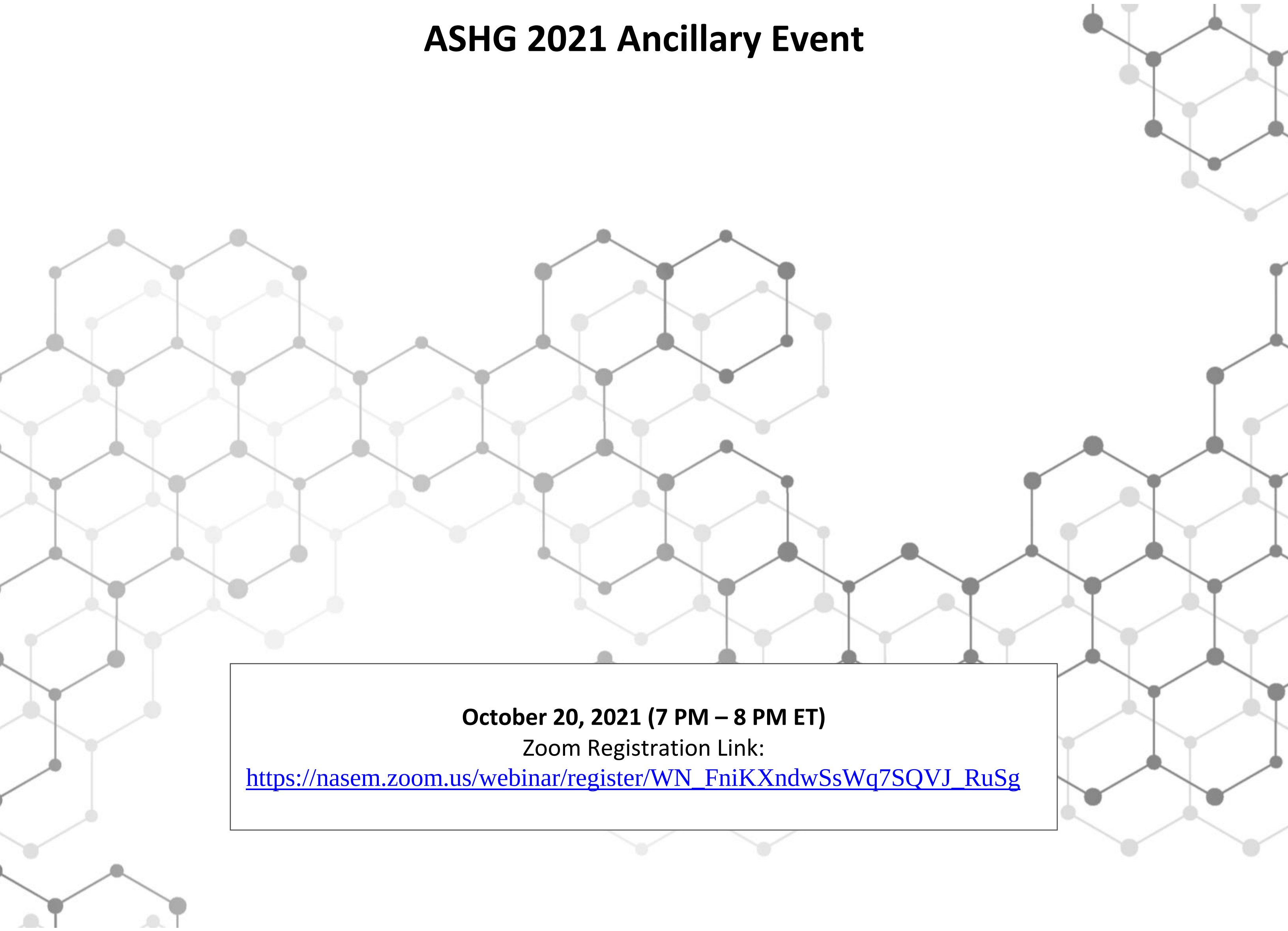
The National Academies of
SCIENCES • ENGINEERING • MEDICINE

Board on Health Sciences Policy

Realizing the Full Potential of Health for All Through Genomics and Precision Health:

A discussion with the National Academies Genomics Roundtable

ASHG 2021 Ancillary Event



October 20, 2021 (7 PM – 8 PM ET)
Zoom Registration Link:
https://nasem.zoom.us/webinar/register/WN_FniKXndwSsWq7SQVJ_RuSg

Roundtable on **GENOMICS** and **PRECISION HEALTH**

The National Academies of
SCIENCES • ENGINEERING • MEDICINE

Board on Health Sciences Policy

Roundtable on Genomics and Precision Health

ASHG 2021 Ancillary Event

October 20, 2021

Table of Contents

Roundtable on GENOMICS and PRECISION HEALTH

EVENT INFORMATION

The National Academies of
SCIENCES • ENGINEERING • MEDICINE

Board on Health Sciences Policy

Realizing the Full Potential of Health for All Through Genomics and Precision Health:

A discussion with the National Academies Genomics Roundtable

An Ancillary Event to ASHG's Annual Meeting

A Virtual Meeting
October 20, 2021
7:00 – 8:00 PM ET
Zoom Webinar Registration Link:
https://nasem.zoom.us/webinar/register/WN_FniKXndwSsWq7SQVJ_RuSg

AGENDA:

7:00 pm ET **Welcoming Remarks**

Geoffrey Ginsburg, M.D., Ph.D.
Director, Duke Center for Applied Genomics & Precision Medicine
Professor, Medicine, Pathology, and Biomedical Engineering
Duke University Medical Center

7:05 pm **Accelerating Genomic Advances for All**

Vence Bonham, Jr, J.D.
Acting Deputy Director
National Human Genome Research Institute
National Institutes of Health

7:20 pm **How Equity Can Enable Adoption and Innovation**

Mira Irons, M.D.
President and CEO
College of Physicians of Philadelphia

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

Roundtable on **GENOMICS** and **PRECISION HEALTH**

7:30 pm **Getting Involved in the Roundtable's Work**

Sarah Beachy, Ph.D.
Director, Roundtable on Genomics and Precision Health
Board on Health Sciences Policy
The National Academies of Sciences, Engineering, and Medicine

7:35 pm **Questions and Open Dialogue with the Community**
Moderated by: Geoffrey Ginsburg

7:55 pm **Closing Comments**

8:00 pm **Meeting Adjourns**

The National Academies of
SCIENCES · ENGINEERING · MEDICINE

Board on Health Sciences Policy
Roundtable on Genomics and Precision Health

Realizing the Full Potential of Health for All Through Genomics and Precision Health:

A discussion with the National Academies Genomics Roundtable

October 20, 2021

Speaker Biographies

Sarah Beachy, Ph.D., is a Senior Program Officer with the National Academies of Sciences, Engineering, and Medicine in Washington, DC. In this capacity, Sarah serves as Director of the Roundtable on Genomics and Precision Health and the Forum on Regenerative Medicine. Each standing convening activity brings together leaders from a variety of sectors, including government, industry, academia, and patient advocacy organizations to discuss key challenges and emerging scientific and policy issues related to the translation of scientific discoveries into clinical use. In these roles and in leading other projects, Sarah has facilitated impactful public workshops and discussions such as Improving Diversity of the Genomics Workforce; Exploring the Current Landscape of Consumer Genomics; Understanding Disparities in Access to Genomic Medicine; Applying an Implementation Science Approach to Genomic Medicine; Changing the Culture of Data Sharing and Management; and An Examination of Emerging Bioethical Issues in Biomedical Research, among others. Prior to her time at the National Academies, Sarah completed a AAAS Science and Technology Policy Fellowship in diplomacy at the U.S. Department of State, working closely with the Office of the Science and Technology Adviser to the Secretary. Sarah was selected as a Mirzayan Science and Technology Policy Fellow and worked with the Board on Health Sciences Policy at the National Academies in 2011. Prior to moving into science policy, Sarah was a postdoctoral fellow in the Genetics Branch at the National Cancer Institute, where she generated and characterized transgenic mouse models of leukemia and lymphoma. Sarah earned her Ph.D. in biophysics from the Roswell Park Cancer Institute Graduate Division at the University at Buffalo.

Vence Bonham, Jr., J.D., received his bachelor of arts from James Madison College at Michigan State University and his juris doctor degree from the Moritz College of Law at the Ohio State University. Mr. Bonham was a fellow in the American Association of Medical Colleges Health Services Research Fellowship Program. Mr. Bonham was a faculty member at Michigan State University in the Colleges of Medicine and Law. Since 2003, Mr. Bonham has served as an associate investigator in the National Human Genome Research Institute (NHGRI) within the Division of Intramural Research's Social and Behavioral Research Branch. He leads the Health Disparities Genomics Unit, which conducts research that evaluates approaches to integrating new genomic knowledge and precision medicine into clinical settings without exacerbating inequities in healthcare delivery. His research focuses primarily on the social influences of new genomic knowledge, particularly in communities of color. He studies how genomics influences the use of the constructs of race and ethnicity in biomedical research and clinical care and the role of genomics in health inequities. The Bonham group has expanded to study sickle cell disease, a condition with a significant health disparity impact both in the United States and globally. Mr. Bonham also serves as the senior advisor to the NHGRI director on genomics and health disparities. This role complements Mr. Bonham's research work as it enables him to ask conceptually based research questions grounded in the science of health disparities and genomics. From 2011 until 2015, Mr. Bonham was the project leader and co-curator for the NHGRI/Smithsonian exhibition “Genome: Unlocking Life’s Code.”

Geoffrey Ginsburg, M.D., Ph.D., is the founding director for the Center for Applied Genomics & Precision Medicine at the Duke University Medical Center and for MEDx, a partnership between the Schools of Medicine and Engineering to spark and translate innovation. His research addresses the challenges for translating genomic and digital information into medical practice and the integration of precision medicine into healthcare. In 2017 he received Duke’s Translational Research Mentorship Award and is a finalist in the NIH/BARDA Antimicrobial Resistance Prize. He is a member of the Advisory Council to the Director of NIH and is co-chair of the National Academies Roundtable on Genomics and Precision Health and is founder and president of the Global Genomic Medicine Collaborative, a not for profit organization aimed creating international partnerships to advance the implementation of precision medicine. He has recently served as a member of the Board of External Experts for the NHLBI, the advisory council for the National Center for Accelerating Translational Science, the chair of the review for Genome Canada’s Large Scale Applied Research Competition in Genomics and Precision Medicine, and the World Economic Forum’s Global Agenda Council on the Future of the Health Sector. He is a founder of Predigen Inc and MeTree&You Inc. He was previously Vice President of Molecular Medicine at Millennium Pharmaceuticals, Inc and a faculty member at Harvard Medical School.

Mira Bjelotomich Irons, M.D., is the President and CEO and the Thomas W. Langfitt Chair of the College of Physicians of Philadelphia. Prior to coming to the College of Physicians as its first female CEO, she held the position of Chief Health and Science Officer; Group Vice President, Health, Science and Ethics, at the American Medical Association (AMA). In that role she provided intellectual leadership for AMA initiatives at the interfaces of practice, science, evidence, policy, ethics, and technology. Dr. Irons is a board-certified physician in both Medical Genetics and Pediatrics and has an impressive breadth and depth of experience in academic medicine, clinical practice, medical professional leadership and scientific research. Prior to joining the AMA, she was the Senior Vice President for Academic Affairs at the American Board of Medical Specialties where she led core ABMS programs and staff that served to advance improved standards and methods for certification, continuing certification and lifelong assessment, as well as leading the ABMS International programs. Prior to coming to ABMS, Dr. Irons spent 30 years in academic medicine in Boston, initially at Tufts Medical Center and more recently at Boston Children’s Hospital where she oversaw clinical operations of the Division of Genetics and

Metabolism, served as the residency director for Medical Genetics and fellowship director for the Harvard Laboratory Genetics programs, led a clinical research program, and served as an Associate Professor of Pediatrics at Harvard Medical School, in addition to running a busy clinical practice. She has held numerous local and national roles in both undergraduate, graduate, and continuing medical education, including; service as the Chair of the ACGME Residency Review Committee for Medical Genetics; serving on the Boards of Directors of the Accreditation Council for Continuing Medical Education and the American College of Medical Genetics and Genomics; and serving as the President of the Association of Professors of Human and Medical Genetics. She has over 100 publications in the medical literature and is the Co-author of the 4th edition of the textbook Human Genetics and Genomics (2013). She has previously held faculty appointments at Harvard Medical School, Tufts University School of Medicine and Northwestern University Feinberg School of Medicine.

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.

ROUNDTABLE INFORMATION

The National Academies of
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 stakeholders.

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) Goldfinch Bio

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

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

Stephanie Devaney, Ph.D.
All of Us Research Program, NIH

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

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

Eric Gustafson, Ph.D.
Merck & Co.

Jill Hagenkord, M.D., FCAP
Optum Genomics

Richard Hodes, M.D.
National Institute on Aging

Geoff Hollett, Ph.D.
American Medical Association

Mira Irons, M.D.
College of Physicians Philadelphia

Praduman Jain, M.S.
Vibrent 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

James Lu, M.D., Ph.D.
Helix

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

Maximilian Muenke, M.D., FACMG
American College of Medical Genetics and Genomics

Susan E. Old, Ph.D.
National Institute of Nursing Research

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.

Wendy Rubinstein, M.D., Ph.D.
Food and Drug Administration

Nadeem Sarwar, Ph.D.
Eisai Inc.

Joan A. Scott, M.S., C.G.C.
Health Resources and Services Administration

Sam Shekar, M.D., M.P.H.
American College of Preventive Medicine

Nonniekaye Shelburne, C.R.N.P., M.S., A.O.C.N.
National Cancer Institute

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

Nikoletta Sidiropoulos, M.D.
University of Vermont Health Network Medical Group

Katherine Johansen Taber, Ph.D.
Myriad Genetics

Ryan Taft, Ph.D.
Illumina

The National Academy of Sciences, National Academy of Engineering, and National Academy of Medicine work together as the National Academies of Sciences, Engineering, and Medicine ("the Academies") to provide independent, objective analysis and advice to the nation and conduct other activities to solve complex problems and inform public policy decisions. The Academies also encourage education and research, recognize outstanding contributions to knowledge, and increase public understanding in matters of science, engineering, and medicine.

Jacquelyn Taylor, Ph.D.
Columbia University

Sharon Terry, M.A.
Genetic Alliance

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

Jameson Voss, M.D.
U.S. Air Force

Karen Weck, M.D.
College of American Pathologists

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

Huntington F. Willard, Ph.D.
Geisinger National Precision Health

Sarah Wordsworth, Ph.D.
University of Oxford

Alicia Zhou, Ph.D.
Color Genomics

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

STRATEGIC PLAN

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 stakeholders, 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.

Roundtable on **GENOMICS** and **PRECISION HEALTH**

BACKGROUND INFORMATION

The National Academies' Roundtable on Genomics and Precision Health: Where we have been and where we are heading

Geoffrey Ginsburg,[1,6] Michelle Penny,[2,6] W. Gregory Feero,[3] Mona Miller,[4,7] Siobhan Addie,[5] and Sarah H. Beachy[5,*]

Summary

The clinical application of genetics and genomics to advance precision health is one of the most dynamic and promising areas of medicine. In 2020, building on nearly 15 years of work, the Roundtable on Genomics and Precision Health of the National Academies of Sciences, Engineering, and Medicine undertook a strategic planning process to assess its strengths, consider the current challenges facing the field, and set out new goals for its future work. As a result, the Roundtable has updated its vision and mission and prioritized four major areas of inquiry—innovation, dialogue, equity, and adoption—while keeping true to its founding goal of providing a neutral convening space for the diversity of stakeholders in genomics and precision health. The Roundtable is unique for its breadth of membership and is committed to fostering a new era for precision health built on decades of expanding knowledge and the emergence of new technologies. To achieve its goals, the Roundtable seeks to broaden its membership's diversity and to engage with new audiences. Roundtable members explore how evidence-based discoveries in genomics could be adopted and used in innovative ways to better serve human health, how equitable access to genomic and precision health technologies can be ensured, and how the Roundtable and broader genomics and precision health community can communicate more effectively to inform the public regarding genomics and precision health. As a first principle, the Roundtable is working to support the overall goal that *all people benefit* from genomics for precision health.

Introduction

Over three decades, the growth and application of genomics knowledge has expanded exponentially. Its transformative impact touches all of biomedicine and spans research, clinical care, and consumer applications with leadership in public, private, and philanthropic sectors. As recently noted in a *NEJM* perspective piece celebrating the National Academy of Medicine's 50th anniversary, progress to decode the human genome has transformed science and medicine, and "the discovery of more than 100,000 robust associations between genomic regions and common diseases has pointed to new biologic mechanisms" across specialties and generated new diagnostics, counseling options, treatments, and therapies.[1]

This scientific and health progress is also fueling economic growth and an entire private sector of businesses fostering research and novel applications. The once-nascent global genomics market size has grown to $17.2 billion in 2019, and one private market report projected it to reach $82.6 billion by 2027.[2] Private sector organizations, too, are interwoven in the fabric of genomics advances: diagnostics and technology companies are securing large amounts of public and venture capital to build new tools, biotechnology firms are applying genomic knowledge to create new therapeutics, consumer genomics companies are leveraging large datasets to advance basic and clinical research as well as consumer-driven testing for non-clinical and clinical use, and health systems are rapidly innovating to bring clinically actionable data into health care settings to improve decision-making, operations, and outcomes. An analysis by the American Society of Human Genetics released in 2021 found that human genetics and genomics contributed $265 billion to the U.S. economy in 2019 alone, a 5-fold increase over a decade.[3] Further, the report indicated that, on the basis of tax revenues from and investment in human genetics and genomics, the federal government's overall return on investment in this space was 4.75 to 1. Despite the many recent advances in the field of genomic medicine, concerns remain about managing costs associated with research and development and ensuring that there is demonstrated clinical value, including improved patient outcomes.[4] Mechanisms for gathering additional evidence to support the use of genomics innovations and assessing their potential costs and benefits will be important as the field moves forward.

The potential of genomics to change the practice of medicine was recognized several years ago and is what led the National Academies of Sciences, Engineering, and Medicine

---

[1] Duke Center for Applied Genomics and Precision Medicine, Duke University, Durham, NC 27708, USA; [2] Goldfinch Bio, Cambridge, MA 02142, USA; [3] Maine Dartmouth Family Medicine Residency, Augusta, ME 04330, USA; [4] American Society of Human Genetics, Rockville, MD 20852, USA; [5] Health and Medicine Division, The National Academies of Sciences, Engineering, and Medicine, Washington, DC 20001, USA

[6] These authors contributed equally

[7] Mona Miller is a paid employee of the American Society of Human Genetics, which owns *The American Journal of Human Genetics*. As CEO, she has no editorial input on ASHG journals and played no role in *The Journal*'s review or acceptance of this manuscript.

*Correspondence: sbeachy@nas.edu

https://doi.org/10.1016/j.ajhg.2021.08.015.

(under the former Institute of Medicine) to found the Roundtable on Translating Genomic-Based Research for Health in 2007. Roundtables at the National Academies are convening activities that consist of members from across sectors and disciplines who represent government, for-profit and non-profit private sectors, patient groups, and academia. The members select challenging topics of importance and facilitate a range of discussions in a neutral setting to impact policies and programs, facilitate collaborations, and generate new ideas to shape the field. Roundtable members and the larger audience they reach (e.g., through their activities such as public workshops and perspective papers) apply the insights and lessons learned from these discussions within their own organizations and research activities. Often, the output from the Roundtable shapes policies at member organizations and in the broader community. Roundtables differ from other National Academies activities such as consensus studies, which publish recommendations or conclusions. At its inaugural meeting, the Roundtable members shared their mission "to advance the field of genomics and improve the translation of research findings to health care, education, and policy." Some of the issues at that time included examining standards of evidence for the use of genetic/genomic tests or services in health care, the need for consistent oversight of the quality and validity of new genetic tests, concern over the commercialization of discoveries (e.g., patents and direct-to-consumer uses), adequate genetics training of health care professionals, bioethical issues of privacy and potential harms from the implementation of new genetic tools and technologies, and a recognition of the need for a high-level convening activity across stakeholder groups to foster discussions and inform the field. Since then, the Roundtable has fulfilled the role of a convener of multiple stakeholder groups to explore advances in the field by serving as a unique assembly for discussion, action, and outcomes spanning academia, industry, government, professional societies and associations, patient groups, and more. The members still share a mutual interest in addressing the issues surrounding the translation of genomics research for use in improving human health. Over the years, the group has revised its focal areas through horizon scanning and member input, but the 2020 strategic plan is the first of its kind in the Roundtable's history; it has a formal and deliberate process to ensure we are meeting the needs of the rapidly evolving field in a more project- and output-driven way.

Impact and evolution of the Roundtable

A major impact of the Roundtable on the field has been through its 28 public workshops and companion proceedings which have covered a wide variety of topics, including generating evidence for genome-based diagnostic test and therapeutic development,[4,5] integrating genomics into clinical practice,[6] assessing the economics of genomic medicine,[7] improving genetics education,[8] applying implementation science,[9] understanding disparities in access,[10] and exploring the roles of consumer genomics and digital health technologies in the health care system.[11,12] A full list of the workshop titles, dates, and links to the proceedings reports can be found in Table S1.

The output from the Roundtable's activities are disseminated to a variety of audiences and through publications in *JAMA*, *Nursing Outlook*, *NAM Perspectives*, and the CDC's Office of Genomics and Precision Health blog, to name a few.[13,14] Our work has provided input to NIH and inspired the development of genomic medicine programs at large academic medical centers around the country, and our members have been consulted by legislative staff on Capitol Hill when questions arise about genetic technologies related to new proposed policy. The Roundtable has also been known for its "action collaboratives," which have focused on implementing genomic medicine, population health, and advances in use of electronic medical records for genetics and genomics. (Action collaboratives are developed as *ad hoc* activities associated with the Roundtable on Genomics and Precision Health at the National Academies of Sciences, Engineering, and Medicine [the National Academies]. Action collaboratives do not necessarily represent the views of any one organization, the Roundtable, or the National Academies and have not been subjected to the review procedures of, nor are they a report or product of, the National Academies.) The Roundtable's action collaboratives are *ad hoc*, short-term activities that stem from ideas generated from the Roundtable and often include participants with additional expertise needed for more implementation-oriented projects or products. Any products generated from these activities are independent of the National Academies and are attributed to the work of the action collaborative individuals. The Global Genomic Medicine Collaborative (G2MC), an action collaborative originally comprising 25 participating countries, launched in 2014 under the auspices of the Roundtable on Genomics and Precision Health from the Global Leaders in Genomic Medicine Summit and was incorporated as a 501(c)3 non-profit organization just 2 years later.[15] Displaying and Integrating Genetic Information Through the Electronic Health Record (DIGITizE) was an action collaborative also initiated in 2014 with the goal of examining how genomic information can be uniformly represented and integrated into electronic health records (EHRs) in a standards-based format. DIGITizE resulted in a number of resources, including two implementation guides, one for pharmacogenomic clinical decision support rules for HLA-B*57:01 and TPMT variants and another for clinical decision support rules for patients carrying variants associated with Lynch syndrome. Pilot projects at academic medical centers with EHR developers and laboratories tested the pharmacogenomics implementation guide within the local EHRs. DIGITizE moved onto the HL7 FHIR Foundation to continue with its mission. The Genomics and Population Health Action Collaborative (GPHAC) was an action collaborative founded in 2015 to

Box 1. Future of the Roundtable on Genomics and Precision Health

Vision: realizing the full potential of health for all through genomics and precision health.

Mission: to 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.

Guiding principles:

- 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.

identify challenges and potential best practices for the widespread integration of evidence-based genomics applications in population health programs. Representatives from state public health departments, federal government, health care systems, and other non-profit and academic institutions met to collaborate and develop their ideas around new partnerships and ways to implement and measure progress of population-based screening programs.[16] Additionally, with 14 years of achievement and more than 30 sponsoring members today, the Roundtable has produced dozens of publications that have been downloaded around the world and used to inform graduate classes, policy, and genomic medicine program development and implementation.

Over time, the Roundtable has evolved with the dynamics of the field of genetic testing as it became more widely implemented in clinical care and has increasingly been applied in innovative ways to expand people's access to information about their health. For example, in 2016, the focus of the group expanded to account for the exploration and adoption of new technologies that support the notion of precision health (e.g., digital health technologies such as wearables, sensors, and electronic medical records). Embracing this shift in focus, the membership updated their identity to the Roundtable on Genomics and Precision Health (the group was founded as the Roundtable on Translating Genomic-based Research for Health in 2007).

2020 Roundtable strategic planning

In 2020, the Roundtable undertook a comprehensive strategic planning effort to assess the research and clinical landscape and chart the group's path forward. Phases of the planning included an environmental scan of the precision medicine landscape and context in which we are working, formation of a vision statement and description of our unique role in the community (see Box 1), an analysis of strengths and weaknesses, and decisions around the group's goals and the principles to guide our future strategies.

Early in the strategic planning effort, Roundtable members and staff conducted a landscape analysis and gathered strategic planning information from several stakeholder groups across the field of genomics and genetics to better understand potential opportunities to advance the field and the challenges that lay ahead. This crosswalk of existing strategic plans from organizations working in the fields of genomics included, but was not limited to, the National Human Genome Research Institute (NHGRI) of the National Institutes of Health (NIH); the "All of Us" Research Program at the NIH; the American College of Medical Genetics (ACMG); the American Society of Human Genetics (ASHG); the Association for Molecular Pathology (AMP); the Centers for Disease Control and Prevention (CDC) Office of Genomics and Precision Health; Genetic Alliance; Genome Canada; Genomics England; the Global Alliance for Genomics and Health (GA4GH); and the National Society of Genetic Counselors (NSGC). This information-gathering exercise allowed the group to consider how they could best contribute to overcoming those challenges. Common themes across many of these plans included educating the genomics/precision health workforce, increasing awareness for the power of genomics/precision health, informing policy decisions, and improving data infrastructure and the evidence base. Issues related to ethical, legal, and social implications (ELSIs) and diversity, equity, and inclusion (DEI) were observed as cross-cutting elements in several of the plans as well.[17] A member survey was conducted in advance of the strategic planning sessions; collection of this information provided additional context for the planning and where specific attention should be directed during the planning process.

While encouraged by progress in the genomics and precision health field, the Roundtable members remained focused on the profound societal implications and use of genomic knowledge, as well as honest and sometimes

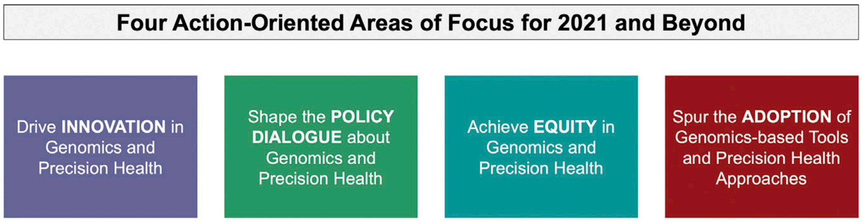


Figure 1. Areas of focus for the Genomics Roundtable

difficult reflection on the field's shortcomings and challenges and related, important choices for society, public policy, and private sector responsibility and stewardship. Open questions and challenges remain around the following:

- how to encourage and facilitate data collection, use, and sharing while maintaining rigor for due privacy and human rights goals;[18]
- deep shortcomings in the diversity of research populations and resulting disparities and importance of focusing on health equity and applicability of tools such as polygenic risk scores;
- the legacy of genetics and genomics research and its misuse or misapplication, especially in relationship to underrepresented or marginalized populations;
- root causes underlying gaps in adoption of evidence-based clinical practices in genetics and genomics in health care and public health systems; and
- approaches for enhancing diversity in the genomics workforce.

Strategic planning discussions illuminated the challenge of focusing on genomics and precision health while also acknowledging the interface of genomics with many other facets of medicine, health, and society. Roundtable members recognized the role of the social determinants of health (i.e., the environmental conditions where people are born, live, learn, work, play, worship, and age)[19] in contributing to improving population health outcomes.[20] The strategic planning conversations among Roundtable members surfaced many topics—such as the interplay between genomics and the social determinants of health and the role of digital technologies, privacy policies, and data management, among others. The group considered these important topics on the basis of timeliness, membership interest, and the potential ability of the Roundtable's actions to advance the subject area for the good of the biomedical community. While there were many important topics raised, certain topics were chosen as fruitful areas for future collaboration with other stakeholder groups that were already focused on those challenges, such as the National Academies' Roundtable on Population Health Improvement.

Not surprisingly, with the diverse spectrum of expertise on the Roundtable, more than a dozen potential areas of emphasis emerged from the strategic planning efforts, so Roundtable members used a logic model in combination with a discussion-driven process to consider where the group's resources would have the most impact over the next few years. Ultimately, the group decided to focus on and prioritize four specific action-oriented areas for 2021, which are listed in Figure 1.

Areas of focus

Following the information gathering stage and throughout 2020, the Roundtable members took stock of the group's progress, and potential, to foster this vital, transformative community and ensure broad and equitable access to genomic benefits. Over 12 months, Roundtable members assessed the group's progress, identified areas of shared interest and unique opportunity, and renewed their commitment to be a leading forum for discussion—and for *action*. The updated vision and mission of the Roundtable can be found below in Box 1.

Innovation

The goal of this workstream is to consider the barriers and facilitators of innovation for genomics-based diagnostics, risk assessment tools, and therapies such that policies, tools, and pathways can be developed to enhance innovation in genomics and precision health. The initial focus will be on case studies from the past decade to provide best practices and lessons learned that can then be used to promote innovative approaches that can accelerate commercialization and integration to drive impact of genomics on precision health. In this regard, the Roundtable could also provide a venue for discussing the potential value of genomics innovations to enable the field to move forward more efficiently and effectively.

Adoption

The goal of this workstream is to understand barriers and facilitators to the adoption of genomics as a tool for improved health and to use this knowledge to accelerate the appropriate adoption of evidence-based practices in genomics and precision health. Initial work will include gathering information from existing genomic medicine programs where widespread integration of genomics has been undertaken in order to identify gaps in adoption, root causes underlying these gaps, and potentially generalizable solutions. From this initial work, the adoption group will engage with the broad community of stakeholders represented on the Roundtable to develop activities that might serve to facilitate advancement in equitable access to genomic and precision health technologies, including informing policy deliberations, educational efforts, health technology standards development, and funding prioritization. The end goal of the group is to help ensure that health care and public health systems have a well-developed and clear path for adoption of genomic and precision health technologies in order to benefit the widest number of individuals possible.

Equity

The goal of this workstream is to foster action related to underrepresentation of diverse populations and inequities in genomic research, workforce, and access to genomic services by people who need them. The initial approach will involve an introspective view on the processes and practices the Roundtable employs to achieve its mission as well as learning from the external genomics communities broadly grappling with equity challenges. The aim of the group's activities will be to offer opportunities for researchers, clinicians, and others to consider how to more fully represent underserved groups in genomics research and also to reduce the genomic and precision medicine divide in health care.

Shaping the dialogue

The goal of this workstream is to accelerate the dissemination of actionable knowledge to shape practice and increase public awareness and to inform and influence how decisions are made across the Roundtable's broad stakeholder community. One approach that this group will take is to implement an infrastructure within and across the redefined workstreams for the purpose of increasing awareness, transparency, and accountability to the Roundtable's commitment to action. Success of the Roundtable is dependent on effective communication and dialog with our community of participants, researchers, clinicians, industry, and policy makers.

Plan implementation

With the working group themes finalized in the fall of 2020, members chose their areas of interest to commit to, and co-facilitators were chosen for each group and asked to work with the members to develop quarterly plans and metrics for assessing progress for 2021. The equity group began their work as early as November 2020, hosting a series of educational sessions for the Roundtable members on systemic racism and diversity needs of the field. In early 2021, all of the new working groups convened to take action around their quarterly goals—some of which are to collect information to inform their steps toward papers (such as this one) or public discussions.

A path to impact

The Roundtable is poised to impact the field of genomics and precision health over the next decade. This cannot be accomplished without the engagement of the broadest possible community of stakeholders and experts both in the United States and globally. Realizing the full potential of Genomics and Precision Health will require your engagement, your partnership, and your voice in our work. For our work to be as impactful as we aspire it to be, we call on you to join us in the debate and dialogue, the strategies and policy agenda, and ultimately in its implementation. To learn more about how to connect with the Roundtable and its members, please visit the group's website (available in the web resources section below).

Supplemental information

Supplemental information can be found online at https://doi.org/10.1016/j.ajhg.2021.08.015.

Acknowledgments

The authors would like to thank Vence Bonham (NIH/NHGRI), Nikoletta Sidiropoulos (University of Vermont Medical Center), and Catherine Wicklund (Northwestern University) for their careful review of the paper and thoughtful comments and edits.

Declaration of interests

Geoffrey Ginsburg reports the following: consulting for Konica-Minolta and Fabric Genomics and ownership interest in Peer Medical, Origin Commercial Advisors, Predigen, MeTree&You, and Coprata. He receives royalties from Elsevier.

Web resources

Global Genomic Medicine Collaborative (G2MC), https://g2mc.org/
Roundtable on Genomics and Precision Health, https://www.nationalacademies.org/our-work/roundtable-on-genomics-and-precision-health

References

1. Collins, F.S., Doudna, J.A., Lander, E.S., and Rotimi, C.N. (2021). Human Molecular Genetics and Genomics - Important Advances and Exciting Possibilities. N. Engl. J. Med. *384*, 1–4.
2. Fortune Business Insights (2020). Pharmaceutical – Genomics Market Analysis. https://www.fortunebusinessinsights.com/industry-reports/genomics-market-100941.
3. American Society of Human Genetics (ASHG) (2021). The Economic Impact and Functional Applications of Human Genetics and Genomics. https://www.ashg.org/wp-content/uploads/2021/05/ASHG-TEConomy-Impact-Report-Final.pdf.
4. IOM (2012a). Genome-Based Therapeutics: Targeted Drug Discovery and Development: Workshop Summary (Washington, DC: The National Academies Press).
5. IOM (2011). Generating Evidence for Genomic Diagnostic Test Development: Workshop Summary (Washington, DC: The National Academies Press).
6. IOM (2012b). Integrating Large-Scale Genomic Information into Clinical Practice: Workshop Summary (Washington, DC: The National Academies Press).

7. IOM (2013). The Economics of Genomic Medicine: Workshop Summary (Washington, DC: The National Academies Press).
8. IOM (2015). Improving Genetics Education in Graduate and Continuing Health Professional Education: Workshop Summary (Washington, DC: The National Academies Press).
9. NASEM (2016). Applying an Implementation Science Approach to Genomic Medicine: Workshop Summary (Washington, DC: The National Academies Press).
10. NASEM (2018). Understanding Disparities in Access to Genomic Medicine: Proceedings of a Workshop. (Washington, DC: The National Academies Press).
11. NASEM (2020a). Exploring the Current Landscape of Consumer Genomics: Proceedings of a Workshop. (Washington, DC: The National Academies Press).
12. NASEM (2020b). The Role of Digital Health Technologies in Drug Development: Proceedings of a Workshop. (Washington, DC: The National Academies Press).
13. Chambers, D.A., Feero, W.G., and Khoury, M.J. (2016). Convergence of Implementation Science, Precision Medicine, and the Learning Health Care System: A New Model for Biomedical Research. JAMA *315*, 1941–1942.
14. Williams, J.K., Feero, W.G., Leonard, D.G.B., and Coleman, B. (2017). Implementation science, genomic precision medicine, and improved health: A new path forward? Nurs. Outlook *65*, 36–40.
15. Ginsburg, G.S. (2019). A Global Collaborative to Advance Genomic Medicine. Am. J. Hum. Genet. *104*, 407–409.
16. Murray, M.F., Evans, J.P., Angrist, M., Chan, K., Uhlmann, W., Doyle, D.L., Fullerton, S.M., Ganiats, T., Hagenkord, J., Imhof, S., et al. (2018). A Proposed Approach for Implementing Genomics-Based Screening Programs for Healthy Adults. NAM Perspectives, Discussion Paper, National Academy of Medicine, Washington, DC.
17. Green, E.D., Gunter, C., Biesecker, L.G., Di Francesco, V., Easter, C.L., Feingold, E.A., Felsenfeld, A.L., Kaufman, D.J., Ostrander, E.A., Pavan, W.J., et al. (2020). Strategic vision for improving human health at The Forefront of Genomics. Nature *586*, 683–692.
18. Meagher, K.M., Cadigan, R.J., Henderson, G.E., and Juengst, E. (2019). Public Health Genomics, Biobanking, and Ethics. Oxford Handbooks Online. https://www.oxfordhandbooks.com/view/10.1093/oxfordhb/9780190245191.001.0001/oxfordhb-9780190245191-e-57.
19. U.S. Department of Health and Human Services (HHS) (2021). Healthy People 2030: Social Determinants of Health. https://health.gov/healthypeople/objectives-and-data/social-determinants-health.
20. Braveman, P., and Gottlieb, L. (2014). The social determinants of health: it's time to consider the causes of the causes. Public Health Rep. *129* (*Suppl 2*), 19–31.

The American Journal of Human Genetics, Volume *108*

Supplemental information

The National Academies' Roundtable on Genomics and Precision Health: Where we have been and where we are heading

Geoffrey Ginsburg, Michelle Penny, W. Gregory Feero, Mona Miller, Siobhan Addie, and Sarah H. Beachy

Workshop	Year	Link to Workshop Proceedings
EVIDENCE FOR POLICY AND PRACTICE		
Improving the Efficiency and Effectiveness of Genomic Science Translation	2012	https://www.nap.edu/catalog/18549/improving-the-efficiency-and-effectiveness-of-genomic-science-translation-workshop
Integrating Large-Scale Genomic Information into Clinical Practice	2011	https://www.nap.edu/catalog/13256/integrating-large-scale-genomic-information-into-clinical-practice-workshop-summary
Generating Evidence for Genomic Diagnostic Test Development	2010	https://www.nap.edu/catalog/13133/generating-evidence-for-genomic-diagnostic-test-development-workshop-summary
Value of Genetic and Genomic Technologies	2010	https://www.nap.edu/catalog/12947/the-value-of-genetic-and-genomic-technologies-workshop-summary
Systems for Research and Evaluation for Translating Genome-Based Discoveries for Health	2009	https://www.nap.edu/catalog/12691/systems-for-research-and-evaluation-for-translating-genome-based-discoveries-for-health
Innovations in Service Delivery in the Age of Genomics	2008	https://www.nap.edu/catalog/12601/innovations-in-service-delivery-in-the-age-of-genomics-workshop
DIAGNOSTIC APPLICATIONS		
Assessing Genomic Sequencing Information for Health Care Decision Making	2014	https://www.nap.edu/catalog/18799/assessing-genomic-sequencing-information-for-health-care-decision-making-workshop
Conflict of Interest in Medical Innovation: Assuring Integrity while Facilitating Innovation in Medical Research	2013	https://www.nap.edu/catalog/18723/conflict-of-interest-and-medical-innovation-ensuring-integrity-while-facilitating
Refining Processes for the Co-Development of Genome-Based Therapeutics and Companion Diagnostic Tests	2013	https://www.nap.edu/catalog/18617/refining-processes-for-the-co-development-of-genome-based-therapeutics-and-companion-diagnostic-tests
Challenges and Opportunities in Using Newborn Screening Samples for Translational Research	2010	https://www.nap.edu/catalog/12981/challenges-and-opportunities-in-using-residual-newborn-screening-samples-for-translational-research
EDUCATION, ENGAGEMENT, AND CULTURAL CHANGE		
Improving Genetics Education in Graduate and Continuing Health Professional Education	2014	https://www.nap.edu/catalog/18992/improving-genetics-education-in-graduate-and-continuing-health-professional-education
Sharing Clinical Research Data	2012	https://www.nap.edu/catalog/18267/sharing-clinical-research-data-workshop-summary
BUILDING LEARNING HEALTH CARE SYSTEMS		
Genomics-Enabled Learning Health Care Systems: Gathering and Using Genomic Information to Improve Patient Care and Research	2014	https://www.nap.edu/catalog/21707/genomics-enabled-learning-health-care-systems-gathering-and-using-genomic
IMPLEMENTATION, PUBLIC HEALTH, AND DISPARITIES		
Understanding Disparities in Access to Genomic Medicine	2018	https://www.nap.edu/catalog/25277/understanding-disparities-in-access-to-genomic-medicine-proceedings-of-a

Implementing and Evaluating Genomic Screening Programs in Health Care Systems	2017	https://www.nap.edu/catalog/25048/implementing-and-evaluating-genomic-screening-programs-in-health-care-systems
Applying an Implementation Science Approach to Genomic Medicine	2015	https://www.nap.edu/catalog/23403/applying-an-implementation-science-approach-to-genomic-medicine-workshop-summary
Assessing the Economics of Genomic Medicine	2012	https://www.nap.edu/catalog/18276/the-economics-of-genomic-medicine-workshop-summary
Diffusion and Use of Genomic Innovations in Health and Medicine	2007	https://www.nap.edu/catalog/12148/diffusion-and-use-of-genomic-innovations-in-health-and-medicine
DISCOVERING AND DEVELOPING PRECISION THERAPEUTICS		
The Role of Digital Health Technologies in Drug Development	2020	https://www.nap.edu/catalog/25850/the-role-of-digital-health-technologies-in-drug-development-proceedings
Enabling Precision Medicine: The Role of Genetics in Clinical Drug Development	2017	https://www.nap.edu/catalog/24829/enabling-precision-medicine-the-role-of-genetics-in-clinical-drug
Deriving Drug Discovery Value from Large-Scale Genetic Bioresources	2016	https://www.nap.edu/catalog/23601/deriving-drug-discovery-value-from-large-scale-genetic-bioresources-proceedings
Drug Repurposing and Repositioning	2013	https://www.nap.edu/catalog/18731/drug-repurposing-and-repositioning-workshop-summary
Genome-Based Diagnostics: Demonstrating Clinical Utility in Oncology	2012	https://www.nap.edu/catalog/18275/genome-based-diagnostics-demonstrating-clinical-utility-in-oncology-workshop-summary
Genome-Based Therapeutics: Targeted Drug Discovery and Development	2012	https://www.nap.edu/catalog/13436/genome-based-therapeutics-targeted-drug-discovery-and-development-workshop-summary
Genome-Based Diagnostics: Clarifying Pathways to Clinical Use	2011	https://www.nap.edu/catalog/13359/genome-based-diagnostics-clarifying-pathways-to-clinical-use-workshop-summary
Establishing Precompetitive Collaborations to Stimulate Genomics Driven Drug Development	2010	https://www.nap.edu/catalog/13015/establishing-precompetitive-collaborations-to-stimulate-genomics-driven-product-development-workshop
CONSUMER GENOMICS		
Exploring the Current Landscape of Consumer Genomics	2019	https://www.nap.edu/catalog/25713/exploring-the-current-landscape-of-consumer-genomics-proceedings-of-a
Direct-to-Consumer Genetic Testing	2009	https://www.nap.edu/catalog/13021/direct-to-consumer-genetic-testing-summary-of-a-workshop

Links to Additional Resources

- Call for nominations to serve on an anticipated committee on Use of Race, Ethnicity, and Ancestry as Population Descriptors in Genomics Research: https://survey.alchemer.com/s3/6559180/Call-for-Nominations-Use-of-Race-Ethnicity-and-Ancestry-as-Population-Descriptors-in-Genomics-Research

- Roundtable on Genomics and Precision Health. NASEM. https://www.nationalacademies.org/our-work/roundtable-on-genomics-and-precision-health

- Feero, 2017. Introducing "Genomics and Precision Health." *Journal of the American Medical Association* 317(18): 1842-1843. https://jamanetwork.com/journals/jama/fullarticle/2625302

- Feero, Wicklund, & Veenstra, 2018. Precision Medicine, Genome Sequencing, and Improved Population Health. *Journal of the American Medical Association* 319(19): 1979-1980. https://jamanetwork.com/journals/jama/fullarticle/2675723?resultClick=1

- Feero & Wicklund, 2020. Consumer Genomic Testing in 2020. *Journal of the American Medical Association* 323(15): 1445-1446. https://jamanetwork.com/journals/jama/fullarticle/2763486?resultClick=1