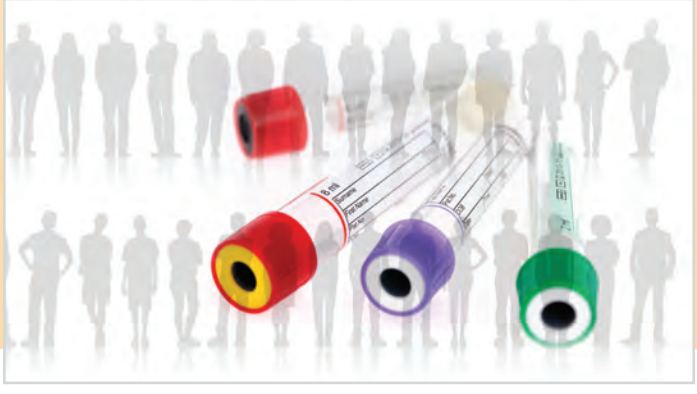


Roundtable on Genomics and Precision Health

2016 Annual Report

Message from the Co-Chairs

Geoffrey S. Ginsburg and Sharon F. Terry

This past year we intentionally reflected on our efforts to date and brought a renewed energy to the issues that face the rapidly evolving field of genomic medicine. We are mindful that our Roundtable began at a time when genetics was rarely integrated into medicine, and so to identify and focus on new opportunities and challenges associated with the implementation of genomics research into clinical care, the Roundtable chose to officially update its name to the Roundtable on Genomics and Precision Health. Along with our updated name came an exciting new mission: to explore strategies for improving health through the implementation of genomics research findings, as well as other related technologies that inform individual health choices, into medicine, public health, education, and policy. During the course of 2016, the Roundtable focused on several exciting and important topics, including mobile health (mHealth) technologies that hold transformative potential for the collection of phenotypic data, the integration of evidence-based genomic applications at the population health level, and large-scale genomic studies that can facilitate drug discovery and development.

The *All of Us*™ cohort, part of the Precision Medicine Initiative (PMI), is one such example of a large-scale, nationwide research effort that aims to expand our understanding of the genetic and environmental influences on human health and disease. The Roundtable members are enthusiastic about this program and are eager to contribute to the policy agenda that will allow this innovative study to lead to health care transformations and therapeutic and diagnostics discoveries. Our Roundtable remains dedicated to addressing issues associated with large-scale genetic studies such as expanding participant engagement, increasing the public's literacy and comfort with genomics research, and helping to shape policies that will reduce health disparities.

One of the primary functions of the Roundtable is to provide an environment of trust for stakeholders from academia, industry, government, foundations, associations, and advocacy organizations to collaboratively identify, highlight, and develop potential solutions for critical scientific and policy issues in research and clinical environments. In this role, the Roundtable fosters robust dialogue, seeks to clarify complex concepts, and identifies pathways to solutions. Since its establishment in 2007, the Roundtable has organized and hosted 30 meetings, 23 public workshops, and published 23 workshop proceedings. In 2016, the Roundtable convened a public workshop that explored novel business models and partnerships that facilitate the use of genetic data in the drug discovery process. As part of an ongoing collaboration with the Forum on Drug Discovery, Development, and Translation, the Roundtable will host a follow-up public workshop in 2017 focused on exploring methods for utilizing genetic information to boost clinical trial enrollment rates, reduce inefficiencies in the current drug development paradigm, and enable precision medicine.

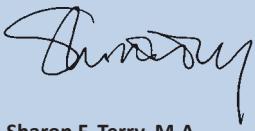
Over the past year, the Roundtable's action collaboratives also made great strides in advancing genomic medicine. The DIGITizE (Displaying and Integrating Genetic Information Through the EHR) action collaborative created an implementation guide for two important pharmacogenomics use cases, and pilot testing of the guide is now under way at several medical centers across the country. In 2016, the Global Genomic Medicine Collaborative (G2MC) was incorporated as a nonprofit corporation, began hosting virtual Grand Rounds on topics in genetics education, and started planning for its third international meeting in Athens, Greece. The G2MC will transition out of the Roundtable in early 2017—a successful sunset of one of the Roundtable's first action collaboratives. Lastly, the Genomics and Population Health action collaborative began compiling information for an online resource guide, which will provide information and support to state public health officials who are interested in launching or expanding genomics programs at the population level.

The Roundtable members will continue to identify key issues in the evidence evaluation process for genomic technologies, approaches to educating health care providers about genomics, integrating genomic information into electronic health records (EHRs), and addressing the challenges of genomic medicine with the global community of stakeholders.

We were very pleased to welcome new sponsoring members to the Roundtable from 23andMe and GeneDx in 2016. We look forward to another productive year with the participation of all stakeholders.

Sincerely,

Geoffrey S. Ginsburg, M.D., Ph.D.
Director, Center for Applied Genomics
& Precision Medicine
Director, MEDx
Duke University



Sharon F. Terry, M.A.
President & CEO
Genetic Alliance

Reflecting Back

Roundtable Activities in 2016

Roundtable Meetings

Roundtable members met three times in 2016 to discuss opportunities and challenges in the fields of genomics and precision health. These meetings brought various stakeholders from government agencies, academic institutions, industry, and professional associations together with the Roundtable members to discuss collaborations that can enable genetically-guided drug development, the implementation of genomics at the population health level, and how to utilize mHealth technologies in genomic medicine to improve health. The Roundtable also convened a public workshop titled Deriving Drug Discovery Value from Large-Scale Genetic Bioresources.

Roundtable Workshop

Deriving Drug Discovery Value from Large-Scale Genetic Bioresources (March 22, 2016)

Given the advances in how genetic data could be used to improve the efficiency of discovery and development of therapies for clinical use, questions remain about how large cohort studies are designed with such objectives in mind, the types of data that should be collected, and which business models could engage stakeholders effectively. The Roundtable on Genomics and Precision Health and the Forum on Drug Discovery, Development, and Translation hosted a workshop that assessed the current landscape of genomic-enabled drug discovery and development activities in industry, academia, and government, examined enabling partnerships and business models, and considered gaps and best practices in how data from populations could be collected with the goal of improving the drug discovery process.

Working Groups

Discovery

While novel technologies and therapies based on genomic information are being implemented in clinical practice, 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 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. They are exploring methods for recruiting patients based on genetics, identifying biomarkers for responders, and focusing on how to garner the types of resources that can enable precision clinical trials.

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 the public health system has arisen. This group will begin to explore the interface between the public health system and health care delivery systems to understand the barriers and challenges that exist for implementing genomics programs at the population health level. Members are interested in understanding communication strategies, data systems, programs, and practices that are currently in place and identifying opportunities to improve collaboration between public health and health care delivery systems.

Mobile Health and Genomics

This group is exploring the landscape at the intersection between mHealth technologies and genomic data by examining how the two fields can interface to enable precision health, advance research (e.g., patient reported outcomes, continuous phenotypes), and improve clinical care (e.g., monitoring, feedback, adherence). The mHealth group is interested in applying what has been learned from the consumer genomics space (e.g., regarding consumer access, consumer comprehension, regulatory jurisdiction, health care provider readiness, clinical and personal utility, potential burden on the health system, and quality standards) to the developing field

of mHealth for more widespread applications for precision medicine. Other areas that may be explored by the group include the role of mHealth platforms as a possible mechanism to link disparate data sources and information that is not currently in the EHR as well as ensuring that the 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 mHealth to illuminate areas of synergy and to inform one another about successes and challenges encountered in each sector.

Overview Group

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

Looking Forward

Roundtable Activities in 2017

Roundtable Meetings

Roundtable members will meet three times in 2017 to discuss major initiatives and areas of focus for future activities. These discussions will examine current issues in genomics and precision health, such as recruiting genetically-identified patients for clinical trials to enable drug development, best practices for implementing genomics in public health systems, and the current landscape of mHealth technologies and how these can be used in conjuction with genomics. In addition, the Roundtable will continue to discuss policy and implementation challenges surrounding global applications of genomic medicine, integrating genetics and genomics into the EHR, and enabling a new drug development paradigm through genomics.

Action Collaboratives

The Action Collaborative on ***Displaying and Integrating Genetic Information Through the EHR (DIGITizE)***, convenes key stakeholders from health information technology (IT) 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 EHRs in a standards-based format. An implementation guide containing the message structures and clinical decision support rules for pharmacogenomic use cases was finalized at the end of 2015 in preparation for pilot programs, which began in 2016. Pilot participants include Boston Children's Hospital, Duke University Hospital, Intermountain Healthcare, Johns Hopkins University Hospital, Mission Health, Partners HealthCare, St. Jude Children's Research Hospital, and University of Utah Hospital. The new use case group is exploring a use case for familial hypercholesterolemia for further phases of the pilot projects in conjunction with a collaborative partner.

The ***Global Genomic Medicine Collaborative*** (G2MC) was launched in 2014 and was incorporated as a 501(c)3 nonprofit organization in June 2016. Since that time, G2MC has hosted two international meetings, bringing together 25 countries to work toward creating a global toolbox for genomic medicine implementation, facilitating collaborations that could enable effective implementation, and discussing solutions for obstacles encountered during implementation. As a result of the collaborative's work, participants have published papers in journals such as *Science Translational Medicine* and began hosting

virtual Grand Rounds on topics related to genetics education. G2MC currently has six working groups including IT/Bioinformatics, Education, Pharmacogenomics, Evidence, Policy, and Sequencing Projects and is planning a third international meeting in Athens, Greece, in 2017.

A ***Genomics and Population Health Action Collaborative*** was initiated at the end of 2015 to explore opportunities for genomics to be used to improve health and prevent disease and death; 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. As an activity of the Roundtable, the Action Collaborative on Genomics and Population Health is seeking to develop methods for assessing best practices and outcomes for implementation of genomic applications and their impact in practice; develop a framework for tools and education materials to increase uptake of genomic applications in health care and disease prevention (such as a plan for a public health genomics toolkit); inform and engage providers, policy makers, and the general public; and explore pilot projects for implementation of genomic applications at the interface of public health and health care that may focus on surveillance and measurement, policy impact, education, and programs.

Roundtable Members

(as of December 2016)

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American Heart Association

Thomas Lehner, Ph.D., M.P.H.
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Laura Nisenbaum, Ph.D.
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Roundtable Sponsors

(as of December 2016)

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National Institute of Nursing Research
National Institute on Aging
National Society of Genetic Counselors
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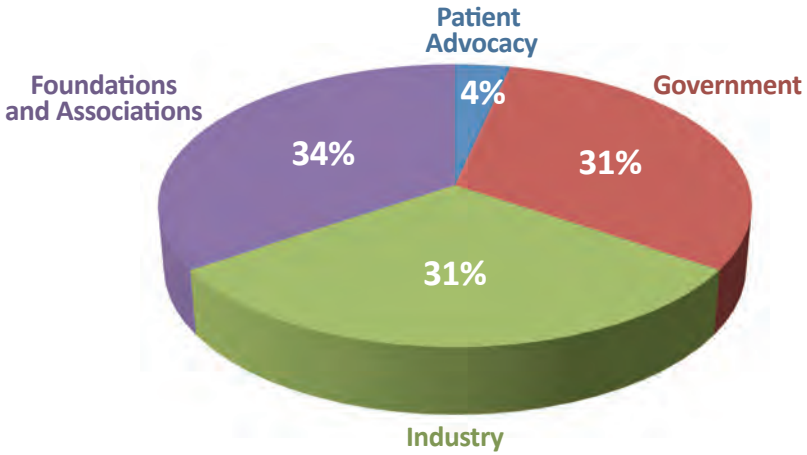


Timeline

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*** | Exploring the Integration of Genomics into Population-Level Health Programs | ***November 2*** Meeting 33

Reports Released in 2016

Applying an Implementation Science Approach to Genomic Medicine: Workshop Summary

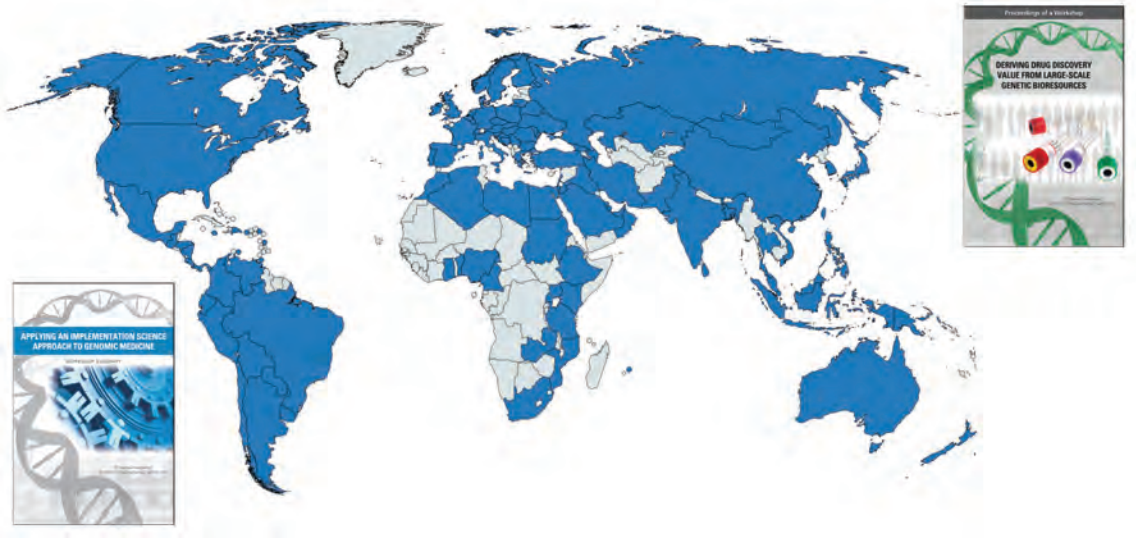
Deriving Drug Discovery Value from Large-Scale Genetic Bioresources: Proceedings of a Workshop

2016 Selected Impacts

Publications

Deriving Drug Discovery Value from Large-Scale Genetic Bioresources: Proceedings of a Workshop
Individuals who downloaded the proceedings indicated that they will use the publication for informing strategic planning for internal programs and potential collaborations, crafting and evolving an open source platform, accelerating health care innovation, and supporting policy development.

Applying an Implementation Science Approach to Genomic Medicine: Workshop Summary
170 individuals who downloaded the workshop summary indicated that they will use the publication for research purposes. More than 20 individuals who downloaded the workshop summary indicated that they will use the publication to inform their efforts in implementing genomics programs.



2016 Roundtable publications have been downloaded in more than 110 countries, for a total of more than 4,600 times.

Collaboratives and Membership

Global Genomic Medicine Collaborative (G2MC)
G2MC hosted eight Grand Rounds on topics related to implementation and genomic programs including pharmacogenomics initiatives in Thailand, using precision medicine to improve risk assessment, the role of genomics in public health, educational technologies and genetic counseling in population genetic screening, and tools for delivering online genetics education resources.

Genomics and Population Health Action Collaborative
Participants in this collaboration developed components for an online toolkit related to horizon scanning and implementation outcome measures. The online toolkit will be used as a resource for public health programs interested in integrating genomics.

Displaying and Integrating Genomics Through the EHR (DIGITizE)
The DIGITizE action collaborative began pilot implementation projects at eight institutions nationwide. The implementation guide created by participants was mentioned in a 2016 NIH Precision Medicine Initiative Funding Opportunity Announcement as "emerging standards to facilitate data exchange and analysis."

About the Genomics Roundtable

The National Academies of Sciences, Engineering, and Medicine established a Roundtable on Genomics and Precision 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.

For more information about the Roundtable on Genomics and Precision Health, please visit our website at nas.edu/genomicsRT or call Sarah Beachy at (202) 334-2217.

About the National Academies of Sciences, Engineering, and Medicine

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.

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