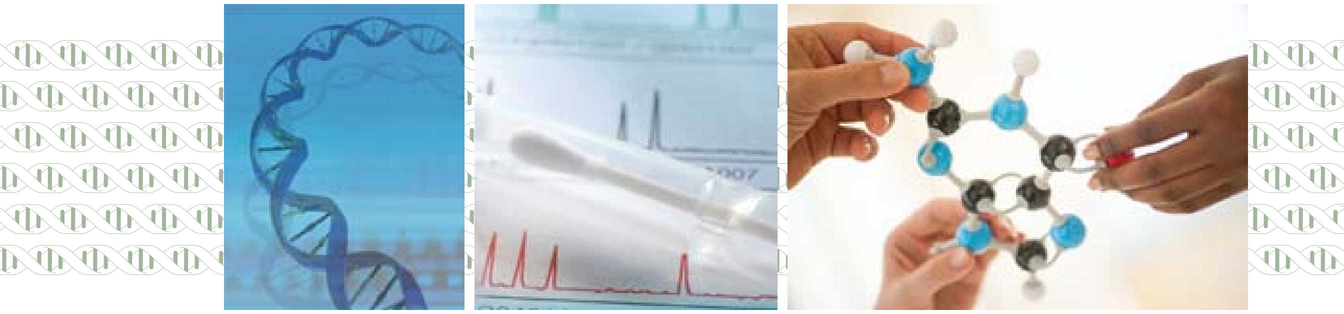
BOARD ON HEALTH SCIENCES POLICY

Roundtable on Translating Genomic-Based Research for Health

2010 Annual Report



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Message from the Chair

Over the past decade, remarkable progress has been made through advances in genomics, from the identification of genes associated with disease processes to the development of pharmacogenetic tests which can minimize adverse side effects and increase treatment efficacy. These discoveries reveal the significant potential for benefit from this rapidly growing area of science—however, the pathway from discovery to health benefit is still unclear for much of genome research and many questions remain. How can the discovery of gene variants or linkages between diseases and biological pathways be leveraged to improve treatment or prevention? What are ethically sound procedures for developing biobanks or using existing samples to address a new emerging question? What research infrastructures are needed to expedite genomics translation and how should they be funded?

The Roundtable on Translating Genomic-Based Research for Health offers a unique venue for experts from academia, industry, patient and provider groups, government, and others to collaborate in addressing these challenging issues in the translation of genomics-based research findings. Our goal is to stimulate honest and frank discussion among members and outside experts, to clarify barriers to effective translation, and promote partnerships that will expedite the benefits of genomic research.

During this past year, the Roundtable sponsored four workshops and three separate discussions which have addressed key questions in the pathway of translating genomic discoveries. Each activity has brought together diverse stakeholder views and explored controversies and common ground as well as potential solutions for advancing the field. We have an ambitious agenda for 2011 which includes examinations of issues such as the economics of personalized medicine, health information technology and genomics, education of the healthcare workforce, and the implementation of industrial scale genomic information into clinical practice. We look forward to another year where the Roundtable can help progress the field toward realizing the promise of genomics.

Sincerely,

Wylie Burke

Wylie Burke, MD, PhD

Professor and Chair
Department of Bioethics & Humanities
University of Washington

Roundtable Activities in 2010

Roundtable Meetings

The Roundtable met three times in 2010 to discuss various issues related to the translation of genomics into health care advances. Topics which were discussed included impediments to bringing basic research discoveries into clinical trials, bioethical considerations surrounding the translation of genomics, and funding challenges and solutions for progressing innovative translational research.

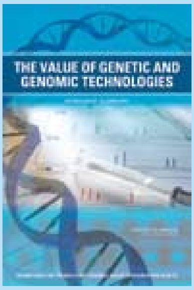
The Value of Genetic and Genomic Technologies: A Workshop (March 2010)

Different stakeholders have very different views on what they require to integrate genomic technologies into their health care decisions. The Roundtable held a workshop to examine what would be needed to compel an individual to utilize a genomic technology if one were available. Some of the issues raised at this workshop included the available research infrastructure and funding for studies to produce the evidentiary basis for clinical utility; the need for an adequate evidence base for clinical and public health decision-making; and the requirements for implementing genetic and genomic technologies into clinical practice. A summary of this workshop is available from the Roundtable website.

Challenges and Opportunities in Using Residual Newborn Screening Samples for Translational Research: A Workshop (May 2010)

Over 4 million infants a year are screened for serious or life-threatening treatable disorders in the United States through the collection and testing of blood samples shortly after birth. Individual states store these samples not only for use in newborn screening, but also for quality control of current tests, the development of new tests, forensic studies, and epidemiological purposes. The Roundtable hosted a workshop to examine the need to balance the public health benefits of using these samples for research purposes with the ethical, legal, and social issues surrounding such use by ensuring that different stakeholder perspectives were identified and considered. This workshop helped inform the report on the use of dried blood spots to the Secretary of Health and Human Services from the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. A summary of this workshop is available from the Roundtable website.

New Publications



The Value of Genetic and Genomic Technologies: Workshop Summary (2010)

Challenges and Opportunities in Using Residual Newborn Screening Samples for Translational Research: Workshop Summary (2010)

Establishing Precompetitive Collaborations to Stimulate Genomics Driven Product Development: Workshop Summary (2010)

Establishing Precompetitive Collaborations to Stimulate Genomics-Driven Product Development: A Workshop (July 2010)

Biological specimens stored by investigators in industry and academia provide a significant untapped resource of genetic and genomic information that can be used to develop individualized treatment regimens or even drug and diagnostic devices. Having human specimens available from well-designed clinical studies for precompetitive use can offer substantial benefits to all parties; however, with this type of sharing a number of issues have to be overcome. The Roundtable hosted a workshop that performed an in-depth examination of the issues that are currently preventing the precompetitive sharing of resources with a goal to facilitate and enable precompetitive collaborations. Discussions addressed specific examples from other industries, examined best practices that make collaboration work, and sought to develop a framework for advancing these partnerships. A summary of this workshop will be available from the Roundtable website.

Evidence Generation for Genomic Diagnostic Test Development: A Workshop (November 2010)

Since sequencing the human genome, scientists have discovered a number of links between genetic variations and disease, and have developed genomic and genetic tests based on these findings. However, clinical practitioners have yet to employ many of these tests as there is a lack of evidence of clinical utility. The Roundtable hosted a workshop to explore current approaches to, new models for, and barriers to generating evidence as well as potential solutions to overcome these obstacles. A summary of this workshop will be available from the Roundtable website.

Roundtable Activities in 2011

Roundtable Meetings

The Roundtable members will meet in February, July, and November of 2011 to continue their discussions on emerging and enduring issues to advance the field of genomics. Meeting topics currently include the economics of genomics and personalized medicine, health information technology and genomics, and workforce and education issues surrounding the application of genomics to clinical practice.

The Implementation of Industrial Scale Genomic Information into Clinical Practice—The Intersection of Bioinformatics and Clinical Genomics: A Workshop (July 2011)

The integration of the large amount of patient-specific data that are generated through genetic and genomic analyses into clinical practice will pose a number of questions and challenges for the current health care system. The Roundtable will host a workshop to examine issues related to the implementation of these technologies including the bioinformatics needs for proper evaluation and re-evaluation of clinically relevant data, cross communication abilities of the bioinformatics and clinical genetics workforce, the addition and interpretation of external factors such as epigenetics and environmental influences on the genome, and ethical issues such as informed consent and stewardship over this information.

Working Groups

The Roundtable has formed four working groups which focus on various aspects of translating genomic advances into health. Members of each group meet regularly by teleconference as well as in-person at Roundtable meetings to converse about matters of mutual interest. Potential workshop, seminar, or commissioned paper topics are also developed by each group and presented to the full Roundtable membership for discussion.

Clinical Practice and Public Health

This working group was formed to focus on issues related to the application of genetic and genomic technologies to clinical and public health practice. Members examine issues such as research and infrastructure requirements, service delivery and clinical decision-making needs, and workforce concerns to develop and apply genomic advances in health care settings. The group is currently developing a workshop aimed at understanding and preparing for the integration of data from technologies such as whole genome sequencing in a clinically relevant manner.

Drug Development Informed by Genetics and Genomics

The Roundtable established a working group to examine the translation of genomic knowledge into drug treatment. Topics of discussion include advancing pharmacogenomics, the sharing of biological samples and associated data, and bridging the "valley of death" in drug development. This group is currently following-up on efforts begun during the Roundtable's July 2010 workshop to determine the necessary framework for using precompetitive collaborations to speed the development of new drugs.

Diagnostic Applications

The Diagnostic Applications working group discusses a variety of issues related to the development and application of genomics and genetics based diagnostic, prognostic, predictive, and population screening tests. Members have discussed issues ranging from the development of new model pathways of moving discoveries to the clinic to the evidence gap for genomic tests.

Emerging Issues

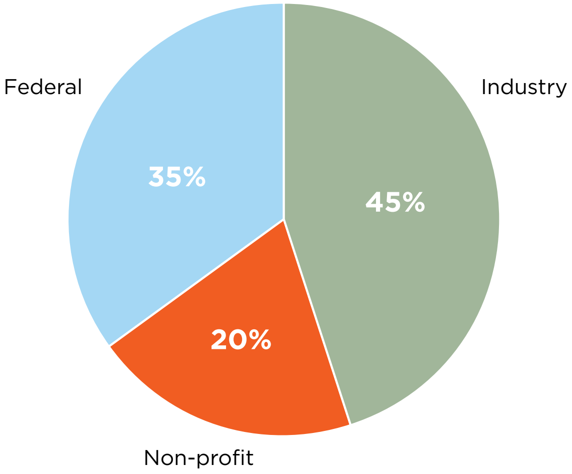
This working group was established by the Roundtable in order to identify, examine, and quickly respond to timely issues of significant importance where the input of the Roundtable would be a valuable asset to the larger public discussion. Members meet to discuss potential topics and have developed activities such as the May 2010 workshop on the use of residual newborn screening samples for translational research. Members will continue to identify, prioritize, and engage on issues as they arise.

Sponsors

(as of December 2010)

American College of Medical Genetics
American Medical Association
American Nurses Association
Blue Cross and Blue Shield Association
Centers for Disease Control and Prevention
College of American Pathologists
Department of the Air Force
Department of Veterans Affairs
Eli Lilly and Company
Genetic Alliance
Health Resources and Services Administration
Johnson & Johnson
Kaiser Permanente
National Cancer Institute
National Heart, Lung, and Blood Institute
National Human Genome Research Institute
National Society of Genetic Counselors
Pfizer Inc.

2010 Roundtable Funding



Roundtable Members

(as of December 2010)

Wylie Burke, M.D., Ph.D. (Chair)
University of Washington

Naomi Aronson, Ph.D.
Blue Cross and Blue Shield Association

Bruce Blumberg, M.D.
Kaiser Permanente

Denise E. Bonds, M.D., M.P.H.
National Heart, Lung, and Blood Institute

C. Thomas Caskey, M.D., FACP
University of Texas Health Science Center at Houston

Stephen Eck, M.D., Ph.D.
Eli Lilly and Company

W. Gregory Feero, M.D., Ph.D.
National Human Genome Research Institute

Andrew N. Freedman, Ph.D.
National Cancer Institute

Geoffrey Ginsburg, M.D., Ph.D.
Duke University

Sharon Kardia, Ph.D.
University of Michigan School of Public Health

Mohamed Khan, M.D., Ph.D.
American Medical Association

Muin Khoury, M.D., Ph.D.
Centers for Disease Control and Prevention

Debra Leonard, M.D., Ph.D.
College of American Pathologists

Michele Lloyd-Puryear, M.D., Ph.D.
Health Resources and Services Administration

Elizabeth Mansfield, Ph.D.
Food & Drug Administration

Garry Neil, M.D.
Johnson & Johnson

Robert L. Nussbaum, M.D.
University of California San Francisco
School of Medicine

Aidan Power, M.B., B.Ch., M.Sc., M.R.C.Psych.
Pfizer Inc.

Ronald Przygodzki, M.D.
Department of Veterans Affairs

Allen D. Roses, Ph.D.
Duke University

Kevin A. Schulman, M.D.
Duke University

Sharon Terry, M.A.
Genetic Alliance

Martha Turner, Ph.D., R.N., B.C., C.N.A.A.
American Nurses Association

Michael S. Watson, Ph.D.
American College of Medical Genetics

Daniel Wattendorf, M.D. (Lt. Col)
Department of the Air Force

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

About the Roundtable

The Institute of Medicine's Roundtable on Translating Genomic-Based Research for Health brings together leaders from academia, industry, government, foundations, associations, and representatives of patients and consumers who have a mutual interest in addressing the issues surrounding the translation of genomic-based research. The purpose of the Roundtable is to explore and implement strategies for improving health through the translation of genomic and genetic research findings into medicine, public health, education, and policy.

Translating genomic innovations involves many disciplines, and takes place within different economic, social, and cultural contexts, generating a need for increased communication and understanding across these fields. Furthermore, these innovations have produced a diversity of new issues to be addressed, including issues such as evidence of utility, economic implications, equal access, and public perspectives. As a convening mechanism for interested parties with different perspectives to meet and discuss complex issues of mutual concern in a neutral setting, the Roundtable fosters dialogue across sectors and institutions and fosters collaboration among stakeholders.

The priorities and areas of emphasis for the Roundtable include: 1) issues related to the translation of genomics into medicine and public health; 2) issues related to the evolving requirements for the health professional community, and the need to be able to understand and responsibly apply genomics to medicine and public health; and 3) ethical, legal, and social issues such as the potential for misuse of genetic information; the medical implications for family members; and the rights of an individual, family, or community to control the use and dissemination of genetic information. To achieve its objectives, the Roundtable conducts structured discussions, workshops, and symposia, and publishes workshop summaries. Specific issues and agenda topics are determined by the Roundtable membership and span a broad range of issues relevant to the translation process.

Roundtable Staff

Adam C. Berger, Ph.D.
Project Director

Claire Giammaria, M.P.H.
Research Associate

Tonia Dickerson
Senior Program Assistant

Board on Health Sciences Policy Staff

Andrew Pope, Ph.D.
Director

Contact

For more information about the Roundtable on Translating Genomic-Based Research for Health, please visit our website at www.iom.edu/genomicroundtable or call Adam Berger at (202) 334-3756.

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