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

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Cofounder
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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