

*The National
Academies of*

SCIENCES
ENGINEERING
MEDICINE

Roundtable on
GENOMICS
AND
**PRECISION
HEALTH**

2017 ANNUAL REPORT

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About the Roundtable

The Roundtable on Genomics and Precision Health brings together leaders from government, academia, industry, foundations, associations, patient communities, and other stakeholder groups to meet and discuss global issues surrounding the translation of genomics and genetics research findings into medicine, public health, education, and policy. The primary purpose of the Roundtable is to foster dialogue across sectors and among interested parties and institutions and to illuminate and scrutinize critical scientific and policy issues where Roundtable engagement and input will help further the field.

The Roundtable membership identifies scientific and policy issues where discussion and collaboration will help enable the translation of genomics into health care applications. Specific issues and agenda topics are determined by the Roundtable members and span a broad range of areas relevant to the translation process. Current areas of emphasis include the development of precision therapeutics; clinical implementation of genomic medicine; health care disparities; health information technology and digital health; public health genomics; the use of genomic information for health care decision making; using genomic information and data science to generate knowledge for clinical practice and research; and education and ethical, legal, and social issues.

To achieve its objectives, the Roundtable conducts structured discussions, public workshops, and meetings; enters into information-gathering activities; develops authored viewpoints and perspectives; organizes and supports collaboratives; and publishes workshop proceedings.

nationalacademies.org/GenomicsRT

Working Groups & Areas of Interest

The Roundtable identifies interest areas that need highlighting and attention based on member areas of expertise and the evolution of the field of genomics and precision health. These groups are fluid in that they adapt to address challenges and areas of focus over time. The working groups for 2017-18 include:



Precision Therapeutics

The cost of developing new therapies has been on the rise, leading many pharmaceutical companies to examine innovative strategies to revitalize and create efficiencies in their drug discovery and development processes, including the adoption of genetically guided strategies to reduce attrition rates and increase the odds of success. This group is interested in enabling precision medicine by identifying which patients should be treated with a particular medicine. Group members are exploring methods for recruiting patients to clinical trials based on genetics, identifying biomarkers for responders, and developing new resources that can enable genetics-based clinical trials. This group is also assessing innovative collaborative models in practice to leverage cross-sector contributions to accelerate drug discovery and development.



Implementation and Public Health Systems

With technological advances in gene sequencing driving down costs and a growing demonstration of utility for large-scale sequencing in disease diagnosis or to identify and monitor treatments for patients, an opportunity for more widespread use in health care systems and the public health system has arisen. This group is exploring the use of genomic screening programs across health care systems and the opportunities for generating, collecting, and sharing data and evidence on a large scale; the return on investment and value that these programs can bring to a system; and the challenges that exist for reaching a diverse and representative population when the goal is to have genetic testing available to all.



Digital Health and Genomics

This group is exploring the intersection between digital health technologies and genomic data by determining how the two fields can interface to enable precision health, improve research (patient-reported outcomes, continuous phenotypes), and clinical care (monitoring, feedback, adherence). Group members are interested in applying what has been learned from the consumer genomics space (e.g., about consumer access, consumer comprehension, regulatory jurisdiction, health care provider readiness, clinical utility, personal utility, potential burden on the health system, and quality standards) to the developing field of digital health for more widespread applications for precision medicine and to enable large-scale genetic/genomic testing initiatives. Other areas that may be explored by the group include the role of digital health platforms as a possible mechanism to link disparate data sources and information that is not currently in the electronic health record (EHR) as well as ensuring that integration with the EHR can be achieved for both research and clinical care. Overall, the group seeks to convene experts from the fields of genomics and digital health to illuminate areas of synergy and to inform each other about successes and challenges encountered in each sector.



Overview

The Overview group examines cross-cutting issues for all Roundtable members and working groups and responds to emerging issues within the genetics and genomics field. Currently, this group is focusing its efforts on precision medicine and precision health.

Public Workshops

Enabling Precision Medicine: The Role of Genetics in Clinical Drug Development

A Workshop

MARCH 8, 2017

<http://bit.ly/PrecisionMedWS>

Together with the Forum on Drug Discovery, Development, and Translation, the Roundtable co-hosted this workshop as a way to explore the successes, challenges, and best practices for effectively utilizing genetic information in the design and implementation of clinical trials to support the development of precision medicines. Discussions explored how clinical trials with genetically identified participants can enable more efficient and effective drug development and advance precision medicine. Speakers also examined ongoing genetics-based clinical trials across a variety of diseases, focusing on logistical design challenges, best practices and lessons learned, and novel mechanisms to engage participants and improve enrollment into clinical trials based on genetic characteristics.

Implementing and Evaluating Genomic Screening Programs in Health Care Systems

A Workshop

NOVEMBER 1, 2017

Proceedings of a Workshop available spring 2018

<http://bit.ly/ImplementingWS>

This workshop explored the implementation of genomics-based screening programs into health care systems. Case studies of ongoing or future genomics screening programs were highlighted during the workshop as a way to understand successes, lessons learned, and challenges regarding evidence and economic considerations (e.g., clinical utility, value), policy environments (e.g., alleviating privacy and discrimination concerns for participants), and data sharing within and across institutions.



PUBLICATION

Enabling Precision Medicine:
The Role of Genetics in Clinical Drug Development
Proceedings of a Workshop

<http://bit.ly/PrecisionMedWSPub>

RELEASED: JULY 10, 2017

Meetings

March 9, 2017

Roundtable members and speakers discussed challenges in the field including coverage for and access to genomic medicine services, considerations about return on investment for health systems as they make decisions about implementing genomics-based programs, and data issues related to incorporating digital health applications into clinical care workflows.

July 17–18, 2017

Members explored the issues that could impact the future of genomics research, considered the role of the National Academies in the new presidential administration, and discussed opportunities to interact with the efforts of the All of Us research program. Speaker panels discussed the opportunities to utilize best practices from health and social sciences in genomics research to address health care disparities and access to services as well as the regulatory and clinical challenges in utilizing digital health data for patient care.

November 2, 2017

Roundtable members and speakers discussed current research on health care disparities in genomic medicine and explored potential avenues for addressing the issues of cost, coverage, and access to genomic testing and services, including learning from and developing a resource of best practices of programs that are effectively engaging and caring for diverse populations. The Roundtable also engaged with speakers working in the field of artificial intelligence to learn about intersections between genomics and artificial intelligence—such as deep learning using genomic sequencing data—and the potential for improved outcomes for drug response based on phenotypic and genotypic information.



Action Collaboratives

The following action collaboratives are ad hoc activities under the auspices of the Roundtable on Genomics and Precision Health at the National Academies of Sciences, Engineering, and Medicine (the National Academies). Products produced by an action collaborative 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.



Global Genomic Medicine Collaborative (G2MC)

<https://g2mc.org>

The Global Genomic Medicine Collaborative (G2MC), an action collaborative launched in 2014 under the auspices of the Roundtable on Genomics and Precision Health following the Global Leaders in Genomic Medicine meeting, was incorporated as a 501(c)3 nonprofit organization in 2016 and transitioned from the Roundtable in 2017 to continue its work in collaboration with the Global Alliance for Genomics and Health (GA4GH). G2MC aims to develop knowledge, resources, and collaborations that can enable genomic medicine implementation globally.



Displaying and Integrating Genetic Information Through the EHR (DIGITizE)

<http://bit.ly/DIGITizEAC>

The action collaborative on Displaying and Integrating Genetic Information Through the EHR (DIGITizE), convenes key stakeholders from health information technology and management vendors, academic health centers, government agencies, and other organizations to work together to examine how genomic information can be uniformly represented and integrated into electronic health records in a standards-based format. An implementation guide containing the message structures and clinical decision support rules for a pharmacogenomics use case was finalized at the end of 2015 in preparation for pilot programs that began in 2016. Pilot participants include Intermountain Healthcare, Boston Children's Hospital, Duke University, Johns Hopkins University, Mission Health, Partners Healthcare, St. Jude Children's Research Hospital, University of Iowa, and University of Utah. Working groups explored additional use cases for further phases of the pilot projects in conjunction with a collaborative partner. In 2018, DIGITizE will become part of the FHIR Foundation, whose goal is to provide support for the expansion of the FHIR community by promoting the global adoption and implementation of this standards platform.



Genomics and Population Health Action Collaborative (GPHAC)

<http://bit.ly/GPHACAC>

The Genomics and Population Health Action Collaborative (GPHAC) was initiated at the end of 2015 to explore opportunities for genomics research and applications to be used to improve health and prevent disease; inform and engage various stakeholders about the implementation of genomics in practice; and assess how evidence-based genomic applications could be integrated into implementation activities and programs at the health care-public health interface. GPHAC is currently organized into three working groups: population screening, cascade screening, and implementation. Together, these groups are assessing best practices and outcomes for implementation of population-level genomics activities; creating tools and educational materials to facilitate the uptake of genomics-based applications in health care and disease prevention; informing and engaging stakeholders; and exploring critical aspects of pilot projects for cascade screening/testing of at-risk relatives.

Roundtable Membership

(AS OF DECEMBER 2017)

Geoffrey S. Ginsburg, M.D., Ph.D. (Co-Chair)

Duke University

Sharon F. Terry, M.A. (Co-Chair)

Genetic Alliance

Naomi Aronson, Ph.D.

Blue Cross and Blue Shield Association

Nazneen Aziz, Ph.D.

Kaiser Permanente

Rebecca Blanchard, Ph.D.

Merck and Co., Inc.

Ruth Brenner, Lt Col, USAF, MC

Air Force Medical Support Agency

Ann Cashion, Ph.D.

National Institute of Nursing Research

Robert B. Darnell, M.D., Ph.D.

The Rockefeller University

Barry Dickinson, Ph.D.

American Medical Association

Joseph Donahue

Accenture

W. Gregory Feero, M.D., Ph.D.

Journal of the American Medical Association

Andrew N. Freedman, Ph.D.

National Cancer Institute

Marc Grodman, M.D.

Columbia University

Jill Hagenkord, M.D.

Color Genomics

Richard Hodes, M.D.

National Institute on Aging

Katherine Johansen Taber, Ph.D.

Counsyl, Inc.

Muin Khoury, M.D., Ph.D.

Centers for Disease Control and Prevention

Thomas Lehner, Ph.D., M.P.H.

National Institute of Mental Health

Debra Leonard, M.D., Ph.D.

College of American Pathologists

David Litwack, Ph.D.

U.S. Food and Drug Administration

Laura Lyman Rodriguez, Ph.D.

National Human Genome Research Institute

Robert McCormack, Ph.D.

Johnson & Johnson

Jennifer Moser, Ph.D.

Department of Veterans Affairs

Michelle Penny, Ph.D.

Biogen

Victoria M. Pratt, Ph.D., FACMG

Association for Molecular Pathology

Nadeem Sarwar, Ph.D.

Eisai Inc.

Derek Scholes, Ph.D.

American Society of Human Genetics

Sheri Schully, Ph.D.

NIH Office of Disease Prevention

Joan A. Scott, M.S., C.G.C.

Health Resources and Services Administration

Sam Shekar, M.D., M.P.H.

Northrop Grumman Information Systems

Ryan Taft, Ph.D.

Illumina

Joyce Tung, Ph.D.

23andMe, Inc.

David Veenstra, Pharm.D., Ph.D.

University of Washington

Michael S. Watson, Ph.D.

American College of Medical Genetics and Genomics

Catherine A. Wicklund, M.S., C.G.C.

National Society of Genetic Counselors

Huntington F. Willard, Ph.D.

Geisinger National Precision Health

Janet K. Williams, Ph.D., R.N., FAAN

American Academy of Nursing

Roundtable Sponsors

23andMe, Inc.

Accenture

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American College of Medical Genetics and Genomics

American Medical Association

American Society of Human Genetics

Association for Molecular Pathology

Biogen

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College of American Pathologists

Color Genomics

Counsyl, Inc.

Department of Veterans Affairs

Eisai Inc.

Geisinger

Health Resources and Services Administration

Illumina

Johnson & Johnson

Kaiser Permanente

Marc Grodman

Merck Research Laboratories

National Cancer Institute

National Human Genome Research Institute

National Institute of Mental Health

National Institute of Nursing Research

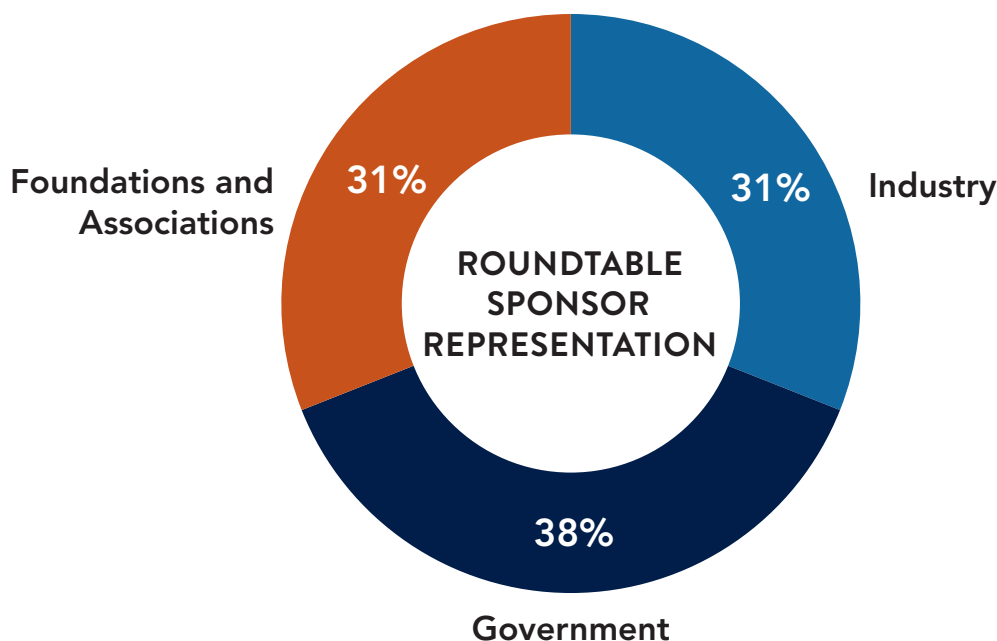
National Institute on Aging

National Institutes of Health Office of Disease Prevention

National Society of Genetic Counselors

Northrop Grumman

United States Air Force Medical Service



Roundtable Timeline

2007

July 11–12: Meeting 1

December 4: Diffusion and Use of Genomic Innovations in Health and Medicine Workshop

December 5: Meeting 2

2008

April 1–2: Meeting 3

July 28: Innovations in Service Delivery in the Age of Genomics Workshop

July 29: Meeting 4

October 6–7: Meeting 5

2009

February 12: Systems for Research and Evaluation for Translating Genome-Based Discoveries for Health Workshop

February 13: Meeting 6

June 9–11: Meeting 7

August 31–September 1: Direct-to-Consumer Genetic Testing Workshop

November 16–17: Meeting 8

2010

March 22: The Value of Genetic and Genomic Technologies Workshop

March 23: Meeting 9

May 24: Challenges and Opportunities in Using Newborn Screening Samples for Translational Research Workshop; Meeting 10

July 22: Establishing Precompetitive Collaborations to Stimulate Genomics-Driven Drug Development Workshop

July 23: Meeting 11

November 17: Generating Evidence for Genomic Diagnostic Test Development Workshop

November 18: Meeting 12

2011

February 22–23: Meeting 13

July 19: Integrating Large-Scale Genomic Information into Clinical Practice Workshop

July 20: Meeting 14

November 15: Facilitating Development and Utilization of Genome-Based Diagnostic Technologies Workshop

November 16: Meeting 15

2012

March 21: New Paradigms in Drug Discovery: How Genomic Data Are Being Used to Revolutionize the Drug Discovery and Development Process Workshop

March 22: Meeting 16

May 24: Evidence for Clinical Utility of Molecular Diagnostics in Oncology Workshop

July 17–18: Assessing the Economics of Genomic Medicine Workshop

July 18: Meeting 17

October 4–5: Sharing Clinical Research Data Workshop

December 3: Improving the Efficiency and Effectiveness of Genomic Science Translation Workshop

December 4: Meeting 18

2013

February 27: Refining Processes for the Co-Development of Genome-Based Therapeutics and Companion Diagnostic Tests Workshop

February 28: Meeting 19

June 5: Conflict of Interest and Medical Innovation: Ensuring Integrity While Facilitating Innovation in Medical Research Workshop

June 24: Genomics-Enabled Drug Repositioning and Drug Repurposing Workshop

June 25: Meeting 20

December 4–5: Meeting 21

2014

February 3: Assessing Genomic Sequencing Information for Health Care Decision Making Workshop

February 4: Meeting 22

August 18: Improving Genetics Education in Graduate and Continuing Health Professional Education Workshop

August 19: Meeting 23

December 8: Genomics-Enabled Learning Health Care Systems: Gathering and Using Genomic Information to Improve Patient Care and Research Workshop

December 9: Meeting 24

2015

March 10–11: Meeting 25

July 14–15: Meeting 26

November 19: Applying an Implementation Science Approach to Genomic Medicine Workshop

November 20: Meeting 27

2016

March 22: Deriving Drug Discovery Value from Large-Scale Genetic Bioresources Workshop

March 23: Meeting 28

July 19–20: Meeting 29

November 9: Meeting 30

2017

March 8: Enabling Precision Medicine: The Role of Genetics in Clinical Drug Development Workshop

March 9: Meeting 31

July 17–18: Meeting 32

November 1: Implementing and Evaluating Genomic Screening Programs in Health Care Systems Workshop

November 2: Meeting 33

2018

March 20: Meeting 34

June 27: Understanding Disparities in Access to Genomic Medicine Workshop

June 28: Meeting 35

December 4–5: Meeting 36

Roundtable Staff

Sarah H. Beachy, Ph.D.

Senior Program Officer and Roundtable
Director

Siobhan Addie, Ph.D.

Program Officer

Meredith Hackmann

Research Associate

Rebecca Ray

Senior Program Assistant

Board on Health Sciences Policy Staff

Andrew Pope, Ph.D., Senior Board Director

Hilary Bragg, Program Coordinator (until August 2017)

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To learn more about the Roundtable,
visit nationalacademies.org/GenomicsRT