NATIONAL ACADEMIES Sciences Engineering Medicine

Board on Health Sciences Policy

Roundtable on Genomics and Precision Health

Next-Generation Screening - The Promise and Perils of DNA Sequencing of Newborns at Birth: A Workshop

Genomics Roundtable Hybrid Workshop
June 7, 2023

In-person Location
Washington, DC

Remote Login Information
https://events.nationalacademies.org/06-07-2023_next-generation-screening-the-promise-and-perils-of-dna-sequencing-of-newborns

NATIONAL ACADEMIES *Sciences Engineering Medicine*

Board on Health Sciences Policy

Roundtable on Genomics and Precision Health

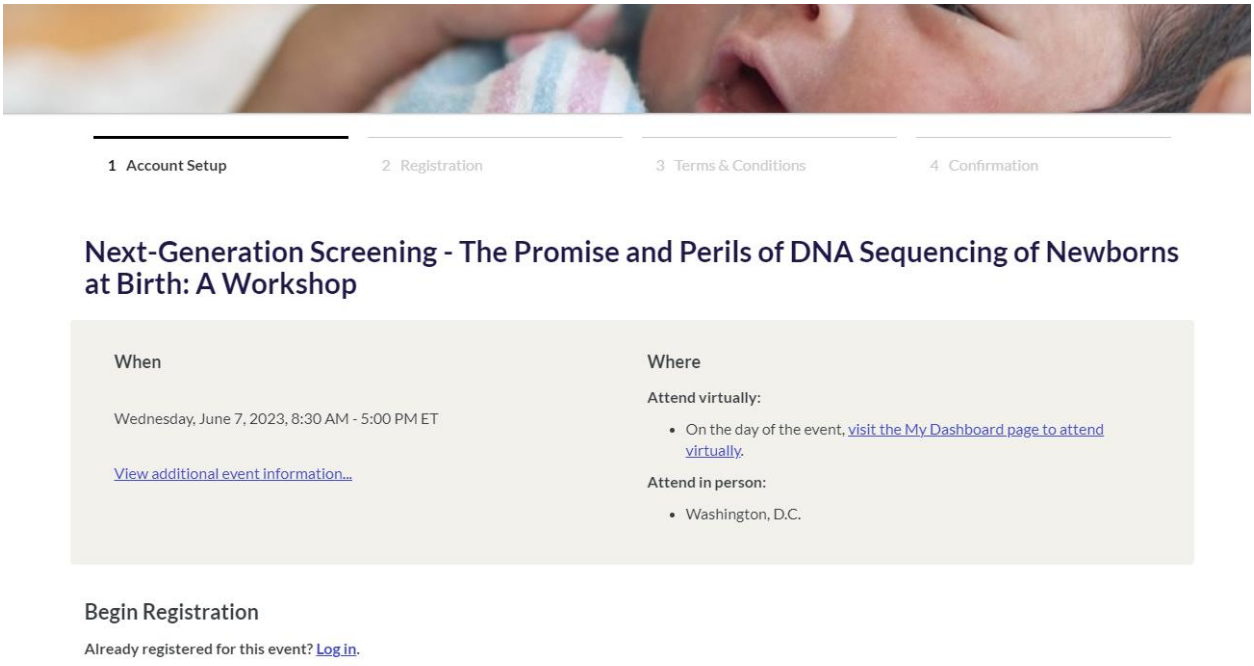
Next-Generation Screening - The Promise and Perils of DNA Sequencing of Newborns at Birth: A Workshop

June 7, 2023

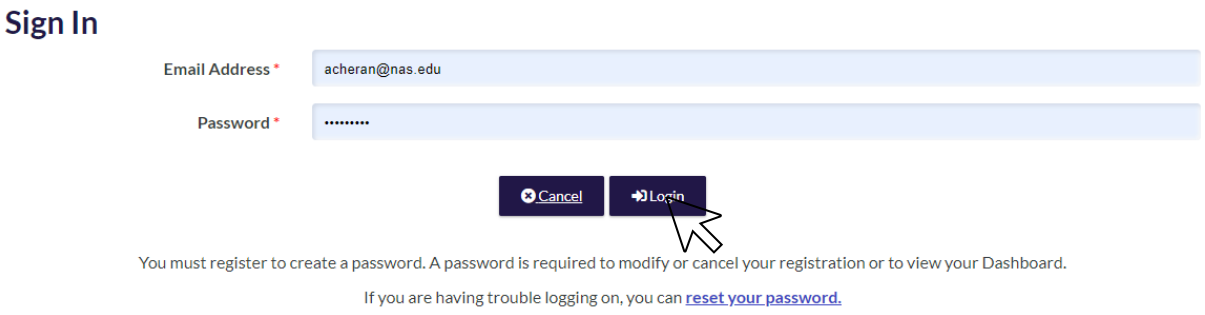
Table of Contents

How To Join Virtually

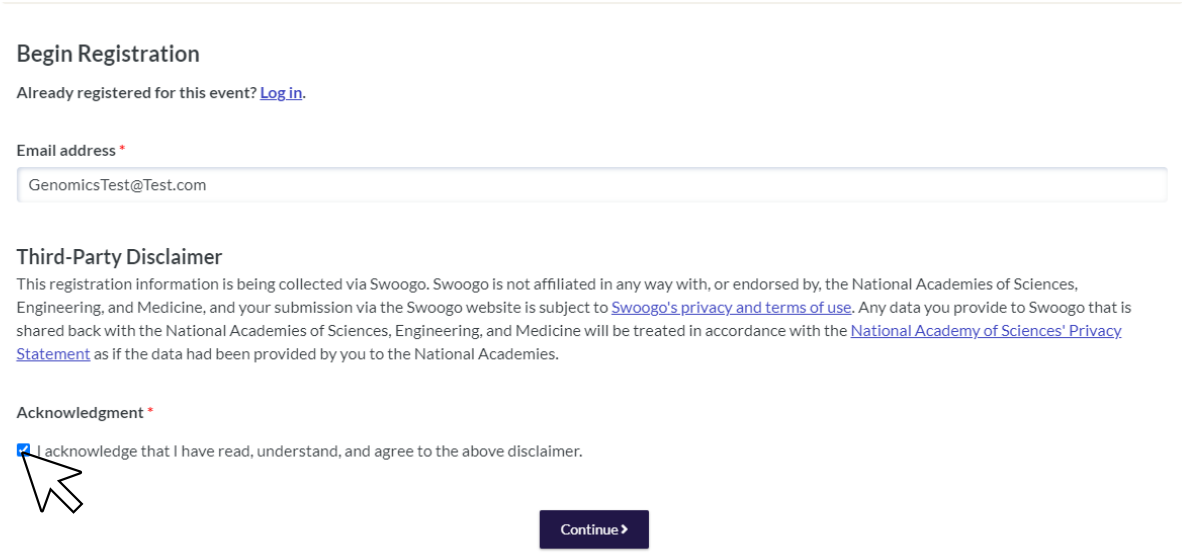
1. Proceed to the Registration webpage.



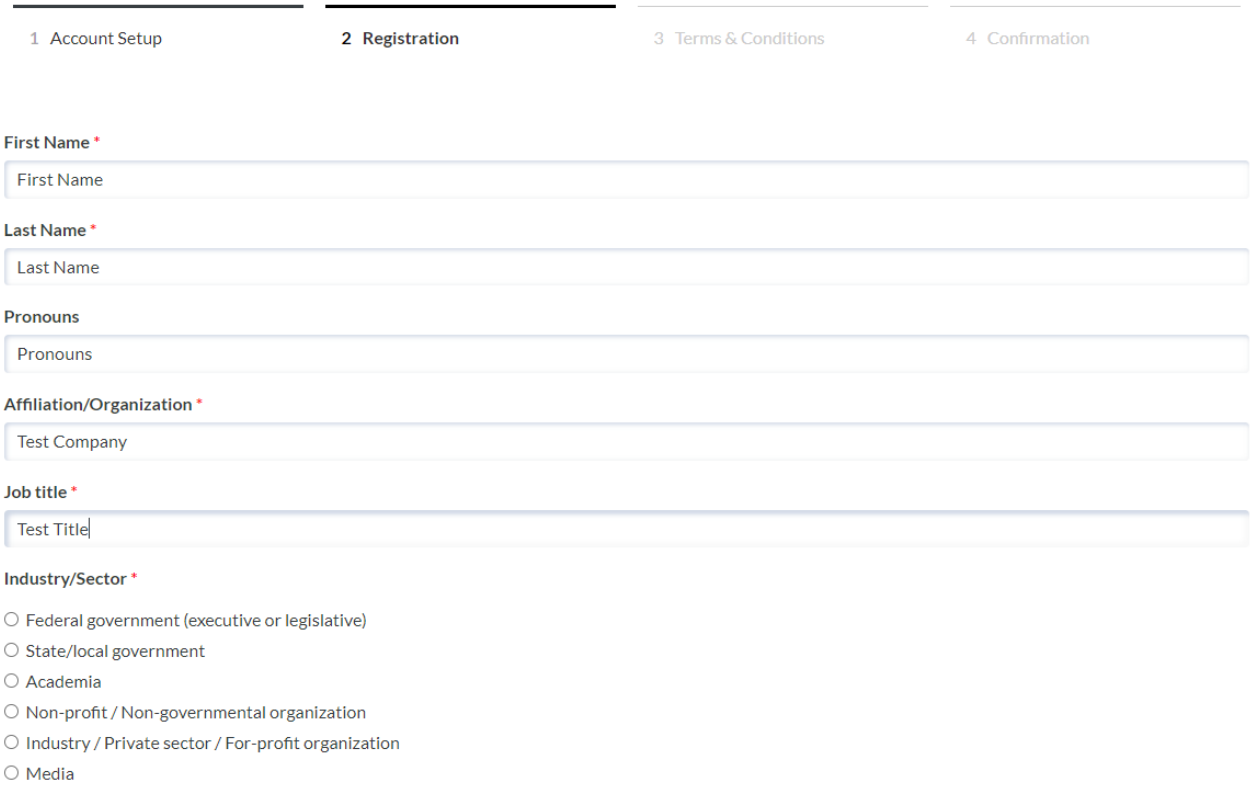
2. If you already have registered for this event, please click “Login”. Input your username and password, and proceed to Step 7.



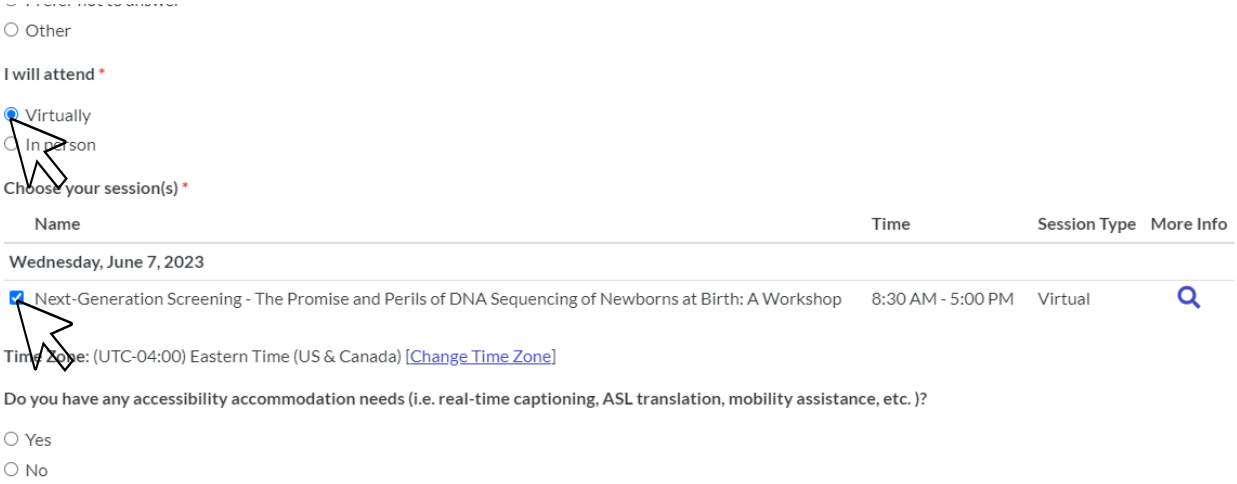
3. If you have not yet created an account on Swoogo, please input your email address, then read and acknowledge the Third-Party Disclaimer.



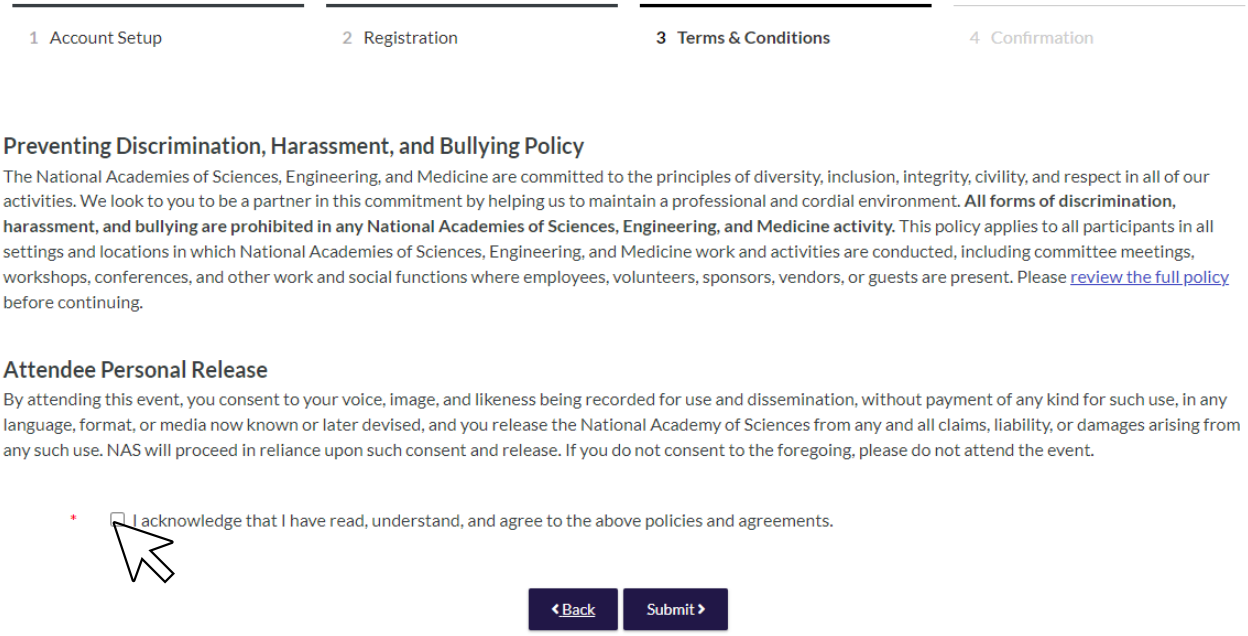
4. Continue to register by filling out the form.



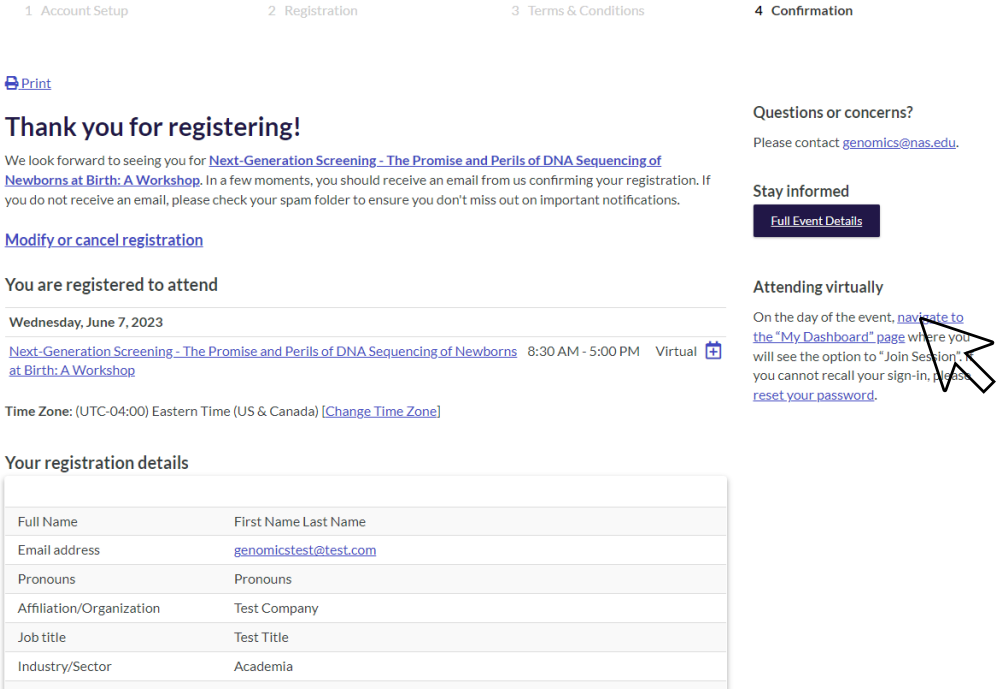
5. For attendance, mark “Virtual” and select the workshop on June 7.



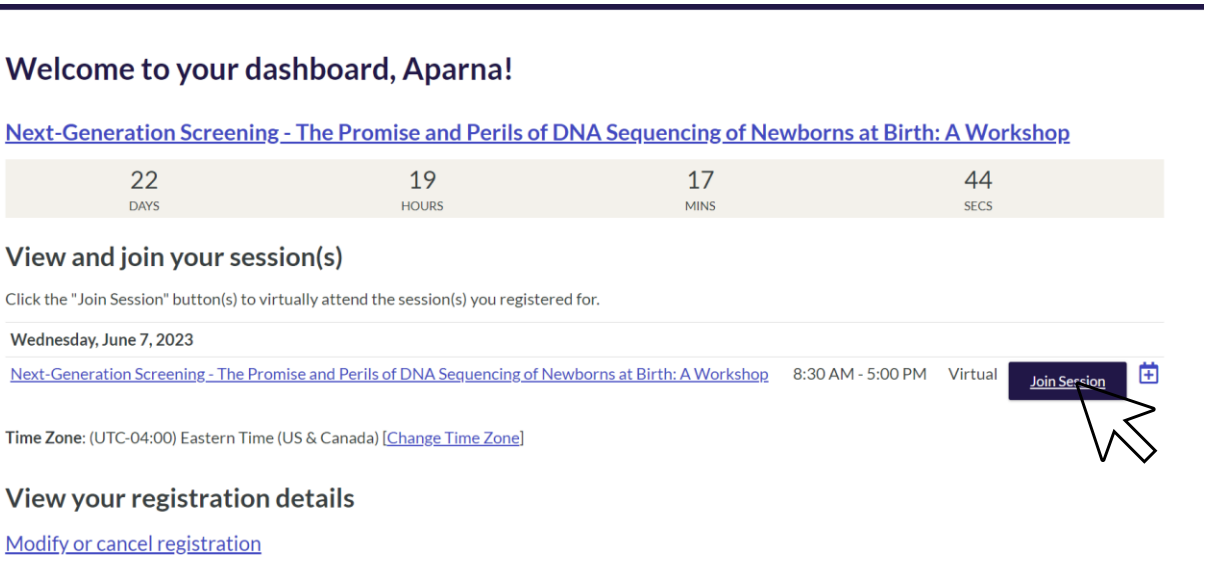
6. Read and acknowledge the Terms & Conditions.



7. After completing registration, you will receive an email confirmation. On the day of the workshop, click the link to your Dashboard.



8. To view the webcast, please click “Join Session”. If you click this link greater than 5 minutes before the workshop, you will see a countdown. At 8:25 AM ET, you will be automatically redirected to the webcast.



9. During the workshop, you can ask questions in the Slido box under the webcast.

AGENDA

Next-Generation Screening – The Promise and Perils of DNA Sequencing of Newborns at Birth: A Workshop

June 7, 2023

PURPOSE

A planning committee of the National Academies of Sciences, Engineering, and Medicine will organize and conduct a one-day public workshop to examine the utilization of DNA sequencing as a supplement to traditional newborn screening for conditions that are treatable, but not clinically evident in the newborn phase. The overarching goals of the workshop are to (1) examine the known and expected benefits, and potential harms, of the widespread utilization of newborn DNA sequencing, (2) explore the ethical and data security and ownership issues associated with DNA sequencing of newborns at birth, and (3) address issues of next-generation newborn screening equity in the United States.

The public workshop will feature invited presentations and discussions to:

- Explore the scope of recently initiated programs, such as those in the US, UK, and Australia, investigating newborn DNA sequencing as a screening tool in diverse healthy newborn populations and their relationship with established newborn screening efforts.
- Engage families, patient advocates, public health system representatives, and members of professional societies to provide their views on the need, impact, readiness, and risks of newborn DNA sequencing.
- Address equity of access to screening, on the assumption that newborn DNA sequencing may be less available, and less likely to identify pathogenic variants, in individuals from groups who are under-represented in genetic databases.

The planning committee will organize the workshop, develop the agenda, select and invite speakers and discussants, and moderate or identify moderators for the discussions. Proceedings-in-brief of the presentations and discussions at the workshop will be prepared by a designated rapporteur in accordance with institutional guidelines.

WEDNESDAY, JUNE 7, 2023

SESSION I: OPENING REMARKS & KEYNOTE

8:30 AM ET

Welcoming Remarks
Michelle Penny, *Roundtable Co-Chair*
Executive Vice President, Research & Development
Embark, Inc.

Greg Feero, *Roundtable Co-Chair*
Representing *Journal of the American Medical Association*
Professor, Department of Community and Family Medicine, Geisel School of Medicine
Faculty, Maine Dartmouth Family Medicine Residency Program

8:40–8:50 AM

Introduction and Charge to the Workshop Speakers and Participants
Natasha Bonhomme, *Workshop Planning Committee Co-Chair*
Founder
Expecting Health

Catherine Wicklund, *Workshop Planning Committee Co-Chair*
Representing National Society of Genetic Counselors
Co-Director, Graduate Program in Genetic Counseling
Past President, National Society of Genetic Counselors
Professor, Department of Obstetrics and Gynecology
Feinberg School of Medicine, Center for Genetic Medicine
Northwestern University

8:50–9:05 AM

Keynote
Aaron Goldenberg
Professor and Vice Chair
Department of Bioethics
Case Western Reserve University School of Medicine

9:05–9:30 AM

Panel of Discussants
Moderator: Karen Weck, Representing College of American Pathologists, University of North Carolina at Chapel Hill

Ellen Wright Clayton
Craig-Weaver Professor of Pediatrics, Center for Biomedical Ethics and Society
Professor of Law
Vanderbilt University Medical Center
Vanderbilt University

Robert Green
Professor, Medicine
Harvard Medical School
Geneticist, Medicine
Brigham and Women's Hospital

Crystal Grant
Former Technology Fellow
ACLU Speech, Privacy, and Technology Project

Mike Hu
Cofounder
Project GUARDIAN

SESSION II: LESSONS LEARNED FROM NEWBORN GENOMIC TESTING AND SCREENING

Moderator: April Adams, Baylor College of Medicine

Objectives

- Discuss lessons learned from programs that have implemented newborn whole genome sequencing:
    - What is informative for patient care (genes, conditions assessed)?
    - What information may provide value over the lifespan and where is there uncertainty or potential harms?

  - What are considerations around equity in clinical utility?
  - How are families dealing with the implications of having this information?
- Examine challenges in test interpretation and return of results.

9:30–9:45 AM

NC Nexus
Cynthia Powell
Professor of Pediatrics and Genetics
University of North Carolina School of Medicine

9:45–10:00 AM

NBSeq
Steven Brenner
Professor
Department of Bioengineering
Department of Molecular & Cell Biology
Department of Plant and Microbial Biology
University of California, Berkeley

10:00–10:15 AM

BeginNGS
Nathaly Sweeney
Assistant Professor of Pediatrics
University of California San Diego
Rady Children's Institute for Genomic Medicine

10:15–10:30 AM

BabySeq
Robert Green
Professor, Medicine
Harvard Medical School
Geneticist, Medicine
Brigham and Women's Hospital

10:30–10:50 AM

Panel Discussion

10:50–11:10 AM

Break

SESSION III: IMPLEMENTING NEWBORN SEQUENCING AT SCALE - HEALTH SYSTEM CHALLENGES & OPPORTUNITIES

Moderator: Greg Feero, Representing Journal of the American Medical Association, Maine Dartmouth Family Medicine Residency Program

Objectives

- Explore how well the current workforce is constituted to address the complexities surrounding newborn sequencing.
- Discuss logistics challenges that health systems may face including long term follow up and care and data management/integration/privacy/security.

11:10–11:25 AM ET

Sylvia Mann
Supervisor, Genomics Section
State of Hawaii Department of Health

11:25–11:40 AM

Holly Peay
Director, Early Check Program
Senior Research Scientist
RTI International

11:40–11:55 AM

David Veenstra
Professor
University of Washington

11:55 AM–12:25 PM **Panel Discussion**

12:25–1:20 PM **Lunch Break**

SESSION IV: DEPLOYING NEWBORN SEQUENCING RESPONSIBLY AND EQUITABLY

Moderator: Amy Gaviglio, Connetics Consulting, LLC

Objectives

- Discuss what defines readiness for system-wide deployment of newborn DNA sequencing. Explore how sequencing can meet the criteria for adoption of testing within newborn screening.
- Explore how barriers to access (e.g., cultural, workforce) could be addressed to decrease inequities.
- Examine the policy landscape for newborn sequencing in the U.S. and how this may affect access and reimbursement across geography and socioeconomic groups.
- Examine best practices for education and ongoing engagement of patients and communities, with particular emphasis on groups historically excluded from clinical research.

1:20–1:35 PM

Faith Fletcher
Assistant Professor
Baylor College of Medicine Center for Medical Ethics and Health Policy
Faculty Scholars Program
The Greenwall Foundation

1:35–1:50 PM

Cheedy Jaja
Associate Professor & Fulbright Scholar
College of Nursing
University of South Florida

1:50–2:10 PM **Patient Perspectives**

Teonna Woolford
CEO
Sickle Cell Reproductive Health Education Directive

Terri Klein
President and CEO
National MPS Society

2:10–2:40 PM **Panel Discussion**

2:40–3:00 PM **Break**

SESSION V: HOW WILL NEWBORN SEQUENCING CHANGE THE TRAJECTORY OF PRECISION HEALTH?

Moderator: Ryan Taft, Illumina

Objectives

- Discuss how genetic information ascertained at birth could be used across the lifespan and how this could help or hinder efforts to address health disparities.
- Discuss potential legal and ethical issues that should be addressed (e.g., informed consent, data privacy, regulatory landscape).

3:00–3:15 PM
David Bick
Clinical Advisor
Newborn Genomes Programme
Genomics England

3:15–3:30 PM
Noura Abul-Husn
Vice President of Genomic Health
23andMe

3:30–3:45 PM
Jeff Brosco
Director
Division of Services for Children with Special Health Needs
Health Resources and Services Administration

3:45–4:15 PM **Panel Discussion**

SESSION VI: FINAL REFLECTIONS

Moderator: Aaron Goldenberg, Case Western Reserve University

Objectives

- Explore what a world with public health NBS and clinical newborn sequencing looks like and how systems might interact with one another.
- What policies need to be put in place to ensure sequencing in newborn is appropriately supported, implemented and benefits the population?

4:15–5:00 PM
Panel Discussion
David Bick
Clinical Advisor
Newborn Genomes Programme
Genomics England

Ellen Wright Clayton
Craig-Weaver Professor of Pediatrics, Center for Biomedical Ethics and Society
Professor of Law
Vanderbilt University Medical Center
Vanderbilt University

Crystal Grant
Former Technology Fellow
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Mike Hu
Cofounder
Project GUARDIAN

Sylvia Mann
Supervisor, Genomics Section
State of Hawaii Department of Health

5:00–5:10 PM

Wrap Up
Natasha Bonhomme, *Workshop Planning Committee Co-Chair*
Founder
Expecting Health

ANNOUNCEMENTS

NATIONAL ACADEMIES Sciences Engineering Medicine

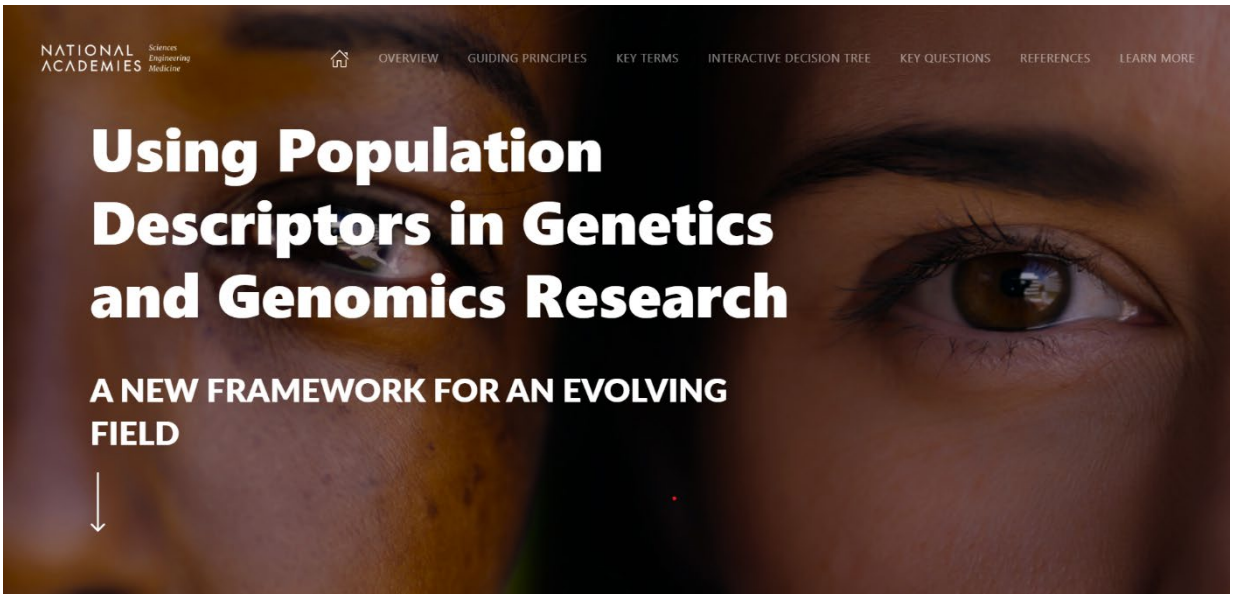
NASEM Consensus Study Interactive and Report

Using Population Descriptors in Genetics and Genomics Research:

A New Framework for an Evolving Field

Research using human genetic data has grown exponentially over the past decade, but challenges persist in the use of race, ethnicity, and other population descriptors. This new resource page summarizes our recent report which describes a framework for change, including guiding principles and best practices for research. It also features an interactive decision tree to help guide researchers in selecting appropriate population descriptors based on the type of study they are conducting.

Access the interactive: https://nap.edu/resource/26902/interactive/



Report Link: https://nap.nationalacademies.org/catalog/26902/

More resources are available through the project page

A recording of the report release webinar can be found here

New Releases

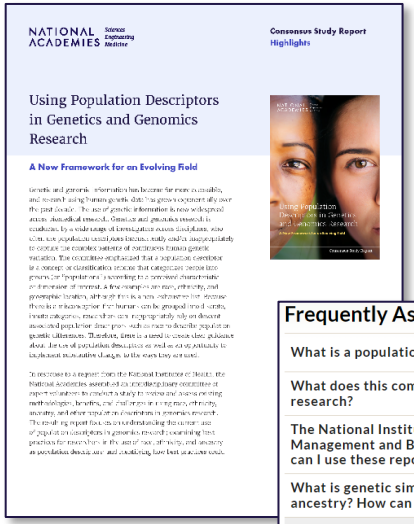
Action Guides

- Journals & Professional Societies
- Funders & Research Institutions

Release Webinar Recording

Briefing Slides

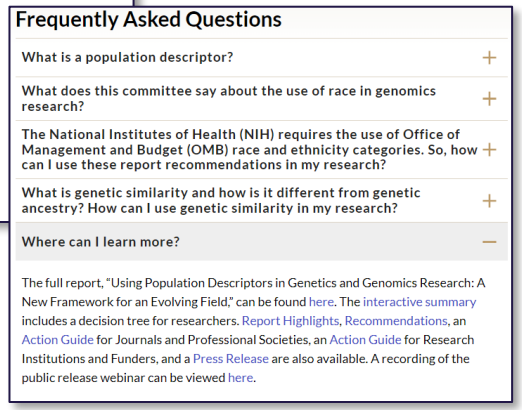
FAQs Section


NATIONAL ACADEMIES Sciences Engineering Medicine

Consensus Study Report Highlights

Using Population Descriptors in Genetics and Genomics Research

A New Framework for an Evolving Field


Frequently Asked Questions

What is a population descriptor? +

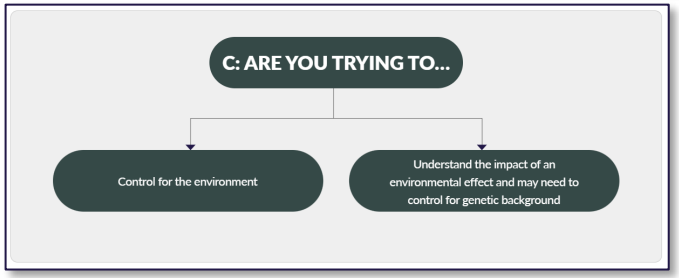
What does this committee say about the use of race in genomics research? +

The National Institutes of Health (NIH) requires the use of Office of Management and Budget (OMB) race and ethnicity categories. So, how can I use these report recommendations in my research? +

What is genetic similarity and how is it different from genetic ancestry? How can I use genetic similarity in my research? +

Where can I learn more? −

The full report, "Using Population Descriptors in Genetics and Genomics Research: A New Framework for an Evolving Field," can be found here. The interactive summary includes a decision tree for researchers. Report Highlights, Recommendations, an Action Guide for Journals and Professional Societies, an Action Guide for Research Institutions and Funders, and a Press Release are also available. A recording of the public release webinar can be viewed here.



Report Resources

Report Highlights

Recommendations 3-pager

NEW! Action Guides

- Journals & Professional Societies
- Funders & Research Institutions

Interactive Webpage and Decision Tree

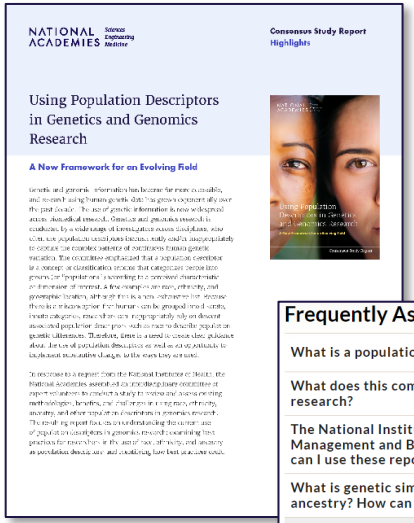
NEW! Release Webinar Recording

NEW! Briefing Slides

NEW! FAQs Section

Ancestry Figure Animation *(in progress)*

Audio Files *(in progress)*

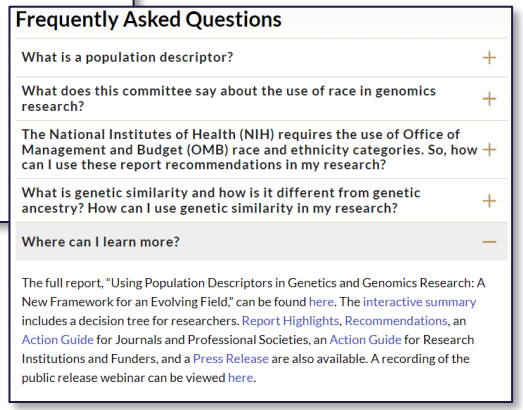

NATIONAL ACADEMIES Sciences Engineering Medicine

Consensus Study Report Highlights

Using Population Descriptors in Genetics and Genomics Research

A New Framework for an Evolving Field

Access these resources through the study page or report page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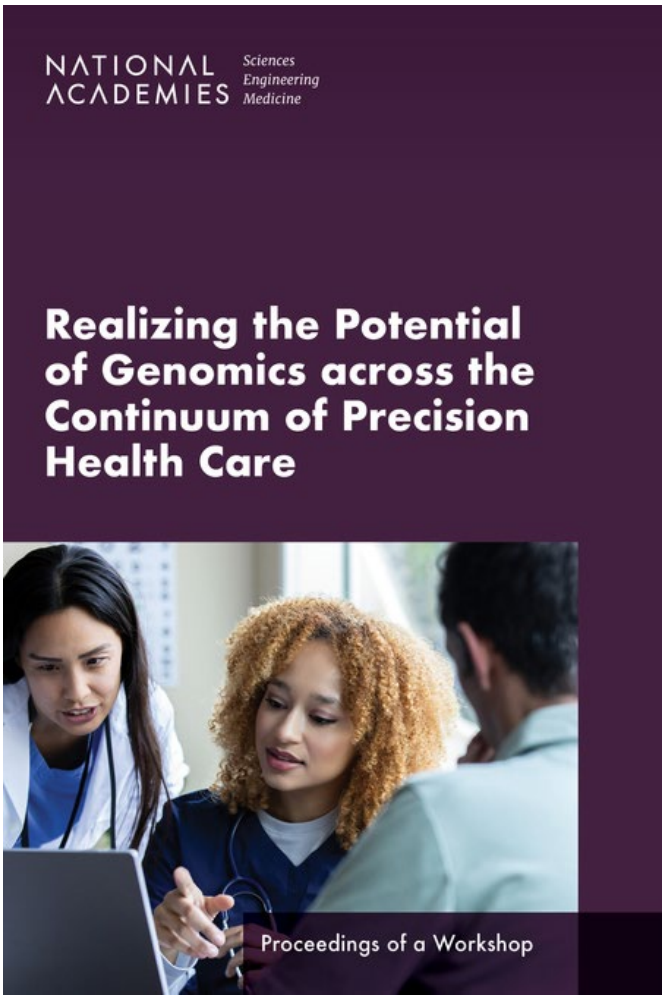
NATIONAL ACADEMIES Sciences Engineering Medicine

New Proceedings of a Workshop!

Realizing the Potential of Genomics across the Continuum of Precision Health Care

Link: https://nap.edu/26917

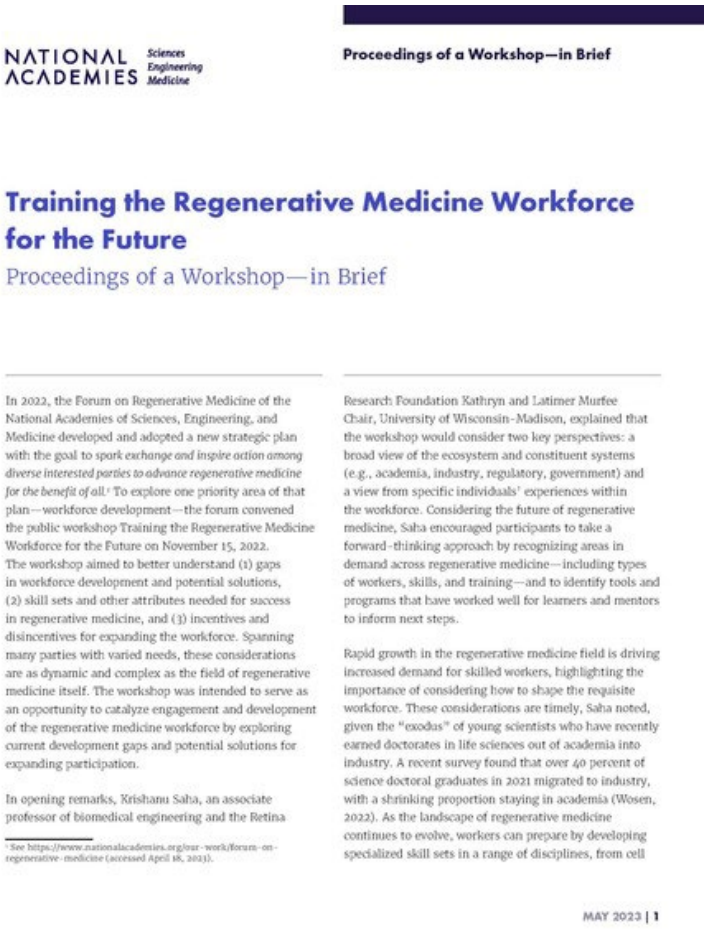


NATIONAL ACADEMIES Sciences Engineering Medicine

New Proceedings in Brief of a Workshop!

Training the Regenerative Medicine Workforce for the Future

Link: https://doi.org/10.17226/27013



NATIONAL ACADEMIES Sciences Engineering Medicine

Proceedings of a Workshop—in Brief

Training the Regenerative Medicine Workforce for the Future

Proceedings of a Workshop—in Brief

In 2022, the Forum on Regenerative Medicine of the National Academies of Sciences, Engineering, and Medicine developed and adopted a new strategic plan with the goal to *spark exchange and inspire action among diverse interested parties to advance regenerative medicine for the benefit of all.*[1] To explore one priority area of that plan—workforce development—the forum convened the public workshop Training the Regenerative Medicine Workforce for the Future on November 15, 2022. The workshop aimed to better understand (1) gaps in workforce development and potential solutions, (2) skill sets and other attributes needed for success in regenerative medicine, and (3) incentives and disincentives for expanding the workforce. Spanning many parties with varied needs, these considerations are as dynamic and complex as the field of regenerative medicine itself. The workshop was intended to serve as an opportunity to catalyze engagement and development of the regenerative medicine workforce by exploring current development gaps and potential solutions for expanding participation.

In opening remarks, Krishanu Saha, an associate professor of biomedical engineering and the Retina Research Foundation Kathryn and Latimer Murfee Chair, University of Wisconsin–Madison, explained that the workshop would consider two key perspectives: a broad view of the ecosystem and constituent systems (e.g., academia, industry, regulatory, government) and a view from specific individuals' experiences within the workforce. Considering the future of regenerative medicine, Saha encouraged participants to take a forward-thinking approach by recognizing areas in demand across regenerative medicine—including types of workers, skills, and training—and to identify tools and programs that have worked well for learners and mentors to inform next steps.

Rapid growth in the regenerative medicine field is driving increased demand for skilled workers, highlighting the importance of considering how to shape the requisite workforce. These considerations are timely, Saha noted, given the "exodus" of young scientists who have recently earned doctorates in life sciences out of academia into industry. A recent survey found that over 40 percent of science doctoral graduates in 2021 migrated to industry, with a shrinking proportion staying in academia (Wosen, 2022). As the landscape of regenerative medicine continues to evolve, workers can prepare by developing specialized skill sets in a range of disciplines, from cell

[1] See https://www.nationalacademies.org/our-work/forum-on-regenerative-medicine (accessed April 18, 2023).

MAY 2023 | 1

NATIONAL ACADEMIES *Sciences Engineering Medicine*

New Consensus Study!

Toward Sequencing and Mapping of RNA Modifications

Statement of Task:

The National Academies of Sciences, Engineering, and Medicine will convene an ad hoc committee to conduct a study on direct sequencing of modifications of RNA (referred to as epitranscriptome) from humans and model organisms. As part of this study, the committee will develop a roadmap for achieving direct sequencing of modifications of RNA. The National Academies' committee will examine:

- Scientific needs for sequencing of modifications of RNA.
- Current RNA sequencing methodologies and technologies and their current limitations, documenting which RNA bases and modifications can be sequenced and whether the RNA is sequenced directly or indirectly.
- Current RNA and RNome* databases, including the types of RNA included, the organisms from which the RNA sequences were obtained, and limitations of the databases.
- Challenges associated with current methodologies and technologies for sequencing RNA and its modifications for scientific, clinical, and public health analyses.
- Scientific and technological hurdles including computational and analytic technologies, that need to be overcome to achieve direct sequencing of RNA modifications.
- Computational and analytic technologies needed to analyze modified RNA.
- Data ecosystem for supporting sequencing and analysis of RNA modifications.
- Policy, workforce, and infrastructure needs to support sequencing and analysis of RNA modifications.
- Potential for existing and new technologies to discover previously-unrecognized RNA modifications.

The National Academies will produce a consensus report that focuses on defining the science and technology roadmap and actionable recommendations toward achievement of direct sequencing of RNA modifications. During the study, the committee will hold a series of coordinated activities (e.g., workshops, meeting of experts, ideation challenges) to provide platforms for creative collaboration among experts from multiple scientific disciplines and organizations. Individual products summarizing key outcomes from these activities may be produced as interim products of the consensus study.

* The RNome refers to all of the RNA species in a cell at a given time (Arora, 2018).

NATIONAL ACADEMIES Sciences Engineering Medicine

Toward Sequencing and Mapping of RNA Modifications

Upcoming Public Event:

Meeting of Experts on Education and Workforce Development
June 21, 2:30-4:30 PM ET
See more details here!

Committee Member Roster:

Co-Chairs

Brenda Bass
University of Utah

Taekjip Ha
Johns Hopkins University

Members

Nicholas M. Adams
Thermo Fisher Scientific

Juan D. Alfonzo
The Ohio State University

Jeffery Baker
NIIMBL

Susan Baserga
Yale University

Lydia M. Contreras
University of Texas at Austin

Markus Hafner
NIAMS/NIH

Sarath C. Janga
Indiana University School of Medicine

Patrick A. Limbach
University of Cincinnati

Julius B. Lucks
Northwestern University

Mary A. Majumder
Baylor College of Medicine

Nicole M. Martinez
Stanford University

Kate D. Meyer
Duke University School of Medicine

Keith Robert Nykamp
Invitae Corporation

Tao Pan
University of Chicago

NATIONAL ACADEMIES Sciences Engineering Medicine

Workshop Recordings Posted!

The Potential Contribution of Cancer Genomics Information to Community Investigations of Unusual Patterns of Cancer: A Workshop

Link: https://www.nationalacademies.org/event/04-13-2023/the-potential-contribution-of-cancer-genomics-information-to-community-investigations-of-unusual-patterns-of-cancer-a-workshop

GENOMICS ROUNDTABLE INFORMATION

NATIONAL ACADEMIES Sciences Engineering Medicine

Roundtable on GENOMICS and PRECISION HEALTH

The sequencing of the human genome is rapidly opening new doors to research and progress in biology, medicine, and health care. At the same time, these developments have produced a diversity of new issues to be addressed.

The National Academies of Sciences, Engineering, and Medicine has convened a Roundtable on Genomics and Precision Health (previously the Roundtable on Translating Genomic-Based Research for Health) that brings together leaders from academia, industry, government, foundations and associations, and representatives of patient and consumer interests who have a mutual concern and interest in addressing the issues surrounding the translation of genome-based research for use in maintaining and improving health. The mission of the Roundtable is to advance the field of genomics and improve the translation of research findings to health care, education, and policy. The Roundtable will discuss the translation process, identify challenges at various points in the process, and discuss approaches to address those challenges.

The field of genomics and its translation involves many disciplines, and takes place within different economic, social, and cultural contexts, necessitating a need for increased communication and understanding across these fields. As a convening mechanism for interested parties from diverse perspectives to meet and discuss complex issues of mutual concern in a neutral setting, the Roundtable: fosters dialogue across sectors and institutions; illuminates issues, but does not necessarily resolve them; and fosters collaboration among interested parties.

To achieve its objectives, the Roundtable conducts structured discussions, workshops, and symposia. Workshop summaries will be published and collaborative efforts among members are encouraged (e.g., journal articles). Specific issues and agenda topics are determined by the Roundtable membership, and span a broad range of issues relevant to the translation process.

Issues may include the integration and coordination of genomic information into health care and public health including encompassing standards for genetic screening and testing, improving information technology for use in clinical decision making, ensuring access while protecting privacy, and using genomic information to reduce health disparities. The patient and family perspective on the use of genomic information for translation includes social and behavioral issues for target populations. There are evolving requirements for the health professional community, and the need to be able to understand and responsibly apply genomics to medicine and public health.

Of increasing importance is the need to identify the economic implications of using genome-based research for health. Such issues include incentives, cost-effectiveness, and sustainability.

Issues related to the developing science base are also important in the translation process. Such issues could include studies of gene-environment interactions, as well as the implications of genomics for complex disorders such as addiction, mental illness, and chronic diseases.

Roundtable sponsors include federal agencies, pharmaceutical companies, medical and scientific associations, foundations, and patient/public representatives. For more information about the Roundtable on Genomics and Precision Health, please visit our website at nationalacademies.org/GenomicsRT or contact Sarah Beachy at 202-334-2217, or by e-mail at sbeachy@nas.edu.

500 Fifth Street, NW, Washington, DC 20001
Phone 202.334.2217 E-mail sbeachy@nas.edu nationalacademies.org/GenomicsRT

Roundtable on Genomics and Precision Health Membership

W. Gregory Feero, M.D., Ph.D. (Co-Chair) *JAMA*
Michelle Penny, Ph.D. (Co-Chair) Embark, Inc.

Naomi Aronson, Ph.D.
BlueCross/BlueShield Association

Aris Baras, M.D., M.B.A.
Regeneron Pharmaceuticals

Vence Bonham, Jr., J.D.
National Human Genome Research Institute

Jeffrey P. Brosco, M.D., Ph.D.
Health Resources and Services Administration

Bernice Coleman, Ph.D., ACNP-BC, FAHA, FAAN
American Academy of Nursing

Robert B. Darnell, M.D., Ph.D.
The Rockefeller University / NY Genome Center

Daniel Edelman, Ph.D.
Food and Drug Administration

Geoffrey Ginsburg, M.D., Ph.D.
Global Genomic Medicine Collaborative (G2MC)

Jennifer Goldsack, MChem, M.A., M.B.A.
Digital Medicine Society (DiMe)

Richard Hodes, M.D.
National Institute on Aging

Geoff Hollett, Ph.D.
American Medical Association

Mira Irons, M.D., FACMG
College of Physicians of Philadelphia

Praduman Jain, M.S.
Vibrent Health

Katherine Johansen Taber, Ph.D.
Myriad Women's Health

Sekar Kathiresan, M.D.
Massachusetts General Hospital

Muin Khoury, M.D., Ph.D.
Centers for Disease Control and Prevention

Charles Lee, Ph.D., FACMG
The Jackson Laboratory for Genomic Medicine

Christa Lese Martin, Ph.D., FACMG
Geisinger

Mona Miller, M.P.P.
American Society of Human Genetics

Adele Mitchell, Ph.D.
Biogen

Jennifer Moser, Ph.D.
U.S. Department of Veterans Affairs

Kenneth Offit, M.D.
American Society of Clinical Oncology

Kathryn Phillips, Ph.D.
University of California, San Francisco

Victoria M. Pratt, Ph.D., FACMG
Association for Molecular Pathology

Murray Ross, Ph.D.
Kaiser Foundation Health Plan, Inc.

Daniel C. Edelman, Ph.D.
U.S. Food and Drug Administration

Nadeem Sarwar, Ph.D.
Eisai Inc.

Sheri Schully, Ph.D.
All of Us Research Program, NIH

Geetha Senthil, Ph.D.
National Institute of Mental Health

Anil Shanker, Ph.D.
Meharry Medical College

Nonniekaye Shelburne, CRNP, M.S., AOCN
National Cancer Institute

Ryan Taft, Ph.D.
Illumina

Jacquelyn Taylor, Ph.D.
Columbia University

Sharon Terry, M.A.
Genetic Alliance

Joyce Tung, Ph.D.
23andMe, Inc.

Jameson Voss, M.D., M.P.H., FACPM
U.S. Air Force

Karen Weck, M.D.
College of American Pathologists

Melanie J. Wells, M.P.H., CAE
American College of Medical Genetics and Genomics

Catherine A. Wicklund, M.S., CGC
National Society of Genetic Counselors

Robert S. Wildin, M.D.
University of Vermont Health Network

Huntington F. Willard, Ph.D.
Genome Medical, Inc.

Sarah Wordsworth, Ph.D.
University of Oxford

National Academy of Medicine Fellow
Paule Joseph, Ph.D., M.S., FNP-BC, FAAN
Inaugural American Academy of Nursing Fellow at NAM

Project Staff
Sarah H. Beachy, Ph.D., *Roundtable Director*
Kathryn Asalone, Ph.D., *Associate Program Officer*
Meredith Hackmann, *Associate Program Officer*
Samantha Schumm, Ph.D., *Associate Program Officer*
Lydia Teferra, *Research Associate*
Aparna Cheran, *Senior Program Assistant*

VISION	**Realizing the full potential of health for all through genomics and precision health.**
MISSION	**We bring together diverse voices to encourage innovation and actions that foster the wide adoption of and equitable access to the benefits of genomics and precision health.**

As a group of committed parties, we believe in…

- Creating an inclusive and optimistic environment for discussion
- Learning from successes and missteps in the field
- Demanding reproducible evidence-based science
- Sharing trustworthy information
- Embracing interdisciplinary strategies
- Optimizing data privacy and security
- Advancing health equity in all that we do

The Roundtable focuses its energy and resources on these priorities:

DRIVE **INNOVATION** IN GENOMICS AND PRECISION HEALTH	Identify the competing barriers and facilitators of innovation for genomics-based diagnostics, risk assessment tools, and therapies. Leverage opportunities to learn from and promote innovative approaches that can accelerate commercialization and integration to drive impact of genomics on precision health.
SPUR THE **ADOPTION** OF GENOMICS-BASED TOOLS AND PRECISION HEALTH APPROACHES	Cultivate evidence-based practices across the health care and public health systems for adopting genomics and precision health. Draw attention to gaps in adoption and their root causes and highlight potential solutions.
ACHIEVE **EQUITY** IN GENOMICS AND PRECISION HEALTH	Foster action related to underrepresentation and inequities in genomic research, workforce, and access to genomic services by people who need them. Look internally to improve the processes and practices the Roundtable employs to achieve its mission.
SHAPE THE **POLICY DIALOGUE** ABOUT GENOMICS AND PRECISION HEALTH	Accelerate the dissemination of actionable knowledge to shape practice and increase public awareness. Inform and influence how decisions are made.

DEFINITIONS

Precision Health | Inclusive of precision medicine, precision health is a broader, proactive and people-focused approach to health, relying on individual-focused care and everyday decision-making to better predict, prevent, and treat disease.

Genetics | Study of heredity, genes, and genetic variation.

Genomics | Study of the genome by using DNA sequencing and other technologies to understand gene structure, function, and regulation.

WORKSHOP INFORMATION

Next-Generation Screening – The Promise and Perils of DNA Sequencing of Newborns at Birth: A Workshop

Roundtable on Genomics and Precision Health
June 7, 2023

Planning Committee Member Roster

Co-Chairs

Natasha Bonhomme
Founder
Expecting Health

Cathy Wicklund, M.S., CGC
Co-Director, Graduate Program in Genetic Counseling
Past President, National Society of Genetic Counselors
Professor, Department of Obstetrics and Gynecology
Feinberg School of Medicine, Center for Genetic Medicine
Northwestern University

Members

April Adams, M.D., M.S., FACOG, FACMG
Assistant Professor of Obstetrics & Gynecology, Maternal Fetal Medicine
Assistant Professor of Molecular & Human Genetics, Prenatal Genetics
Assistant Professor of Obstetrics & Gynecology, Reproductive and Prenatal Genetics
Assistant Program Director, Maternal Fetal Medicine/Maternal Fetal Medicine & Medical
Genetics and Genomics Fellowships Assistant Director of Education, Reproductive and Prenatal Genetics
Baylor College of Medicine
Maternal Medical Director, Quality and Safety
Harris Health System, Ben Taub Hospital

Amy Gaviglio, M.S., CGC
Founder
Genetic Counselor
Public Health Genetics and Rare Disease Consultant
Connetics Consulting, LLC

Aaron Goldenberg, M.D., Ph.D.
Director of Research
Professor and Vice Chair
Department of Bioethics
Professor, Department of Genetics and Genome Sciences
School of Medicine
Member, Population and Cancer Prevention Program
Case Western Reserve University

Alex Kemper, M.D., M.P.H., M.S.
Division Chief, Primary Care Pediatrics
Nationwide Children's Hospital
Professor of Pediatrics
The Ohio State University College of Medicine

Molly McGinnis, M.S., CGC
Senior Director, Client and Partner Relations
Population Genomics
Genome Medical, Inc.

Ryan Taft, Ph.D.
Vice President, Scientific Research
Illumina, Inc.

Next-Generation Screening – The Promise and Perils of DNA Sequencing of Newborns at Birth: A Workshop

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Joyce Tung, Ph.D.
Vice President, Research
23andMe, Inc.

Karen Weck, M.D.
Representing College of American Pathologists
Professor of Pathology & Laboratory Medicine
Professor of Clinical Genetics
University of North Carolina at Chapel Hill

NATIONAL ACADEMIES Sciences Engineering Medicine

Board on Health Sciences Policy

Planning Committee Member Biographies

Natasha Bonhomme *(Co-chair)*, is the Founder of Expecting Health and has over 15 years of nonprofit and maternal and child health experience. She launched Expecting Health to bring a range of family, community, and professional stakeholders together to address the need for clearer information, high quality engagement, and scalable solutions in healthcare. Her focus is on centering families' perspectives into policy and program design and implementation. Natasha led and managed an extensive study of women (with more than 2,000 expectant and new mothers) to understand their attitudes towards newborn screening and their preferences on how and when to be educated. She created and oversees Baby's First Test a national resource center which reaches over 600,000 families and health providers annually. As director of Baby's First Test, Natasha has testified before the US Senate Health, Education, Labor, and Pension Committee's Subcommittee on Children and Families on the importance of public education on screening. She sits on numerous committees on maternal and child health including the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.

Outside of the office, Natasha has been involved in numerous community-based initiatives and currently is a Board Member of Whitman Walker Health, a DC-based federally-qualified health center focused on offering affirming community-based health and wellness services to all with a special expertise in LGBTQ and HIV care.

Catherine A. Wicklund, M.S., CGC *(Co-chair)*, is the Co-Director of the Graduate Program in Genetic Counseling at Northwestern University and a Professor in the Department of Obstetrics and Gynecology. She has over 20 years of experience in clinical genetic counseling and has provided prenatal and pediatric genetic services. She served on the Board of Directors of the National Society of Genetic Counselors first as Region V Representative, then as Secretary and was President in 2007. She has served on several national and regional committees including the Advisory Committee on Hereditable Disorders in Newborns and Children, the American Society of Human Genetics representative on the Scientific Program Committee of the 2016 International Congress of Human Genetics and the NSGC representative on the NASEM Roundtable on Genomics and Precision Health since 2007. Ms. Wicklund's research interests include issues regarding genome/exome sequencing and precision medicine, psychosocial and counseling issues, and issues around diversity, equity, inclusion and justice, workforce and access to and delivery of genetic services. She has contributed to the Electronic Medical Records and Genomics (eMERGE) Network, which aims to bring personalized medicine into broader clinical use. Her current research grants are focused on the education of cardiology providers on hereditary cardiomyopathies and utilizing chatbots in living donors with *APOL1* variants regarding kidney donation decision making with nephrologists. She received her Master of Science degree in Genetic Counseling from the University of Texas-Graduate School of Biomedical Sciences and is a diplomat of the American Board of Genetic Counseling.

April Adams, M.D., M.S., FACOG, FACMG, is currently an assistant professor at Baylor College of Medicine in the departments of Obstetrics and Gynecology and Molecular and Human Genetics. She is also the associate program director for the maternal fetal medicine and combined maternal fetal medicine/medical genetics fellowship programs and the Maternal Medical Director for Quality and Safety at Ben Taub Hospital. She completed her completed residency training in obstetrics and gynecology and the University of Minnesota. She then completed fellowship training in both maternal fetal medicine and medical genetics at MedStar Washington Hospital Center and the National Human Genome Research Center. Her research and clinical interests are in placental development, pregnancy loss and stillbirth, health disparities in reproductive genetics.

Amy Gaviglio, M.S., CGC, is a certified genetic counselor and founder of Connetics Consulting, which provides public health genetics, genomics, and rare disease services across the country and has been working in the newborn screening and rare disease space for the past 15 years. Amy currently works with the Centers for Disease Control and Prevention's Newborn Screening and Molecular Biology Branch, the Association of Public Health Laboratories (APHL), Expecting Health, and several other genetics and rare disease organizations. She is co-chair of APHL's New Disorders in Newborn Screening workgroup and is a member of additional national groups including the Legal & Legislative Issues in Newborn Screening workgroups, the Rare Disease Diversity Coalition, and EveryLife Foundation's Community Congress. She also serves as an Advisor for the Midwest Genetics Network and the Innovations in Newborn Screening Interoperability Center. Finally, Amy serves as Chair of the NBS Expert Panel for the Clinical and Laboratory Standards Institute and is currently the Chair of Minnesota's Rare Disease Advisory Council.

Aaron Goldenberg, Ph.D., is a Professor and Vice-Chair in the Department of Bioethics at Case Western Reserve University. He is also Director for the CWRU Bioethics Center for Community Health and Genomic Equity. Dr. Goldenberg has a background in bioethics, health behavior/health education, public health ethics, and public health genetics. He has focused his work on the ethical, legal, and social issues associated with the integration of new genomic technologies into research, clinical and public health settings. Dr. Goldenberg's research program has been grounded by a number of major project areas, including: 1) the ethical and social implications of storing and using of biological specimens and data for future research; 2) implications of genetics and gene-environment interactions for racial/ethnic minorities and other communities experiencing health disparities; and 3) ethical implications of expanding newborn screening programs. He is currently Co-Chair the NBSTRN Bioethics and Legal Workgroup, and with this group, led a recent effort to develop a set ELSI questions that can be integrated into NBS Pilots to help assess the potential benefits and harms of adding new conditions to NBS panels. He is also Co-Chair for the Legal and Legislative Issues in Newborn Screening Workgroup for the Association of Public Health Laboratories.

Alex R. Kemper, M.D., M.P.H., M.S., is the Division Chief of Primary Care Pediatrics at Nationwide Children's Hospital and Professor of Pediatrics at the Ohio State University College of Medicine and Deputy Editor of *Pediatrics*. Dr. Kemper completed his pediatric residency training at Duke University followed by combined fellowship training in health services research and medical informatics with residency training in preventive medicine at the University of North Carolina. His research focuses on preventive services in the primary care practice setting. Dr. Kemper serves as the Chair of the Evidence Review Group for the US Secretary of Health and Human Services Advisory Committee on Heritable Disorders in Newborns and Children.

Molly McGinniss, M.S., CGC, is Senior Director of Client and Partner Relations at Genome Medical, a digital health company and specialty medical group focused on genetics and genomics. She is a board-certified genetic counselor with extensive clinical and commercial expertise. She began her career as a clinical genetic counselor providing prenatal, pediatric, and adult genetic counseling services. Prior to joining Genome Medical, she led global market development at Illumina for the genetic disease clinical segment. Molly serves as an adjunct professor and advisory board member for several genetic counseling graduate training programs and has held past leadership roles for the National Society of Genetic Counselors (NSGC). Ms. McGinnis has served as the Program Review Committee chair prior to being elected President.

Ryan Taft, Ph.D., is Director of Scientific Research at Illumina Inc. with a focus on the development and deployment of diagnostic whole genome sequencing worldwide, particularly for patients with rare and undiagnosed genetic disease. He obtained his Bachelor of Science in Biochemistry and Molecular Biology from the University of California, Davis on a Regent's Scholarship, and his Ph.D. in Genomics and Computational Biology from the University of Queensland on a U.S. National Science Foundation Research Fellowship. He was previously a Group Leader & Senior Research Fellow at the University of Queensland, where he still holds a minor appointment, and is Adjunct Associate Professor at the George Washington University School of Medicine and Health Sciences. He has published in a variety of well-regarded journals – including articles in *Nature Genetics*, *Nature Structural and Molecular Biology*, *the American Journal of Human Genetics*, and others. He is a founding member of the Global Leukodystrophy Initiative (GLIA), and is delighted to help drive solutions for patients with rare disease through Global Genes.

Joyce Tung, Ph.D., joined 23andMe in 2007 and manages the 23andMe research team, which is responsible for consumer health and ancestry research and development, academic and industry collaborations, computational analyses for therapeutics, and new research methods and tools development. While a postdoctoral fellow at Stanford University, Joyce studied the genetics of mouse and human pigmentation. She graduated from Stanford with honors and distinction with a B.S. in Biological Sciences and a minor in computer science, and earned her Ph.D. in Genetics from the University of California, San Francisco where she was a National Science Foundation graduate research fellow.

Karen Weck, M.D., is Clinical Professor of Pathology & Laboratory Medicine and Clinical Professor of Genetics at the University of North Carolina School of Medicine, Director of Molecular Genetics and Pharmacogenomics, and Medical Director of the Clinical Molecular Genetic Pathology Laboratory for UNC Hospitals. She serves as the College of American Pathologists (CAP) liaison to the National Academy of Medicine Roundtable on Genomics and Precision Medicine. Dr. Weck served as President of the Association for Molecular Pathology (AMP) in 2020 and Chair of the AMP Executive Committee and Board of Directors. She is currently Co-chair of the AMP Professional Relations Committee and Co-chair of the AMP Clinical Practice Committee Pharmacogenomics Working Group. She is Past Chair of the College of American Pathologists Molecular Pathology and Genomics Cluster, Biochemical and Molecular Genetics Resource Committee, and Pharmacogenomics Workgroup. She received a Distinguished Service Award from the College of American Pathologists in 2018 and the Jeffrey A. Kant Leadership Award from the Association for Molecular Pathology in 2022.

Dr. Weck received Bachelor of Science and Doctor of Medicine degrees from Duke University, residency training in Laboratory Medicine at Washington University in St. Louis, and is boarded in Clinical Pathology and Molecular Genetic Pathology. She has served as a consultant or advisor to numerous commercial and professional organizations and is a past member of the FDA Molecular and Clinical Genetics Devices Panel. She is an Associate Editor for Genetics in Medicine, the official journal of the American College of Medical Genetics and Genomics, and is a member of the editorial board for the Journal of Molecular Diagnostics, the official journal of the Association for Molecular Pathology.

Dr. Weck has authored over 100 peer reviewed articles, invited editorials and book chapters related to molecular diagnostics and genomics. Dr. Weck has been co-investigator of several NIH-funded grants to implement exome/genome sequencing for diagnosis of genetic diseases from prenatal testing to adults and for newborn screening. She has been co-investigator of institutional clinical trials to study the clinical utility of pharmacogenomics guided therapy and of the UNCseq tumor sequencing study to implement genomic sequencing for somatic mutation testing in cancer. Dr. Weck is a founding member of the steering committee for the UNC Program for Precision Medicine in HealthCare (PPMH) and a member of the UNC Precision Health Genetic Screening Program, EPIC Genomics Implementation Committee, ISD Genetics and Genomics Governance Committee, and Lineberger Comprehensive Cancer Center Genetics Advisory Committee.

NATIONAL ACADEMIES Sciences Engineering Medicine

Board on Health Sciences Policy

Roundtable on Genomics and Precision Health
June 7, 2023

Speaker Biographies

Noura Abul-Husn, M.D., Ph.D., is the Vice President of Genomic Health at 23andMe. She leads 23andMe's clinical strategy to integrate consumer genetics into health care and provides genomic medicine expertise to direct company-wide health initiatives. In addition to her leadership role at 23andMe, Dr. Abul-Husn is an Associate Professor of Medicine at the Icahn School of Medicine at Mount Sinai, where she previously led genomic medicine efforts as founding Clinical Director of the Institute for Genomic Health and founding Chief of the Division of Genomic Medicine in the Department of Medicine. Prior to that, she served as Director of Translational Genetics at the Regeneron Genetics Center.

Dr. Abul-Husn's scientific contributions include pioneering genome-first approaches in ancestrally diverse populations to provide novel clinical insights and inform population genomic screening. Her work has been published in leading journals, including *Science*, *Cell*, and the *New England Journal of Medicine*. At Mount Sinai, Dr. Abul-Husn launched a genomic screening program tailored to ancestrally diverse populations. She established the Genomic Health Clinic to provide the infrastructure for emerging genomic medicine applications and started a Genomic Medicine Track for Internal Medicine residents to expand genomics knowledge across specialties. She is a principal investigator in the NIH-funded eMERGE (electronic Medical Records and Genomics) Network, which is integrating genomic risk assessments into routine clinical care.

Dr. Abul-Husn is a double board-certified Internist and Medical Geneticist. She completed her M.D,. Ph.D., and residency at Mount Sinai in New York, and was elected to the Alpha Omega Alpha Medical Honor Society. She is the recipient of numerous awards, including a 40 under 40 Rising Star Award and the Dr. Michael S. Watson Genetic & Genomic Medicine Innovation Award from the ACMG Foundation for Genetic and Genomic Medicine.

David Bick, M.D., is the Principal Clinician for the Newborn Genomes Programme at Genomics England. Prior to his work in England, he was the Chief Medical Officer and a faculty investigator at the HudsonAlpha Institute for Biotechnology. Dr. Bick also served as the Medical Director of the Smith Family Clinic for Genomic Medicine located on the campus of HudsonAlpha Institute for Biotechnology and the Laboratory Director of the HudsonAlpha Clinical Services Laboratory.

He came to HudsonAlpha from the Medical College of Wisconsin where he was Professor in the Department of Pediatrics and the Department of Obstetrics & Gynecology at the Medical College of Wisconsin. At the Medical College of Wisconsin he was the Director of the Clinical Sequencing Laboratory, Director of the Advanced Genomics Laboratory at Children's Hospital of Wisconsin, Medical Director of the Genetics Clinic at Children's Hospital of Wisconsin, and Chief of the Division of Genetics in the Department of Pediatrics at Medical College of Wisconsin.

Dr. Bick received his medical degree from George Washington University School of Medicine in 1981 and completed his residency in Pediatrics at Yale-New Haven Hospital in New Haven, CT. At the Yale University School of Medicine, Dr. Bick completed a fellowship in Human Genetics and Pediatrics in 1986, followed by a post-doctoral research fellowship in Human Genetics in 1987. Dr. Bick is board certified in Pediatrics, Clinical Genetics, and Clinical Molecular Genetics.

He is a leader in the field of genomic medicine and has published numerous peer-reviewed articles, chapters, and reviews. Dr. Bick's laboratories at the Medical College of Wisconsin and Children's Hospital of Wisconsin were the first in the world to offer whole genome sequencing as a clinical test. He also developed the first Genomic Medicine Clinic in the United States.

Steven Brenner, Ph.D., M.Phil, research is primarily in the area of computational genomics, including genome variation interpretation, genomic privacy, RNA regulation, protein structure evolution, and function prediction. He has a particular interest in the use of genome sequencing in newborns for population screening and new methods for genetic diagnosis.

Brenner's undergraduate research was in the first genome laboratory, mentored by Walter Gilbert at Harvard. He received his M.Phil. from the Department of Biochemistry at Cambridge University, and earned a Ph.D. from the University of Cambridge and the MRC Laboratory of Molecular Biology where he studied with Cyrus Chothia. Brenner had a fellowship at the Japan National Institute of Bioscience, followed by postdoctoral research supervised by Michael Levitt at Stanford University School of Medicine.

Brenner has a commitment to supporting open science and development of a diverse, inclusive scientific community. He is currently a member of the ClinGen Sequence Variant Interpretation Working Group, a director of the Human Genome Variation Society, and a member of the HUGO Pathogenicity Committee. He was founding chair of the Computational Biology graduate program at Berkeley, a founding editor of *PLoS Computational Biology*, a founding chair of the Global Alliance for Genomics and Health's Data Working Group Variant Annotation Task Team, and was a founding director of the Open Bioinformatics Foundation, His recognitions including being a Miller Professor, a

Sloan Research Fellow, a Searle Scholar, an AAAS Fellow, an ISCB Fellow, and a recipient of ISCB's Overton Prize.

Jeffrey P. Brosco, M.D., Ph.D., is a historian and pediatrician who serves as the Director for the Division of Services for Children with Special Health Needs at the Health Resources and Services Administration's Maternal and Child Health Bureau. As DSCSHN Director, Dr. Brosco leads a team tasked with ensuring that every child in the U.S. receive the medical care and family support they need to play, go to school, and grow up to be healthy and productive adults.

Dr. Brosco also continues to teach and practice general pediatrics and developmental-behavioral pediatrics at the University of Miami Miller School of Medicine. He directed the MCHB-funded Leadership Education in Neurodevelopmental Disabilities (LEND) training program at the Mailman Center for Child Development from 2010 to 2022. He has also served as Director, Population Health Ethics, at UM's Institute for Bioethics and Health Policy, and as Associate Chair, Population Health, for UM's Department of Pediatrics. Dr. Brosco's research focuses on history, policy, and ethics regarding child health and disability; he has authored 50 peer review publications and over 90 other publications. For over two decades, Dr. Brosco has held a series of leadership positions for the

Florida Department of Health's Children's Medical Services (CMS), which seeks to improve the health of children with special health care needs. From 2017-19 he was Florida's Deputy Secretary of Health for CMS, and stepped down in 2022 after 4 years as the state's Title V Director for CSHCN. Dr. Brosco has also been active in national health policy groups, such as the Advisory Committee on Heritable Disorders in Newborns and Children and the National Workgroup on Standards for Systems of Care for Children and Youth with Special Health Care Needs. In 2019 he was awarded a Maternal and Child Health Bureau Director's Award for noteworthy national level contributions to the health of infants, mothers, children, adolescents, and children with special health care needs.

Ellen Wright Clayton, M.D., J.D., is the Craig-Weaver Professor of Pediatrics, Professor of Health Policy, and Co-Founder of the Center for Biomedical Ethics and Society at Vanderbilt University Medical Center (VUMC) and Professor of Law at the Vanderbilt Law School. Her work is truly transdisciplinary, combining empirical, normative, and legal analytic methods to address real-world challenges. An internationally respected scholar, she has focused for many years on ethical, legal, and social issues presented by conducting research in genetics and genomics and the impact of translating these advances in clinical care and the broader society as well as issues related to women's and children's health and wellbeing. She has been a member of Tennessee's state genetics committee since the early 1990s and has written numerous articles about genetic testing of children. She is currently co-PI of VUMC's highly transdisciplinary Center of

Excellence in ELSI Research on genomic privacy and identity, GetPreCiSe, as well as being deeply engaged in the eMERGE consortium since its inception and working as part of both AIM-AHEAD and Bridge2AI to ensure the ethical and legal development and deployment of machine learning and artificial intelligence. She is an elected member of the National Academy of Medicine, where she has served on 11 consensus committees and the Executive Committee and is currently Co-Chair of Report Review.

Faith Fletcher, Ph.D., M.A., is an Assistant Professor in the Center for Medical Ethics and Health Policy at Baylor College of Medicine, a senior advisor to The Hastings Center, and a Hastings Center fellow. Drawing from diverse disciplines and methodologies from bioethics, public health, and behavioral science, her empirical research investigates pressing health concerns and inequities facing traditionally marginalized populations. In her scholarship, she engages issues relating to reproductive health and autonomy, informed decision-making, structural stigma, and trustworthiness in both research and healthcare settings.

Her K01 Award, funded through the NIH/ National Human Genomic Research Institute uses a stakeholder engagement approach to develop ethical practices and guidelines for engaging Deep South residents in genomics research. Dr. Fletcher was recently named to the Greenwall Faculty Scholars Program in Bioethics Class of 2026. Her work will examine the role of bioethicists in advancing maternal health equity among Black women in the US.

In collaboration with an antiracism task force, Dr. Fletcher is a co-editor of the Hastings Center Special report entitled, “A Critical Moment in Bioethics: Reckoning with Anti-Black Racism Through Intergenerational Dialogue”. 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.

Aaron Goldenberg, Ph.D., is a Professor and Vice-Chair in the Department of Bioethics at Case Western Reserve University. He is also Director for the CWRU Bioethics Center for Community Health and Genomic Equity. Dr. Goldenberg has a background in bioethics, health behavior/health education, public health ethics, and public health genetics. He has focused his work on the ethical, legal, and social issues associated with the integration of new genomic technologies into research, clinical and public health settings. Dr. Goldenberg's research program has been grounded by a number of major project areas, including: 1) the ethical and social implications of storing and using of biological specimens and data for future research; 2) implications of genetics and gene-environment interactions for racial/ethnic minorities and other communities experiencing health disparities; and 3) ethical implications of expanding newborn

screening programs. He is currently Co-Chair the NBSTRN Bioethics and Legal Workgroup, and with this group, led a recent effort to develop a set ELSI questions that can be integrated into NBS Pilots to help assess the potential benefits and harms of adding new conditions to NBS panels. He is also Co-Chiar for the Legal and Legislative Issues in Newborn Screening Workgroup for the Association of Public Health Laboratories.

Crystal Grant, Ph.D., is a Technologist in Residence at the Federal Trade Commission. She was formerly a Technology Fellow with the American Civil Liberties Union's (ACLU) Speech, Privacy, and Technology Project where she worked on issues in genetic privacy and surveillance as well as racial and ethnic bias in algorithmic decision-making systems and in AI/ML. Dr. Grant is a graduate of Emory University's Laney Graduate School where she was a National Science Foundation Graduate Research Fellow and her research focused on applying bioinformatics techniques to better understand human aging and disease.

After obtaining her Ph.D. in Genetics and Molecular Biology from Emory, Dr. Grant served as a Mirzayan Graduate Science and Technology Policy Fellow at the National Academy of Sciences where she contributed to a consensus report on policy recommendations to increase the number of women of color working in tech. She next served as a Legislative Fellow in Senator Elizabeth Warren's office through the TechCongress Congressional Innovation Scholars program working on a portfolio that spanned both health and technology policy. Dr. Grant earned her undergraduate degree in biological sciences at Cornell University.

Robert Green, M.D., M.P.H., is a board-certified medical geneticist and Professor of Medicine at Harvard Medical School who directs the Genomes2People Research Program at Mass General Brigham, Ariadne Labs and the Broad Institute.

Dr. Green's empirical research and policy development is accelerating the implementation of genomic and precision medicine. His work has established the safety and feasibility of disclosing various forms of genetic risk information, assessed the impact of whole genome sequencing in primary care, created the concept of aggregate penetrance of genomic variants in a prospective population cohort and provided early data on the clinical utility and cost-effectiveness of genomic sequencing in healthy adults (the MedSeq Project), and in active duty military personnel (the MilSeq Project). Most recently Dr. Green is leading the first empirical trial of comprehensive sequencing in healthy newborn infants, the NIH-funded BabySeq Project.

Dr. Green established the world's first academically-affiliated, family-oriented Preventive Genomics Clinic in Boston and led policy development for returning genomic information to research participants within the Global Alliance for Genomics and Health, the Verily-

Google Baseline Project and All of Us Research Program. In 2017, he co-founded the nationwide telegenetics company Genome Medical.

Zhanzhi "Mike" Hu, Ph.D., is a parent of two children with a rare disorder. He is an ardent advocate for newborn screening and works tirelessly to bridge the gap between advanced technologies and the critical need of expansion of the newborn screening public health program. Mike is the cofounder of Project GUARDIAN, a nonprofit organization with the mission of advancing genomics based newborn screening. He also serves as a member of the Steering Committee and the chair of the Researcher Needs Workgroup at the Newborn Screening Translational Research Network (NBSTRN). Additionally, he is an Adj. Associate Research Scientist at Columbia University where he conducts newborn screening related research projects. Previously, Mike was the Associate Director of Product Development at Veracyte and had led the development team to successfully launch multiple diagnostic products during his tenure. Before joining Veracyte, Mike co-founded a precision medicine startup to pioneer advanced reproductive healthcare by genome scale carrier screening. Prior to that, Mike had led development teams at Complete Genomics and Affymetrix to develop next-generation sequencing technologies and products. Mike received his Ph.D. degree in Molecular Genetics and Microbiology from the University of Texas at Austin, and the B.S. degree in Cell Biology and Genetics from Peking University.

Cheedy Jaja, Ph.D., M.P.H., M.S.N., PMHNP-BC, APRN, FAAN, is an Associate Professor at the University of South Florida. His interest in social justice, health disparities, and improving health outcomes in historically marginalized and vulnerable populations such as those with sickle cell disease (SCD) drives his research, clinical, and advocacy initiatives. As a clinician and health science researcher, he noticed with dismay the unsettlingly high under-five mortality rates associated with SCD in sub-Saharan Africa even though standard public-health care packages, including pre-test counseling and carriers for sickle cell hemoglobinopathy, are available and in use in middle-and high-income countries. His professional partnerships and research initiatives in the West African nation of Sierra Leone are efforts to reduce this global health disparity gap. Additionally, Dr. Jaja is one of only a few nurse scientists prepared to use pharmacogenetics strategies in pharmacotherapy for SCD. In his pioneering research, he has made significant discoveries that are likely to make an impact on the way SCD pain could be treated. He has received national recognition for his research contributions as evidenced by being selected as a 2016 NIH Future Research Leader, a 2018 Fulbright Program Scholar, and a Fellow of the American Academy of Nursing in 2020.

Terri Klein, M.P.A., CNPM, is the President and CEO at the National MPS Society in Durham, North Carolina. Her expertise is in operations and management, contract negotiation, and multi-tier fund development. Her portfolio includes over 36 million dollars in secured corporate prospects, and nonprofit gifts. Terri began her nonprofit

career in 2004, and became the first Executive Director of the ISMRD in 2005. Terri works in partnership with her Board of Directors to implement crucial patient programs that include research and family support. The Society has funded over $ 18 million in research and over $4 million in family support programs. She has worked to increase the membership of the Society by over 310% in the last three years, and now provides services to over 2600 households nationwide. Crucial areas of focus are diseases with no treatment, bridging bench and translational research to clinical application, key stakeholder engagement, newborn screening, RUSP nominations for all MPS syndromes, patient secured data, and access to therapies. In addition, she administers advocacy in efforts within the NIH, NCATS, NINDS, FDA, RDCRN, HRSA, and the ACHDNC.

Terri is past Co-Chair of the International MPS Network (IMPSN), which is a nonprofit in Canada. The IMPSN will further drive humanitarian efforts for MPS around the world. Initiatives include: newborn screening, access to treatments; unmet needs of underserved countries, and reimbursement policies.

She is a wife and mother of four, including Jennifer (31), her youngest that suffers from Mucolipidosis III. Terri spends volunteer time working with the local ALS association, and enjoys spending time with her family. Terri's publications have been in the areas of newborn screening and patient advocacy.

Sylvia Mann, M.S., CGC, is a board certified genetic counselor who has worked in the State of Hawaii Department of Health for 30 years. Currently, she is the supervisor of the Genomics Section which includes the genetics, newborn screening, and birth defects programs. Ms. Mann is also the Project Director for the Health Resources and Services Administration funded Western States Regional Genetics Network (WSRGN). The WSRGN includes Alaska, California, Guam, Hawaii, Idaho, Oregon, and Washington and seeks to improve genetic services, information, and education for underserved families.

To improve access and education, Ms. Mann has been working on increasing the use of telegenetics since 2004. She also worked to develop the Minority Genetic Professionals Network which serves to increase the number of racial and ethnic minority students entering genetics professions and supporting practicing minority genetics professionals excel in their career.

Ms. Mann has served on many national committees including the U.S. Health and Human Services Secretary's Advisory Committee on Genetics, Health and Society. She is also currently a board member of the Accreditation Counsel for Genetic Counseling. Ms. Mann received her Bachelor of Science degree from the University of British Columbia and her Master of Science degree in human genetics and genetic counseling from Sarah Lawrence College.

Holly L. Peay, Ph.D., M.S., is a Senior Research Scientist at RTI International. She received her B.A. from University of Virginia in Biomedical Ethics, her M.S. in Genetic Counseling from University of South Carolina School of Medicine, and her Ph.D. in Medicine from Leiden University in the Netherlands. Dr. Peay's research is primarily focused on the ethical integration of new technologies into research, clinical, and public health settings. Dr. Peay is the Director of RTI's Early Check research program, which currently returns research on approximately 450 newborns per month. By mid-2023, RTI will launch the Early Check genome sequencing study to return results for two large panels of single-gene conditions and for genetic risk for type 1 diabetes. In addition to expertise in genomics, Dr. Peay has expertise using qualitative and quantitative social science techniques to explore preferences, attitudes, risk perception, and decision making. Her experience includes the development of approaches and instruments for lower-literacy and historically underrepresented populations. Dr. Peay uses empirical evidence and bioethics scholarship to inform the development of return of results protocols, decision support products, and shared decision-making tools. Dr. Peay's expertise with ethical, legal and social implications of new technologies is reflected in her roles in the National Human Genome Research Institute Genomics and Society Working Group; the All of Us ELSI Brain Trust; and the International Society for Psychiatric Genetics Ethics, Position & Policy Committee.

Cynthia Powell, M.D., is a Professor of Pediatrics and Genetics at the University of North Carolina at Chapel Hill School of Medicine, where she sees patients, teaches students, residents and fellows, and participates in research. She is the program director of the UNC Hospitals Medical Genetics and Genomics Residency Program. She is immediate past Chair of the U.S. federal Advisory Committee on Heritable Disorders in Newborns and Children (HRSA committee that makes recommendations to the Secretary of the Department of Health and Human Services about conditions to be added to the Recommended Uniform Screening Panel). Her research interests include newborn screening, genomics, birth defects and genetic syndromes. She led the North Carolina Newborn Exome Sequencing for Universal Screening (NC NEXUS) project, a five-year project funded by NIH investigating the utility of next generation sequencing in newborns and is currently the site PI for the Early Check Project in North Carolina, a voluntary expanded newborn screening program.

Nathaly M. Sweeney, M.D., M.P.H., M.S., FAAP, is an assistant professor of pediatrics in the neonatology division at the University of California San Diego (UCSD)/ Rady Children's Hospital San Diego (RCHSD) and an investigator with the Rady Children's Institute for Genomic Medicine (RCIGM).

Dr. Sweeney is board certified in general pediatrics, pediatric cardiology and neonatal-perinatal medicine. She earned her undergraduate degree from Springfield College, MA

and her graduate degrees from the University of Texas at El Paso, TX and the Johns Hopkins University- Bloomberg School of Public Health in Baltimore, MD.

She obtained her medical degree from Columbia University College of Physicians and Surgeons, NY, NY and completed her pediatric residency at Johns Hopkins Hospital in Baltimore, MD.

Dr. Sweeney completed her pediatric cardiology fellowship at Lucile Packard Children's Hospital-Stanford University, Stanford, CA and her neonatal-perinatal medicine fellowship at UCSD.

Dr. Sweeney is interested in the application of individualized medicine in the care of children with suspected genetic disease with a focus on the optimization of short and long-term outcomes in children with congenital heart disease.

Her research focuses on identifying genotypic differences via genomic sequencing that may explain the observed phenotypes with the goal of offering prompt, directed, accurate, personalized care to children in the intensive care unit and beyond.

David Veenstra, Pharm.D., Ph.D., is a Professor in the Department of Pharmacy and the Comparative Health Outcomes, Policy & Economics (CHOICE) Institute. He graduated from the University of California San Francisco with doctoral degrees in clinical pharmacy and computational chemistry. He conducted his postdoctoral training in health economics and outcomes research with the University of Washington, including a one-year externship with Roche Global Pharmacoeconomics.

Dr. Veenstra's primary research interests are the clinical, economic, and policy implications of using genomic information in healthcare. His major research projects include evaluation of the cost effectiveness of population-level genomic screening, pharmacogenomics in diverse populations, decision modeling techniques to assess evidence thresholds, and stakeholder preferences for precision medicine.

Dr. Veenstra's research has been funded through grants from the National Human Genome Research Institute, Centers for Disease Control, National Cancer Institute, and the National Institute for General Medical Sciences.

Dr. Veenstra has worked extensively with organizations such as the Academy of Managed Care Pharmacy (AMCP) and the Institute for Clinical and Economic Review (ICER) to further the practical application of cost-effectiveness analysis in managed care decision making.

Dr. Veenstra teaches courses in pharmacoeconomics and managed care and is an author

or co-author of five book chapters and over 180 peer-reviewed publications.

Teonna Woolford was born and raised in Baltimore, Maryland. Since birth, she has overcome many battles in her fight against Sickle Cell Anemia, SS; the most severe form of a hereditary blood disorder that causes life-threatening complications and affects millions of people of color around the world. A true warrior at heart, Teonna has recovered from numerous challenges including, but not limited to, excruciating pain crises, bilateral hip replacements, and a failed bone marrow transplant that resulted in the side effect she feared most, infertility.

When searching for both educational and financial resources for fertility preservation, Teonna was taken aback by the lack of information related to women with Sickle Cell. For far too long, the Sickle Cell Community has faced tremendous disparities and have had to prioritize survival over quality of life, allowing other areas of importance, such as reproductive health, to be overlooked. To combat this, Teonna, alongside two physicians devoted to the cause; John Hopkin's Dr. Lydia Pecker and CHOP's Dr. Kim Smith-Whitley, founded the non-profit organization Sickle Cell Reproductive Health Education Directive (SC RED). SC RED is a collective of Sickle Cell warriors, providers, caregivers, and other key thought leaders advocating for high-quality sexual and reproductive care through awareness, education, advocacy, and various levels of support.

With an intimate understanding of the realities of those impacted by Sickle Cell Disease, Teonna has served on several working groups for the National Institutes of Health (NIH), and the National Heart, Lung, and Blood Institute (NHLBI). Teonna has also published with the American Society of Hematology (ASH). She has been successful carrying her mission and message to schools, churches, and The White House; bringing her face-to-face with former First Lady, Michelle Obama, Dr. Ben Carson, and the late civil rights activist and humanitarian, Congressman John Lewis. In October 2021, Teonna made history as the first patient to give the Charles F. Whitten Memorial Lecture alongside her mentor, Dr. Kim Smith-Whitley.

While Sickle Cell has been a large part of Teonna's life, she does her best not to let it define who she is. In her spare time, she enjoys reading, writing, cooking (therefore eating), shopping, watching movies, keeping up with current events, and most of all, spending time with her family. She has always been active in her church and seeks to put God first in her life. Like many in the Sickle Cell community, Teonna is determined to not only live but to thrive.

NATIONAL ACADEMIES Sciences Engineering Medicine

Next-Generation Screening – The Promise and Perils of DNA Sequencing of Newborns at Birth

June 7, 2023

SPEAKER GUIDANCE: CONTEXT AND QUESTIONS

Following the Genomics Roundtable's strategic plan development in 2020, the Innovation working group has been interested in identifying the competing barriers and facilitators of innovation for genomics-based diagnostics, risk assessment tools, and therapies as well as leveraging opportunities to learn from and promote innovative approaches that can accelerate commercialization and integration to drive impact of genomics on precision health. The goals of this public workshop are to (1) examine the known and expected benefits, and potential harms, of the widespread utilization of newborn DNA sequencing, (2) explore the ethical and data security and ownership issues associated with DNA sequencing of newborns at birth, and (3) address issues of next-generation newborn screening equity in the United States.

Session I: KEYNOTE

Questions to frame the keynote:

1. What distinctions are important when thinking about newborn screening versus newborn sequencing?
2. What is the current landscape of newborn sequencing? What key implementation issues do you foresee as newborn sequencing becomes more prevalent?
3. Why is it important to focus on newborns rather than adult population screening or sequencing?

Key Questions for discussants:

1. What is the current landscape of newborn sequencing? What key implementation issues do you foresee as newborn sequencing becomes more prevalent?
2. What are the key ethical issues to consider as sequencing in healthy newborns is implemented in clinical care?
3. Why is it important to focus on newborns rather than adult population screening or sequencing?

Session II: Lessons Learned from Newborn Genomic Testing and Screening

Questions to frame speakers' talks:

1. Based on your experience, what are you learning about implementing whole genome sequencing in healthy newborns?
2. How was newborn sequencing conducted in your program?
3. Do we have appropriate and well-accepted definitions in place to ensure there is clarity about the assays, test definitions and findings that are being delivered to newborn parents?

Key Questions for Speakers:

1. See list of questions:
    - What is informative for patient care (genes, conditions assessed)?
    - What information may provide value over the lifespan and where is there uncertainty or potential harms?
    - What are considerations around equity in clinical utility?
    - How are families dealing with the implications of having this information?
2. What experiences have you had surrounding interpreting test results? What are the opportunities and challenges?
3. What challenges did you encounter with return of results? How can the process be improved in the future? Have you examined the impacts on families of the results 1-3 years out?

Session III: Implementing Newborn Sequencing at Scale – Health System Challenges & Opportunities

Questions to frame speakers' talks:

1. How can health systems prepare to manage the implementation of genome sequencing in healthy newborns?
2. What needs to change in order to build the workforce needed to address the complexities of newborn sequencing? Are there models that we can be learning from (e.g., other complex systems)?

Key Questions for Speakers:

1. What challenges and opportunities do you foresee with long-term follow-up of patients sequenced at birth? Both for those who are not diagnosed with a condition, but their genomes will be used to inform care throughout their life course, and those whose who receive a diagnosis.
2. What are the data management and privacy issues specific to newborn sequencing? What infrastructure needs to be in place to manage these issues?

Session IV: Deploying Newborn Sequencing Responsibly and Equitably

Questions to frame speakers' talks:

1. What is needed to make newborn sequencing available equitably across communities in the U.S.? What defines readiness for system wide deployment of DNA sequencing in healthy newborns?
2. Would sequencing of health newborns best be done through U.S. public health based newborn screening and is it feasible?
3. How does the policy landscape affect access (to testing, diagnosis, and treatment) and payment across socioeconomic groups and geography?

Key Questions for Speakers:

1. What are some of the major barriers to access both to sequencing and follow up care? How can they be addressed to decrease inequities?
2. What are some best practices for education and engagement of patients and communities? How can we best engage communities that have been historically excluded from clinical research?
3. Given concerns about ableism in genomics, how can the field move forward to be equitable and supportive of individuals and families across a wide range of ability?

Session V: How Will Newborn Sequencing Change the Trajectory of Precision Health?

Questions to frame speakers' talks:

1. How can genomic information gathered at birth be used in health care across an individual's and their family's lifespan?
2. How will public trust be established and maintained?
3. How might the use of these platforms address or exacerbate health disparities?
4. What are some of the legal and ethical issues that must be addressed as healthy newborn's genomes begin to be sequenced?
5. What are the biggest data privacy and security issues that need to be considered?

Key Questions for Speakers:

1. How might precision health be pushed forward through access to a genome from birth?
2. How could health disparities be affected by DNA sequencing health newborns?
3. Given concerns about ableism in genomics, how can the field move forward to be equitable and supportive of individuals and families across a wide range of ability?

Session VI: Final Reflections

Key Questions for Speakers:

1. As more programs are developed to sequence newborn DNA, what are some ways that equity can be considered from the early stages of implementation?
2. What does a world with both clinical newborn sequencing and public health newborn screening look like? How might these systems work together or separately?
3. How can we ensure that proper data privacy and security policies are put in place to protect individuals?
4. What are the implications of having newborn sequencing at a population scale? How can policy be shaped to ensure support, proper implementation, and benefits for the whole population through newborn sequencing?

NATIONAL ACADEMIES Sciences Engineering Medicine

PREVENTING DISCRIMINATION, HARASSMENT, AND BULLYING:
POLICY FOR PARTICIPANTS IN NASEM ACTIVITIES

The National Academies of Sciences, Engineering, and Medicine (NASEM) are committed to the principles of diversity, inclusion, integrity, civility, and respect in all of our activities. We look to you to be a partner in this commitment by helping us to maintain a professional and cordial environment. **All forms of discrimination, harassment, and bullying are prohibited in any NASEM activity.** This policy applies to all participants in all settings and locations in which NASEM work and activities are conducted, including committee meetings, workshops, conferences, and other work and social functions where employees, volunteers, sponsors, vendors, or guests are present.

Discrimination is prejudicial treatment of individuals or groups of people based on their race, ethnicity, color, national origin, sex, sexual orientation, gender identity, age, religion, disability, veteran status, or any other characteristic protected by applicable laws.

Sexual harassment is unwelcome sexual advances, requests for sexual favors, and other verbal or physical conduct of a sexual nature that creates an intimidating, hostile, or offensive environment.

Other types of harassment include any verbal or physical conduct directed at individuals or groups of people because of their race, ethnicity, color, national origin, sex, sexual orientation, gender identity, age, religion, disability, veteran status, or any other characteristic protected by applicable laws, that creates an intimidating, hostile, or offensive environment.

Bullying is unwelcome, aggressive behavior involving the use of influence, threat, intimidation, or coercion to dominate others in the professional environment.

REPORTING AND RESOLUTION

Any violation of this policy should be reported. If you experience or witness discrimination, harassment, or bullying, you are encouraged to make your unease or disapproval known to the individual at the time the incident occurs, if you are comfortable doing so. You are also urged to report any incident by:

- Filing a complaint with the Office of Human Resources at 202-334-3400 or hrservicecenter@nas.edu, or
- Reporting the incident to an employee involved in the activity in which the member or volunteer is participating, who will then file a complaint with the Office of Human Resources.

Complaints should be filed as soon as possible after an incident. To ensure the prompt and thorough investigation of the complaint, the complainant should provide as much information as is possible, such as names, dates, locations, and steps taken. The Office of Human Resources will investigate the alleged violation in consultation with the Office of the General Counsel.

If an investigation results in a finding that an individual has committed a violation, NASEM will take the actions necessary to protect those involved in its activities from any future discrimination, harassment, or bullying, including in appropriate circumstances **the removal of an individual from current NASEM activities and a ban on participation in future activities**.

CONFIDENTIALITY

Information contained in a complaint is kept confidential, and information is revealed only on a need-to-know basis. NASEM will not retaliate or tolerate retaliation against anyone who makes a good faith report of discrimination, harassment, or bullying.

Updated December 2, 2021

BACKGROUND MATERIALS

Links to Additional Resources

SESSION I: OPENING REMARKS & KEYNOTE

- The Hastings Center. The Ethics of Sequencing Newborns: Reflections and Recommendations. https://www.thehastingscenter.org/publications-resources/special-reports-2/ethics-sequencing-newborns-reflections-recommendations/
- The BabySeq Project publications: https://www.genomes2people.org/publications/?_projects=3567
- Grant. 2023. Widespread Newborn DNA Sequencing Will Worsen Risks to Genetic Privacy https://www.aclu.org/news/privacy-technology/widespread-newborn-dna-sequencing-will-worsen-risks-to-genetic-privacy
- Illumina Genomics Forum. 2022. Head start: The promise of universal newborn sequencing. https://www.youtube.com/watch?v=ZFSEzlCpuz0
- Powell, C. 2018. What Genomic Sequencing Can Offer Universal Newborn Screening Programs. https://pubmed.ncbi.nlm.nih.gov/30133725/

SESSION II: LESSONS LEARNED FROM NEWBORN GENOMIC TESTING AND SCREENING

- Roman et al. 2020. Genomic Sequencing for Newborn Screening: Results of the NC NEXUS Project. https://pubmed.ncbi.nlm.nih.gov/32853555/
    - NC Nexus - https://www.med.unc.edu/genetics/berglab/research/past-projects/nc-nexus-project/
- UCSF NSIGHT Project: Newborn Sequencing, NBSeq - https://www.genome.gov/Multimedia/Slides/NIH_NSIGHT_2015/03_Puck.pdf
- BeginNGS - https://radygenomics.org/begin-ngs-newborn-sequencing/
- GUARDIAN Study - https://guardian-study.org/overview/
- The BabySeq Project - https://www.genomes2people.org/research/babyseq/
- Gold et al. 2023. Perspectives of rare disease experts on newborn genome sequencing. https://jamanetwork.com/journals/jamanetworkopen/fullarticle/2804586
- Pereira et al. 2023. Parents' decision-making regarding whether to receive adult-onset only genetic findings for their children: Findings from the BabySeq Project. https://www.sciencedirect.com/science/article/abs/pii/S1098360022010693?via%3Dihub
- Green et al. 2023. Actionability of unanticipated monogenic disease risks in newborn genomic screening: Findings from the BabySeq Project. https://www.genomes2people.org/wp-content/uploads/2023/05/20230605_BabySeq_AJHG_Green_UnanticipatedMonogenic.pdf

SESSION III: IMPLEMENTING NEWBORN SEQUENCING AT SCALE - HEALTH SYSTEM CHALLENGES & OPPORTUNITIES

- Berg et al. 2011. Deploying whole genome sequencing in clinical practice and public health: Meeting the challenge one bin at a time. https://www.nature.com/articles/gim9201182
- Bick et al. 2022. Newborn Screening by Genomic Sequencing: Opportunities and Challenges. https://www.mdpi.com/2409-515X/8/3/40

SESSION IV: DEPLOYING NEWBORN SEQUENCING RESPONSIBLY AND EQUITABLY

- Fletcher et al. A Critical Moment in Bioethics: A Reckoning with Anti-Black Racism through Intergenerational Dialogue. https://onlinelibrary.wiley.com/toc/1552146x/2022/52/S1
- Goldenberg. 2019. Considering Equity in Assessing Familial Benefit From the Return of Genomic Research Results. https://publications.aap.org/pediatrics/article/144/6/e20193111/37939/Considering-Equity-in-Assessing-Familial-Benefit?autologincheck=redirected
- Sickle Cell reproductive Health Education Directive (SCRED) - https://sicklecellred.org/
- Gold et al. 2023. Perspectives of Rare Disease Experts on Newborn Genome Sequencing. https://jamanetwork.com/journals/jamanetworkopen/fullarticle/2804586

SESSION V: HOW WILL NEWBORN SEQUENCING CHANGE THE TRAJECTORY OF PRECISION HEALTH?

- Genomics England - https://www.genomicsengland.co.uk/initiatives/newborns
- Abul-Husn et al. 2021. Implementing genomic screening in diverse populations. https://genomemedicine.biomedcentral.com/articles/10.1186/s13073-021-00832-y
- Abul-Husn et al. 2023. Molecular diagnostic yield of genome sequencing versus targeted gene panel testing in racially and ethically diverse pediatric patients. https://www.gimjournal.org/article/S1098-3600(23)00893-6/pdf
- Berg et al. 2017. Newborn Sequencing in Genomics Medicine and Public Health. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5260149/
- Brosco et al. 2015. Universal State Newborn Screening Programs Can Reduce Health Disparities. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4528613/
- Chung et al. 2022. Newborn screening for neurodevelopmental diseases: Are we there yet? https://onlinelibrary.wiley.com/doi/10.1002/ajmg.c.31988