

*The National
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SCIENCES
ENGINEERING
MEDICINE

Roundtable on
GENOMICS
AND
**PRECISION
HEALTH**

2018 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 discovery and 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, and 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 2018-19 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. Group members are considering patient engagement as a part of precision drug development efforts, education and awareness surrounding genetic resources, and entrepreneurial innovation that can accelerate drug development.



Implementation

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 of genomic medicine in health care systems and the public health system has arisen. This group is exploring the impact on access to genetic testing as new technologies are introduced in health care settings as well as the role of employers in providing access to genomic and genetic services as part of health benefit packages.



Digital Health

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). Other areas that may be explored by the group include strategies for successful digital health implementation in clinical care and privacy and regulatory challenges surrounding digital health applications. 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 disparities in access to genomic medicine.

Public Workshop

Understanding Disparities in Access to Genomic Medicine

A Workshop

JUNE 27, 2018

<http://bit.ly/GenomicDisparitiesWS>

This workshop explored gaps in knowledge related to access to genomic medicine and discussed health care disparities and possible approaches to overcoming differential use of genomic medicine across populations. Model programs of care for diverse patient populations were highlighted. Participants discussed current challenges and possible best practices for alleviating health care disparities as they relate to genomics-based approaches.

Prior to the workshop, several of the speakers, Roundtable members, and workshop registrants participated in a Twitter chat hosted by the National Academies' Health and Medicine Division on the topics of #GenomicsDisparities. Topics discussed during the chat included challenges faced by patients when trying to access genetics-based services, opportunities for increasing access, the role of health systems and providers in improving access, and lessons that can be learned from other areas of medicine in how genetics-based services can be equitably provided.



Panelists at the Understanding Disparities in Access to Genomic Medicine workshop

Meetings

March 20, 2018

Roundtable members and speakers discussed some of the security challenges related to storing large-scale genomic data and opportunities for securing data by using emerging technologies like blockchain. The Roundtable also explored innovative collaborative models in the pre-competitive space that utilize genomic data to accelerate the development and use of precision medicine.

June 28, 2018

The Roundtable debriefed following the workshop on disparities in access to genomic medicine and discussed current precision medicine research initiatives including All of Us and efforts around shaping health systems at the World Economic Forum. The working groups also convened in breakout sessions to identify specific themes and challenges that could be explored in the upcoming year.

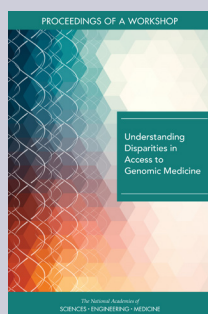
October 5, 2018

Roundtable members met virtually to discuss the recently released National Academies consensus report *Returning Individual Research Results to Participants: Guidance for a New Research Paradigm*. Members were debriefed on the recommendations from the committee as well as implications for genomic information by study staff and members of the consensus committee.

Publications



Implementing and Evaluating Genomic Screening Programs in Health Care Systems
 Proceedings of a Workshop
<http://bit.ly/GenomicScreeningPW>
 RELEASED: MARCH 16, 2018



Understanding Disparities in Access to Genomic Medicine
 Proceedings of a Workshop
<http://bit.ly/GenomicDisparitiesPW>
 RELEASED: NOVEMBER 14, 2018

Perspectives



Accelerating Precision Health by Applying the Lessons Learned from Direct-to-Consumer Genomics to Digital Health Technologies
<http://bit.ly/PrecisionHealthPerspective>
 POSTED: MARCH 19, 2018



A Proposed Approach for Implementing Genomics-Based Screening Programs for Healthy Adults
<http://bit.ly/GenomicsScreeningPerspective>
 POSTED: DECEMBER 3, 2018

Perspectives, published by the National Academy of Medicine (NAM), are individually authored by Roundtable and Forum members and outside experts in health and health care. The views expressed in these papers are those of the authors and not necessarily of the authors' organizations, the National Academy of Medicine (NAM), or the National Academies of Sciences, Engineering, and Medicine (the National Academies). Perspectives are intended to help inform and stimulate discussion. They are not reports of the NAM or the National Academies.

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.



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.



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 became 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 platform.

Roundtable Membership

(AS OF DECEMBER 2018)

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

Duke University

Michelle Penny, Ph.D. (Co-Chair)

Biogen

Naomi Aronson, Ph.D.

Blue Cross and Blue Shield Association

Aris Baras, M.B.A.

Regeneron Pharmaceuticals

John W. Belmont, M.D., Ph.D.

Illumina

Karina Bienfait, Ph.D.

Merck and Co., Inc.

Ann Cashion, Ph.D.

National Institute of Nursing Research

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

The Rockefeller University

Joseph Donahue

Accenture

Katherine Donigan, Ph.D.

U.S. Food and Drug Administration

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

JAMA

Marc Grodman, M.D.

Genosity

Jill Hagenkord, M.D.

Color Genomics

Emily Harris, Ph.D., M.P.H.

National Cancer Institute

Richard Hodes, M.D.

National Institute on Aging

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

Centers for Disease Control and Prevention

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

National Institute of Mental Health

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American Medical Association

Jennifer Moser, Ph.D.

Department of Veterans Affairs

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Nadeem Sarwar, Ph.D.

Eisai Inc.

Sheri Schully, Ph.D.

NIH Office of Disease Prevention

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Genetic Alliance

Joyce Tung, Ph.D.

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Air Force Medical Support Agency

Michael S. Watson, Ph.D.

American College of Medical Genetics and Genomics

Karen E. Weck, M.D., FCAP

College of American Pathologists

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

National Society of Genetic Counselors

Benjamin Wiegand, Ph.D.

Johnson & Johnson

Huntington F. Willard, Ph.D.

Geisinger National Precision Health

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

American Academy of Nursing

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National Institute of Nursing Research

National Institute on Aging

National Institutes of Health Office of Disease Prevention

National Society of Genetic Counselors

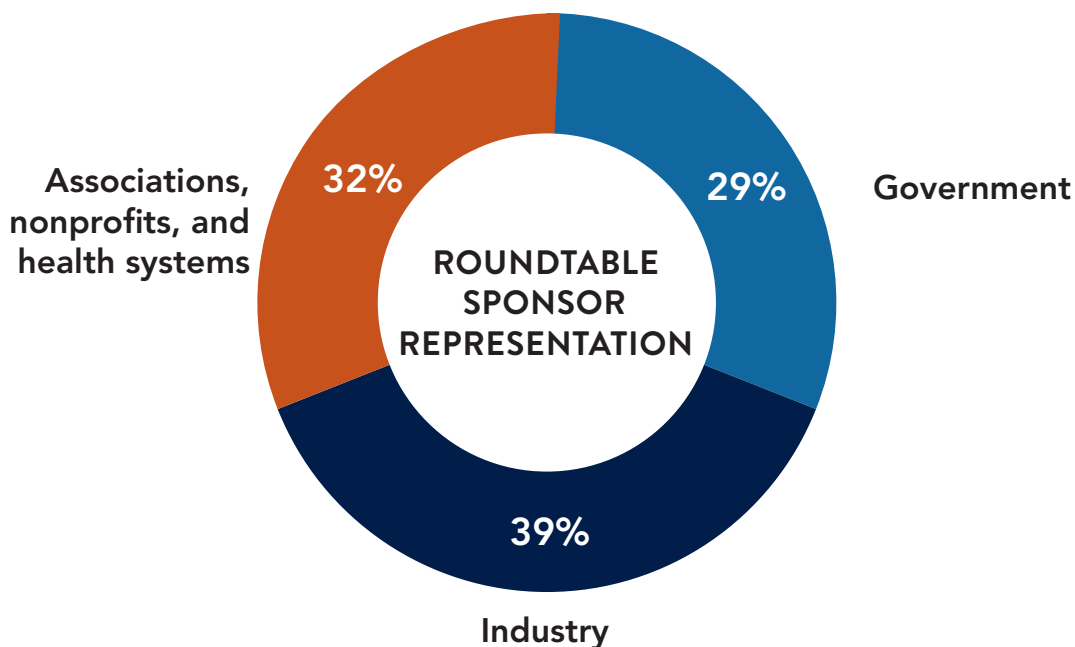
Northrop Grumman

Pfizer Inc.

Regeneron Pharmaceuticals

United States Air Force Medical Service

The University of Vermont Medical Center and The University of Vermont Health Network Medical Group



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

October 5: Virtual Meeting on Return of Research Results

2019

January 23–24: Meeting with Keystone Symposia and American College of Cardiology

March 27–28: Meeting 36

June 25–26: Meeting 37

October 29–30: Meeting 38

Roundtable Staff

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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

Mariam Shelton, M.P.H., CHES, Program Coordinator

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The Health and Medicine Division (HMD), formerly known as the program unit of the Institute of Medicine, is a division of the Academies. HMD's aim is to help those in government and the private sector make informed health decisions by providing evidence upon which they can rely. Each year, more than 3,000 individuals volunteer their time, knowledge, and expertise to advance the nation's health through the work of HMD.

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