

Summary of Future Directions in Genomics and Health Equity Virtual Workshop

April 6-7, 2022

Introduction

As part of the National Human Genome Research Institute's efforts in to increase health equity in genomics, the Office of Training, Diversity, and Health Equity (TiDHE) hosted the virtual "Future Directions in Genomics and Health Equity Workshop" on **April 6 - 7, 2022**. The goal of the workshop was to identify research gaps and opportunities that will help to decrease health disparities and improve health equity in genomics. The workshop had the following objectives:

- 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.

A series of presentations, panels and breakout groups executed these objectives by offering different methods for participants to share their research, expertise, and diverse perspectives. Across both days, over 300 participants attended. In addition to this meeting summary, a video recording of the meeting is available at: <https://www.genome.gov/event-calendar/future-directions-in-genomics-and-health-equity-research>

NHGRI and NIH's Mission for Health Equity and Diversity

Speakers: Eric Green, M.D., Ph.D., Vence Bonham, J.D., Leia Butler, J.D., Karriem Watson, D.H.Sc., M.S., M.P.H.

A series of presentations related to the NHGRI or NIH mission for health equity spanned over both days of the workshop. NHGRI Director, Eric Green, and NHGRI Acting Deputy Director, Vence Bonham, gave remarks about the importance of genomics and health equity research to NHGRI. On Day 1, Dr. Green stated the NHGRI aims to make genomic advances accessible to all communities. When developing the [2020 NHGRI Strategic vision](#), research and issues around diversity and health equity in genomics were a significant part of the conversation. Due to this, the NHGRI's Strategic Vision for the next ten years is to strive for global diversity in all aspects in genomics research and the systemic inclusion of ancestrally diverse and underrepresented groups in genomic studies. The NHGRI also wants to maximize the usability of genomics for all members of the public, which includes the ability to access genomics in healthcare. On Day 2, the NHGRI Acting Deputy Director, Vence Bonham, continued the conversation about the NHGRI's directive for genomics and health equity. Mr. Bonham noted the principle of ensuring the usability of genomics to all should help guide recommendations in developing a research agenda to increase equity and reduce existing inequities in genomics. This research agenda should be advanced across the existing NHGRI research areas, including genomic technology development, genome structure and function, computational genomics, genomic variation and disease, genomic medicine, ELSI and training in genomics. Mr. Bonham also introduced areas where recommendations would be sought in the breakout groups that would be convening later that day: social determinants of health; structural factors; implementation science; data science, and ELSI.

Also on Day 2, the [NIH UNITE](#) initiative Program Manager, Leia Butler, and the [All of Us](#) Research Program Chief Engagement Officer, Karriem Watson, gave presentations that highlighted the NIH's research and mission for improving health equity and diversity. Ms. Butler highlighted the UNITE

Program, which has a commitment to identify and address structural racism within the NIH-supported and the greater scientific community. UNITE has 5 committees: 1) Understanding stakeholders; 2) Facilitating new research on health disparities/minority health/health equity; 3) Improving the NIH culture and structure for equity, inclusion, and excellence; 4) Transparency, communication, and accountability with internal and external stakeholders; and 5) Addressing the extramural research ecosystem by changing policies, cultures, and structures to promote workforce diversity. The program also established an NIH Anti-Racism Steering committee, to work on developing racial and ethnic equity plans (REEP) for all the institutions. Dr. Watson discussed the NIH *All of Us* Research program; a program that aims to build a diverse data set of one million people to help improve healthcare for all populations. The program plans to have diversity in age, disability, geography, disease types, etc., with 80% of the participants from underrepresented in biomedical research. The program also plans to increase diversity in the biomedical research workforce and engage in outreach with communities.

Setting the Stage: A Vision for Health Equity in Genomics

Speaker: Nancy Cox, Ph.D., Vanderbilt University

On Day 1, Dr. Nancy Cox gave a presentation titled "Genomics in Health Equity: Today and Tomorrow." Dr. Cox discussed recent research that aims to understand how rare variant risk, polygenic risk, and the environment interact to result in association with complex traits. The research has shown that for genetic ancestry that is accounted for by reported race, there are different genetic components as well as associations for disease. Therefore, there is a need to use genetics and genomics to clearly identify portions of health disparities that are driven by social determinants of health as well as understand the parts of health equity that are not driven by genetics.

In terms of clinical laboratory research, there needs to be a focus on health equity to do no harm and correct any wrongs quickly. This can be an issue when the reference genome ranges used for lab testing fail to adequately account for the diversity of the population. The lack of precision in these genomic reference ranges have clinical consequences, such as requiring repeated testing, over and under-diagnosis of disease, unnecessary procedures, and failure to obtain needed care and therapies. While this is a complex problem without a simple solution, Dr. Cox detailed an approach to develop PRS-adjusted laboratory values that account for differences across genetic ancestry groups that may help reduce structural health disparities, developed through collaborations within the EndoPhenotype Incorporated Polygenic Risk Scores (EPIC-PRS) group that is part of the PRIMED Consortium.

Panel: Moving Forward: From Health Disparities to Health Equity in Genomics

Moderator: Eliseo J. Pérez-Stable, M.D., Director, NIMHD. Panelists: Esteban Burchard, M.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

The panel responded to the research results and ideas presented by Dr. Cox and discussed what it means to move forward from health disparities to health equity in genomics. The panelists noted race and ethnicity is a self-identified social construct and there is not a scientific test to determine race. In addition, race/ethnicity is a proxy for environmental, structural, and legal components and the use of race is proving to be more complicated as the world becomes more diverse. Due to this, health systems and research studies need to be more systematic in their recording and reporting of race. It is important to remember that socio-economic factors also play a large effect on someone's health, the care they receive, and how physicians communicate with their patients and community. There is also not enough empirical research to determine if race should be included in clinical equations. This is crucial to the

research, as the system needs to understand systematic issues that allow health disparities to continue and its contributions to health equity concerns.

The panel also noted when interacting with Indigenous communities, the recognition of sovereignty is a leading value. As a social and legal construct that gives the ability of tribal nations to make decisions for themselves, which many communities require researchers and others to uphold. Views about data ownership and how researchers interact with tribal nations are all concepts that are built-in and entangled with race.

In addition to racial disparities, the panel discussed the need to address structural, attitudinal, and other barriers that preclude equitable participation of people with disabilities in genomic and precision medicine research, which undermines efforts to shift from health disparities to health equity. It was noted that although people with disabilities are often recruited to genetic studies that aim to identify the underlying cause of the disability, such individuals are often excluded (directly and indirectly) from mainstream genetic and precision medicine research. There is a need to take an intersectional approach to promote diverse and equitable inclusion in precision medicine research.

Lastly, the panel noted that a key step to addressing the issue of health inequity is to become comfortable with uncomfortable conversations that include diverse views is important to achieving health equity; therefore, diverse populations should be recruited into research in a responsible, disability accessible, and safe way and the value of diverse and interdisciplinary teams should be implemented in all areas of research. The effect of discrimination, racism, and ableism is still very present in our scientific system and is something that needs to be addressed. By understanding and fighting structural racism and ableism, the community can create a safer and more inclusive environment.

Current Research in Genomics and Health Equity

Speaker: John Carpten, Ph.D., University of Southern California

Dr. John Carpten gave a presentation on current research in genomics and health equity. Dr. Carpten presented that health equity includes social-structural and socioeconomic factors. He asked, "How can there be health equity if there is no equity?" As such, the community needs to look at the long-lasting detriments of structural racism and financial impacts that go beyond race. As an example, there is limited diversity in The Cancer Genome Atlas ([TCGA](#)) and analysis of many populations are significantly underpowered. The genomic scientific community needs to increase international collaborations and increase genomic analyses across a variety of populations, including oversampling as needed. Dr. Carpten also emphasized the need to diversify the genomic workforce more broadly to ensure precision medicine is accessible to all and thus led to more equitable health outcomes. To move forward, diverse communities must be better engaged. Funding mechanisms that encourage and incentivize investigator diversity and multidisciplinary teams were recommended.

Panel: Identifying Research Gaps and Opportunities

Moderator: Chanita Hughes-Halbert, Ph.D., University of Southern California. Panelists: Rick Kittles, Ph.D., City of Hope, Loren Saulsberry, Ph.D., University of Chicago, Michael Inouye, Ph.D., University of Cambridge

The panel discussed key points presented by Dr. Carpten and expanded on the gaps and opportunities in genomic and health equity research. The panel noted that disparities and health, overall, in the United

States, are influenced significantly by poverty and racism. It was suggested that there needs to be an increased understanding of the influence of race and racism has on health disparities and inequities. Additionally, the panel discussed the establishment of metrics and standards to achieve accountability. Genomic workforce diversity and the support of institutions that are dedicated to training diverse researchers, such as Minority Serving Institutions, is also important. Having diverse people in the room would allow for more diverse ideas and lead to progress in genomics. There is also a lack of representation of diverse populations in datasets, which creates gaps in research outcomes that apply to all populations and leads to differences in implementation of genomic medicine. In addition, the differences in views and perspectives across different populations are not always incorporated into clinical care. Therefore, institutions and diverse settings need to have the necessary resources to appropriately conduct both research and medicine in an equitable manner. The panel noted that bias in the NIH review process against applications from diverse investigators and minority serving institutions works in opposition to the inclusion of diverse institutions and diverse settings thus funding and policy changes are needed. Lastly, the panel reiterated the call for the scientific community to examine how and where genomics plays a role as it relates to broader systemic issues of poverty and racism. There is an intersectionality that has not been examined with enough depth to inform and direct genomics and health equity research.

Current Challenges in Genomic Research and Genomic medicine That Lead to Health Disparities

Speakers: James Hildreth, Ph.D., M.D., President and Chief Executive Officer, Meharry Medical College; Genevieve Wojcik, Ph.D., Johns Hopkins Bloomberg School of Public Health

Dr. Hildreth presented challenges in genomic research and medicine that lead to health disparities. In his presentation, he highlighted that African American and Hispanic populations have a higher rate of chronic health issues and that Covid-19 has disproportionately impacted these groups. Therefore, there needs to be more inclusive genomic research that will lead to inclusive implementation and improved genomic medicine opportunities to increase health equity. Dr. Hildreth also noted that genetic information is frequently used for diagnosis, prognosis, treatment, but there is a significant gap in genomic databases with diverse populations. Polygenic risk scores (PRS) used in disease prediction are less accurate across diverse populations because a majority of the data come from those of European ancestry. So, it is necessary to increase diversity samples in research and databases to identify and improve health outcomes. Dr. Hildreth also emphasized the need for more diverse data to be represented in clinically actionable genetic information and accurate predictive risk scores, to improve personalized medicine and pharmacogenomics. He also suggested that the genomics community should foster community engagement and partnerships including engaging participants and increasing genomic education and understanding. Several examples of educational and scientific activities underway in collaboration with Historically Black Colleges and Universities (HBCU) were given. Finally, the lack of diverse researchers creates bias in research. Investigators have personal connections to ancestry or countries of origin. Lack of funds for researchers from underrepresented backgrounds is a major barrier.

Dr. Genevieve Wojcik continued the discussion of challenges in genomic research and medicine that lead to health disparities by spotlighting that Eurocentric bias is accepted as the default, which permeates every step in research, including the scientific questions that are asked, who is included in the research, and the systems that researchers choose to model. This default contributes to genomic health inequities. So, when developing genomic health tools for complex systems outside of the perceived default, equity is key. A major barrier to achieving health equity is imprecision when describing study populations and the corresponding lack of accountability or ability to assess equity if

what and whom is being measured is unclear. As noted, PRS for European ancestry studies are vastly overrepresented based on the world population. However, the results from the use of PRS improve when adjusted for ancestry, compared to self-identified race or ethnicity. Dr. Wojcik also noted that diverse researchers can work with communities to jointly determine what is best for them.

Panel: Addressing Structural Factors Needed to Support Health Equity Research in Genomics

Moderator: Carol Horowitz, M.D., Icahn School of Medicine at Mount Sinai. 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

The panel included those with expertise in the areas of LGBTQI+ issues and Indigenous and underserved population ethical research. The panel responded to the two presentations and discussed structural factors needed to support health equity research in genomics. The panel voiced a need to change underlying structure, by diversifying the voices heard in the room. Tribal communities have been notably absent from scientific conversations, due to a lack of resources available to them. The panel reiterated the need for more inclusive genomic research as it will lead to improved genomic medicine opportunities. This includes representation of diverse populations among study leadership, not only among study support staff. The panel discussed the importance of viewing all phases of genomics research with a health equity lens to examine and address the multiple forms of participant vulnerability that is rooted in structural inequities. Without a participant's voice, there is potential for over or underestimating participant risk and personal agency, which can lead to exposure to research harms and limit research benefits. The panel noted the importance of including health equity in all stages of genomic research and an increased emphasis on community engagement, including funding community members to be involved in research for longer periods of time. When engaging with the community, it is important to be honest about how much the research will benefit them, discuss how to proactively include them in the research, and adopt a stance of cultural humility. The panel also emphasized that all investigators have a moral and ethically reasonability to work together to dismantle harmful narratives, frames, practices, and policies across the scientific research community.

Common Themes

After the talks and panel discussion, 6 common themes emerged:

1. Diversifying the genomic workforce and landscape.
2. Addressing the lack of diverse data.
3. Understanding how lack of diversity in populations and communities comprising genomic research cohorts impacts health disparities.
4. Addressing inappropriate use of racial categories in research, laboratory values and clinical markers and underutilization of genomic markers.
5. Nurturing long standing relationships with diverse communities to build trust and conduct research equitably.
6. Developing metrics of health equity, such as access to genomic testing, and applying them across genomic studies

Breakout groups

After the presentations and panel discussions, workshop participants were separated into five breakout groups. These breakout groups were created to touch on all aspects of genomics research and medicine. During the breakouts, participants answered questions pertaining to the topic area and were asked to give recommendations for future research on how to decrease health disparities and improve health equity in genomics. The five groups included:

1. Social Determinants of Health and Genomic Equity (*Leaders: Tabia Henry Akintobi, Ph.D., M.P.H., Morehouse School of Medicine and Nanibaa' Garrison, Ph.D., University of California, Los Angeles*)
2. Structural Factors (*Leaders: Malia Fullerton, D.Phil., University of Washington, and Nita A. Limdi, Pharm.D., Ph.D., MSPH, University of Alabama at Birmingham*)
3. From Bench to Bedside: The Implementation Science of Genomics and Health Equity (*Leaders: Denise Dillard, Ph.D., Southcentral Foundation and 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 and Jeff Leek, Ph.D., M.S., John Hopkins Bloomberg School of Public Health*)
5. Health Equity Research in ELSI (*Leaders: Catherine Hammack-Aviran, M.A., J.D., Vanderbilt University School of Medicine and Benjamin Wilfond, M.D., Seattle Children's Hospital*)

Recommendations

The breakout groups were each asked to choose the top two recommendations within the topic area of their breakout to get a pulse of the community's priorities. The top two recommendations were reported back to the larger group, then consolidated into a poll, and all participants voted on the top priority from the list of ten recommendations (Appendix). Based on the poll results, the recommendations receiving the most votes were to: 1) Diversify the genomics workforce by targeting community colleges and MSIs including, HBCUs, HSIs and Tribal Colleges, to build infrastructure, capacity, faculty, and leadership at these institutions; and 2) Ensure sufficient time and equitable resource distribution and funding for appropriate community engagement.

Next Steps

The workshop concluded with an acknowledgment to workshop participants and those who aided with workshop planning and preparation. In addition to this workshop summary, a scientific article is planned to be submitted to a peer-reviewed journal. Finally, NHGRI will consider these recommendations for development of future funding initiatives.

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APPENDIX.

Top Recommendations from the breakout groups (presented as poll options to the full group)

What recommendation is the most important for NHGRI to consider when developing future research opportunities in genomics and health equity? %

Diversify the genomics workforce by targeting HBCUs, MSIs, Tribal Colleges, Community Colleges, for increasing funding, support programs and training to build infrastructure, faculty, leadership	29%
Ensure sufficient time and equitable resource distribution and funding for appropriate community engagement with review and progress reporting and criteria	23%
Include a robust understanding of contextual variables and emphasize diverse settings; should also include clear and measurable metrics to assess health equity	9%
Collect data on wider abilities, demographics (resiliencies) and barriers to participation (vulnerabilities) as a standard reporting element; use this as part of the education & improvement process	7%
Include training on using SDOH measures and an analytic plan for collecting and using SDOH measures	7%
Require training for researchers, reviewers and funders on diversity, sensitivity and inclusion and a participation role for community members in the research and review and funding decisions	7%
Enhance workforce diversity - involving point of care clinicians, other members of the team in understanding and communicating genomic research using a health equity lens	6%
Increase outreach to diversify the demographic and disciplinary background of ELSI researchers	6%
Engage students earlier in the educational pipeline to attract students to computer/data science and then supporting them throughout the career process	3%
Support long-term training opportunities to study underrepresented populations using SDOH measures	3%
Total	100%

Full List of Recommendations from the breakout groups

Social Determinants of Health and Genomic Equity

1. NHGRI should require investigators to include SDOH on some level (special RFAs to include measures)
 1. Types of data - geospatial approaches, self-report, machine learning, artificial intelligence to extract data from clinical record
 2. Race, ethnicity, age, disability, language, sexual orientation, gender, intersex status
 3. Develop working group to flesh out what SDOH measures would be
2. Investigators would need to apply the SDOH measures to the analysis
3. Need mechanisms to support long-term training opportunities to study underrepresented populations using SDOH
4. Need initiatives to support new data collection

Structural Factors

1. Sufficient time and funding for appropriate community engagement with appropriate review and progress reporting and criterion. Need to be allocated equitably and include the community in the resources.
2. Required training for “researchers” on diversity, sensitivity and inclusion and a participation role for community members both in the research and in NIH review and NHGRI decision making.
3. Collect data on wider abilities, demographics (resiliencies) and barriers to participation (vulnerabilities) as a standard reporting element and use this as part of the education and improvement process

From Bench to Bedside: The Implementation Science of Genomics and Health Equity

1. Suggest efforts directed at increasing the ability of clinicians working in these communities to engage in research. They interact with community patients every day and generally have a clear understanding of community priorities. Including these clinicians in the design and implementation of research would improve the relevance to the community. Include a robust understanding of context (be diverse) - involve the community
2. Workforce diversity - clinicians, other members of the team
3. Building consensus and understanding around metrics to assess potential for incidental inequities (we can potentially draw from existing metrics) and hold systems leaders accountable
4. Support for formative research for the exploration of outcomes and constructs that are meaningful for the community
5. Support for research on interventions to mitigate inequities with tailored engagement

Data Science Genomics Equity,

1. Diversify the genomics workforce by targeting HBCUs, MSIs, Tribal Colleges, Community Colleges, for increasing funding and support programs to build infrastructure, capacity, faculty, and leadership at these institutions (focus on training in genomics science and health equity) acknowledging the unique circumstances and needs of faculty at these institutions.
2. Develop funding/support for community-based research where participants are both contributors and beneficiaries to generate a more diverse collection of data acknowledging these communities needs in terms of data ownership and participation. Focus on privileging participants and their perspective.

3. Engage students earlier in the educational pipeline to attract students to computer/data science exposing them to diverse scientists (high school) and then supporting them throughout the career process through graduate school and into faculty roles.

Health Equity Research in ELSI

1. Any evaluation criteria or framework must consider downstream implications on health and not just representativeness of data.
 - a. Consider downstream impacts of focusing on outcomes—diversity in data is necessary but not sufficient, and may exacerbate disparities/inequities
2. Need to consider new frameworks outside of traditional bioethics that are more inclusive of community and population-based models.
3. More research is needed on the impact of genomics on the full range of individuals with disabilities or differing abilities.
4. Consider the goals of communities when defining health equity. Will not be the same for all groups.
5. Need to increase outreach to graduate and undergraduate students in humanities, social sciences, health systems scientists, health economics and other related fields so that future researchers are aware of career paths related to ELSI research.
6. NHGRI must continue funding and collaborating on analysis of the intersection of law with science, clinical care, and ethics. Need continued and ongoing attention to legal barriers and challenges that impact our ability to achieve health equity.