatives that provide early exposure and access to careers in genomics
   - 1.1 : Identify best practices in programming designed to provide early exposure to genomics, including barriers and recommendations to eliminate those barriers
   - 1.2 : Support and participate in programs that are designed to encourage individuals of diverse backgrounds to pursue genomics careers, especially persons from groups who are historically underrepresented in science
2. Develop and support training programs and networks that connect undergraduate and graduate education to careers in genomics
   - 2.1 : Create a systematic network of support for underrepresented students as they move to and through graduate training programs in genomics
   - 2.2 : Ensure that undergraduate minority-serving institutions and community colleges are aware of and tightly connected to this network
   - 2.3 : Encourage inclusive climates at all leading graduate-level genomics training programs so as to mentor and promote cohorts of individuals from underrepresented backgrounds
3. Develop and support training, career development, and research transition programs that lead to independent research and clinical careers in genomics
   - 3.1 : Identify and reduce barriers for individuals from diverse backgrounds who want to enter research and clinical careers in genomics
   - 3.2 : Facilitate the inclusion and retention of individuals from diverse backgrounds in research and clinical careers in genomics
4. Evaluate progress toward achieving greater diversity in the genomics workforce
   - 4.1 : Establish a relevant set of metrics for success for NHGRI diversity training and career development programs
   - 4.2: Use these metrics to develop tracking protocols for all individuals supported by these training programs
   - 4.3: Assess all NHGRI training and career development programs, including diversity-targeted programs, with periodic reports to leadership

A foundation to build upon

The new NHGRI action agenda builds on a set of institute-supported training programs that have been operating for nearly two decades. In 2002, the institute established the Diversity Action Plan (DAP) program to increase the pool of scientists from underrepresented groups in biomedical research that might pursue genomics-related careers. The DAP program was originally associated with NHGRI's large extramural research programs (for example, the Centers of Excellence in Genomic Science, Genome Sequencing Centers, and Model Organism Databases). Since its inception, the program has supported the participation of more than 1,400 trainees in research education projects. In 2014, NHGRI expanded the DAP program and encouraged applicants to propose innovative initiatives and educational activities that would enhance the diversity of genomics trainees. Currently, NHGRI funds 14 DAP awards at 13 different academic institutions across the US.

Meanwhile, at the NIH-wide level, NHGRI participates in several programs that endeavor to promote diversity in the biomedical research workforce, including the Maximizing Opportunities for Scientific and Academic Independent Careers Transition Award to Promote Diversity (MOSAIC) program, the Research Supplements to Promote Diversity in Health-Related Research program, the Ruth L. Kirschstein National Research Service Award Individual Predoctoral Fellowship to Promote Diversity in Health-Related Research, and an opportunity for Small Grants for New Investigators to Promote Diversity in Health-Related Research. All diversity-enhancing funding opportunities can be found on NHGRI's Funding to Promote Diversity in the Genomics Workforce website.

Additionally, in 2019, NHGRI partnered with the American Society of Human Genetics (ASHG) to establish the Human Genetics Scholars Initiative, an effort that aims to develop a community of 250 early-career genomics researchers from underrepresented backgrounds. The program includes an intensive mentoring and skill-building experience for 40 early career scientists from underrepresented backgrounds who are selected to be Human Genetics Scholars. This partnership can serve as a model for developing new innovative and impactful programs through similar collaborations among professional societies, government agencies, industry, and philanthropy.

Ongoing NHGRI efforts to engage students and enhance genomic literacy more broadly are also relevant to fostering a more diverse genomics workforce. On multiple fronts, NHGRI has provided leadership at NIH and beyond in the development of informal and formal science education programs focused on genomics. This has included various programs that

engage the public in understanding genomics and the ethical, legal, and social issues that emanate from genomic advances. Notable examples are:

- A partnership with the Smithsonian's National Museum of Natural History to develop and travel the award-winning exhibition "Genome: Unlocking Life's Code."
- Virtual and in-person genomics programs organized by the institute's Education and Community Involvement Branch that train middle school, high school, and college teachers associated with diverse geographic locations and student populations.
- An education- and engagement-focused collaboration with PBS and WETA public television in support of "Ken Burns Presents The Gene: An Intimate History," a documentary based on the book authored by Siddhartha Mukherjee.

The aim of these NHGRI education and community engagement programs is to expose people of all ages—especially students from underrepresented groups in science—to the field of genomics. With such a commitment, the institute seeks to foster a broader and more diverse cadre of students who are interested in careers that involve genomics.

Like all fields of science and medicine, genomics must reckon with the complex social issues associated with systemic injustices and biases in order to establish a more diverse genomics workforce. To proceed appropriately, NHGRI must gain a more complete view of the current demographic composition of genomics professionals and trainees. Toward that end, the institute has partnered with the ASHG, the American College of Medical Genetics and Genomics (ACMG), and the National Society of Genetic Counselors (NSGC) to conduct a landscape analysis that will provide detailed information about the current makeup of the genomics workforce. These data will enable NHGRI to build upon and expand successful efforts as well as direct additional funding and attention to areas in need.

An ambitious roadmap

The new "Building a Diverse Genomics Workforce: An NHGRI Action Agenda" signifies the institute's strong commitment to provide leadership and resources for enhancing the diversity of the genomics workforce by 2030. Developed by an internal working group over the last year, the action agenda articulates goals and objectives for increasing the number of underrepresented individuals who work in genomics. The action agenda also challenges the broader research and healthcare communities to establish synergistic and durable structures that nurture and support the professional pursuits of those individuals. Important first steps toward achieving the articulated goals include an in-depth evaluation of the institute's current training and diversity programs and a search to develop new partnerships with other motivated groups (such as NHGRI's established partnership with ASHG).

The major thrust of the action agenda is the establishment of new—and an enhancement of existing—programs and strategies that focus on increasing the number of individuals from underrepresented backgrounds who have the necessary training and experiences to pursue careers in genomics. Some of these efforts target the trainee community, specifically by fortifying and expanding the institute's existing training programs to maximize the diversity of individuals who come into genomics at an early stage. Other efforts will help underrepresented, earlystage scientists establish independent genomics research careers and attain leadership positions in the field. In all cases, the institute will work with academic institutions, healthcare organizations, professional societies, and industry to create environments that nurture this more-inclusive vision of a diverse genomics workforce.

NHGRI's roadmap for the coming decade is organized around four goals (Figure 1), each with a set of associated actions (Box 1). Specifically, the institute will:

- Support programs that provide opportunities for individuals from underrepresented backgrounds to pursue genomic careers.
- Advance training programs that connect undergraduate and graduate education to careers in genomics.
- Reduce barriers for individuals from diverse backgrounds to enter research and clinical careers in genomics.
- Continuously evaluate these efforts to determine their success in achieving enhanced diversity in the genomics workforce.

Closing thoughts

The promise of genomics cannot be fully realized without successfully attracting, developing, and retaining a diverse research and clinical workforce that more closely resembles the population of the US. Aligned with the institute's 2020 strategic vision,[14] the new "Building a Diverse Genomics Workforce: An NHGRI Action Agenda" boldly commits to this important goal. Like the monumental challenge of sequencing the human genome for the first time during the Human Genome Project, the institute stands ready to tackle this herculean task. While NHGRI is committed to providing leadership and resources, ultimate success will also depend on the collective efforts of the genomics community and its members. As institute leaders, we invite you to join us in making diversity of the workforce a priority for the entire genomics enterprise.

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Web resources

Education and Community Involvement Branch, https://www.genome.gov/about-nhgri/Division-of-Genomics-and-Society/Education-and-Community-Involvement-Branch

Funding to Promote Diversity in the Genomic Workforce, https://www.genome.gov/careers-training/Funding-to-Promote-Diversity-in-the-Genomic-Workforce

Genome Unlocking Life's Code, https://unlockinglifescode.org/

Human Genetics Scholars Initiative, https://www.ashg.org/membership/awards/hgsi/

Initiative to Maximize Research Education in Genomics: Diversity Action Plan (R25), https://grants.nih.gov/grants/guide/pa-files/PAR-19-380.html

NHGRI Action Agenda to Build a Diverse Workforce to Accelerate Genomics, https://genome.gov/workforcediversity

NIH Faculty Institutional Recruitment for Sustainable Transformation (FIRST) Program: FIRST Cohort (U54 Clinical Trial Optional), https://grants.nih.gov/grants/guide/rfa-files/RFA-RM-20-022.html

NIH Physician-Scientist Workforce Report (2014), https://report.nih.gov/workforce/psw/index.aspx

Notice of NIH's Interest in Diversity, https://grants.nih.gov/grants/guide/notice-files/NOT-OD-20-031.html

PBS Learning Media: The Gene, https://www.pbslearningmedia.org/resource/the-gene-full-film/the-gene-intimate-history/

Professional Status Survey 2020: Executive Summary. National Society of Genetics Counselors, https://www.nsgc.org/p/cm/ld/fid¼468

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