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Workshop report: Future Directions in Genomics and Health Equity

Judy Cho, MD

Icahn School of Medicine, Mount Sinai

National Advisory Council for Human Genome Research

May 16, 2022

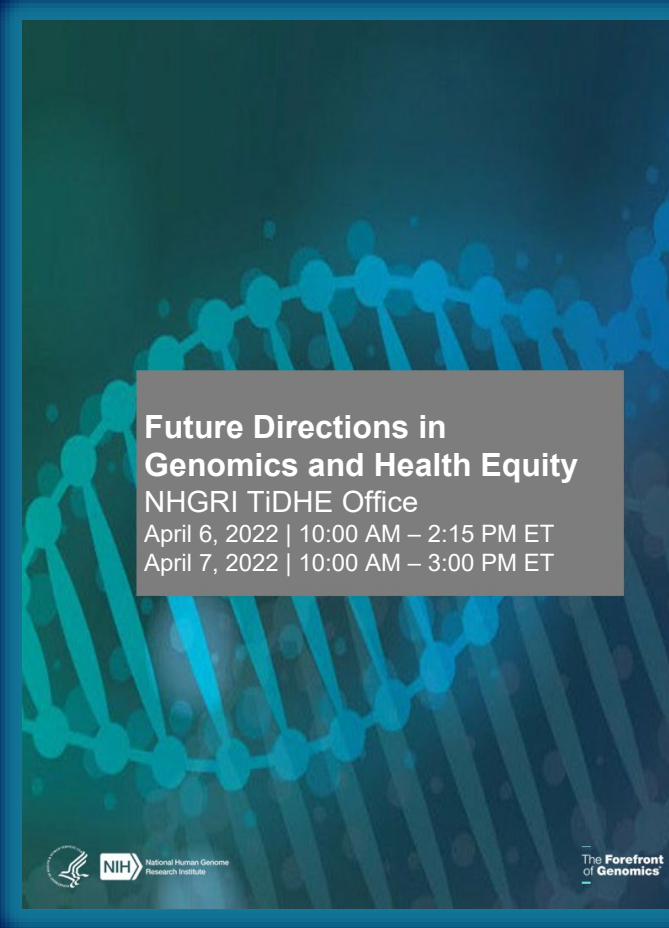


National Human Genome
Research Institute

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Future Directions in Genomics and Health Equity Research Virtual Workshop

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- **Workshop Goal: Identify research gaps and opportunities that will help address health disparities and improve health equity in genomics**
- **Over 300 participants**
- **Co-chairs: Judy Cho, Icahn School of Medicine at Mount Sinai; Sandra Soo-Jin Lee, Columbia University**

Keynote speakers

Setting the Stage: A Vision for Health Equity in Genomics

- Nancy Cox, Vanderbilt University



Current Research in Genomics and Health Equity

- John Carpten, University of Southern California



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

- James Hildreth, Meharry Medical College
- Genevieve Wojcik, John Hopkins University



Moving Forward: From Health Disparities to Health Equity in Genomics



Panelists:

- Esteban Burchard, University of California, San Francisco
- Vanessa Hiratsuka, University of Alaska Anchorage
- Latrice Landry, Harvard University
- Maya Sabatello, Columbia University

Moderator:

- Eliseo Pérez-Stable, National Institute on Minority Health and Health Disparities

Identifying Research Gaps and Opportunities



Panelists:

- Rick Kittles, City of Hope
- Loren Saulsberry, University of Chicago
- Michael Inouye, University of Cambridge

Moderator:

- Chanita Hughes-Halbert, University of Southern California

Addressing Structural Factors Needed to Support Health Equity Research in Genomics



Panelists:

- Kellan Baker, Whitman-Walker Institute
- Rene Begay, University of Colorado Anschutz Medical Campus
- Faith Fletcher, Baylor College of Medicine
- Neil Risch, University of California, San Francisco

Moderator:

- Carol Horowitz, Mount Sinai School of Medicine

Breakout Groups (open to all registrants)

Social Determinants of Health and Genomic Equity

- Tabia Henry Akintobi, Morehouse School of Medicine
- Nanibaa' Garrison, University of California, Los Angeles

Structural Factors

- Nita A.Limdi, University of Alabama at Birmingham
- Stephanie Malia Fullerton, University of Washington

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

- Denise Dillard, Southcentral Foundation, Alaska
- Elizabeth Ofili, Morehouse School of Medicine

Data Science Genomics Equity

- Valentina Di Francesco, National Human Genome Research Institute
- Jeff Leek, John Hopkins School of Public Health

Health Equity Research in ELSI

- Catherine Hammack-Aviran, Vanderbilt University School of Medicine
- Benjamin Wilfond, Seattle Children's Hospital

Selected discussion points

Defaulting to laboratory reference ranges based on European ancestral populations results in misdiagnoses, under- and over-treatment

Address lack of data re: using race or ethnicity in clinical algorithms

Necessity to oversample recent African ancestral populations

Lack of diversity in researchers, peer reviewers has far-reaching impact on what is studied

Evaluating downstream impacts in health equity should go beyond representation in data to include health implications

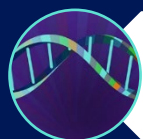
Strengthen bi-directional relationships between researchers and community members

Opportunities for NHGRI

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Diversify the genomic workforce and landscape



Address the lack of diverse genomic data



Understand how lack of diversity in populations and communities comprising genomic research cohorts impacts health disparities



Address inappropriate use of racial categories in research, laboratory values and clinical markers and underutilization of genomic markers

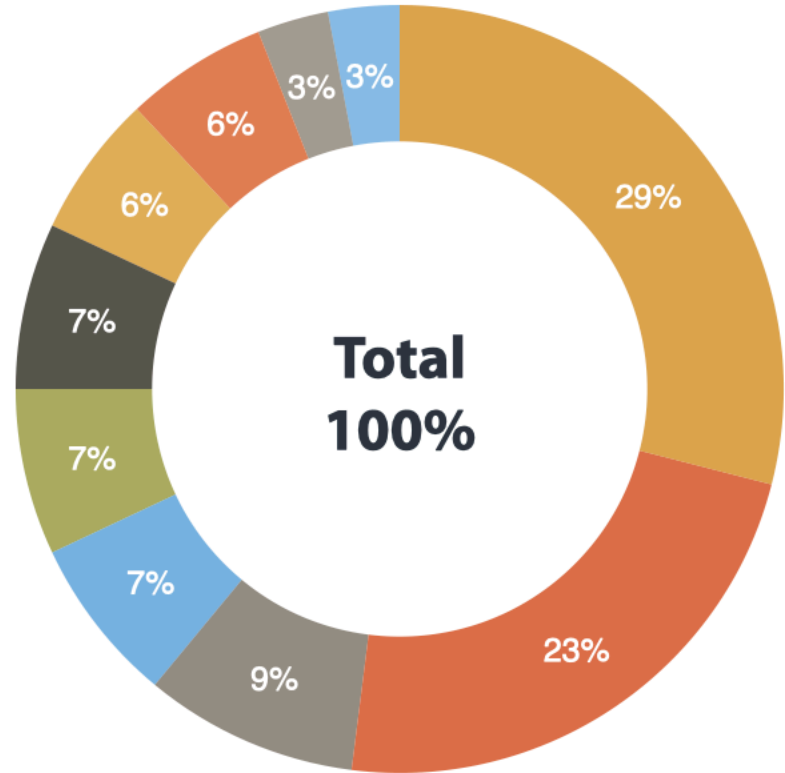


Nurture long standing relationships with diverse communities to build trust and conduct research equitably



Develop metrics of health equity, such as measures of access to genomic testing, and apply them across genomic studies

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



29 %
 Diversify the genomics workforce by targeting HBCUs, MSIs, Tribal Colleges, and Community Colleges

23%
 Ensure sufficient time, equitable resource distribution, and funding for appropriate community engagement

9%
 Include a robust understanding of contextual variables and diverse settings; include health equity metrics

7%
 Routinely collect data on abilities, demographics and barriers to participation, and use for process improvement

7%
 Include training on and an analytic plan for collecting and using SDOH measures

7%
 Require training on diversity, sensitivity and inclusion for funders; participatory role in decision-making for community members

6%
 Enhance workforce diversity of clinicians and clinical teams to communicate genomic research with a health equity lens

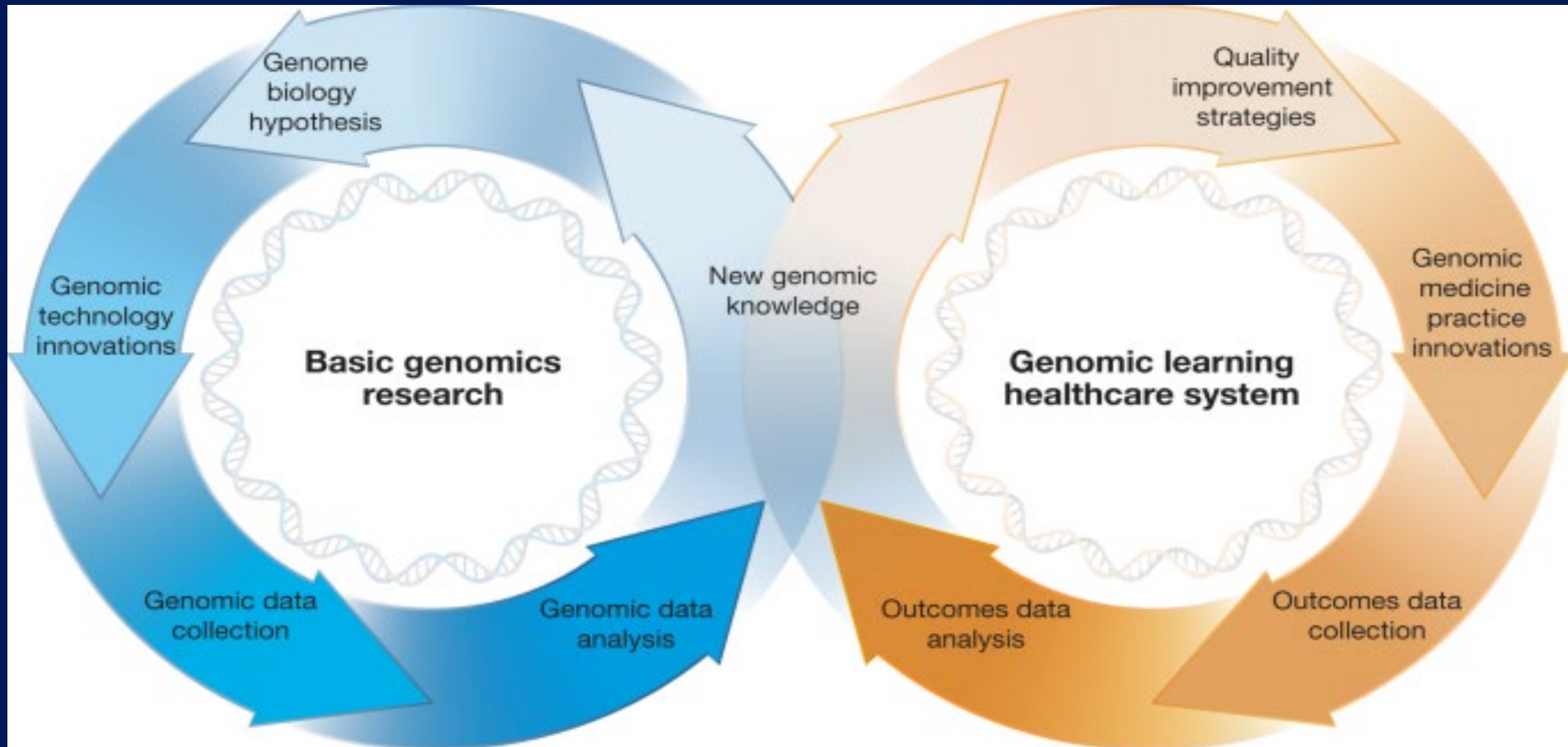
6%
 Diversify the demographic and disciplinary background of ELSI researchers

3%
 Engage students earlier in the educational pipeline to interest and support students in data science

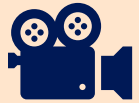
3%
 Support long-term training opportunities to study underrepresented populations using SDOH measures



Health equity is broadly relevant across ALL aspects of genomics research



Next steps



Video and workshop report published at <https://www.genome.gov/event-calendar/future-directions-in-genomics-and-health-equity-research>.



Preparation of manuscript for submission to peer-reviewed publication

Acknowledgments

Workshop co-chairs, speakers, moderators, breakout group leaders, panelists

- NHGRI planning committee
 - Vence Bonham
 - Lucia Hindorff
 - Amber Jackson
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 - Ebony Madden
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- NHGRI communications and informatics
 - Mukul Nerukar
 - Jerryl Somani
- NHGRI Program Analysts
 - Madison Goldrich
 - Kathleen Renna
 - Ella Samer
- Judy Cho, Co-Chair
- Sandra Soo-Jin Lee, Co-Chair
- Kellan Baker
- Rene Begay
- Larry Brody
- Estaban Burchard
- Leia Butler
- John Carpten
- Nancy Cox
- Denise Dillard
- Valentina Di Francesco
- Faith Fletcher
- Malia Fullerton
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- Jeff Leek
- Nita Limdi
- Neil Risch
- Elizabeth Ofili
- Eliseo Pérez-Stable
- Maya Sabatello
- Loren Saulsberry
- Karriem Watson
- Ben Wilfond
- Genevieve Wojick



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