

# Roundtable on Translating Genomic-Based Research for Health

2011 Annual Report



INSTITUTE OF MEDICINE

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## About the Genomics 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.

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



### Message from the Chair

Genomic research continues to advance at a rapid pace, offering new possibilities in research and clinical medicine. Yet many questions remain about how to deploy genomics most effectively and how to evaluate the benefits and harms of new genomic technology. 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 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 workshops and meetings addressing several critical areas in the translation of genomics findings, including the potential risks and benefits of whole genome sequencing in clinical care; clarifying regulatory and reimbursement pathways for genomic tests; and the potential for genomic tools to enhance drug discovery. Each activity brought together diverse stakeholder views and explored controversies and common ground as well as potential solutions for advancing the field.

Our agenda for 2012 will include further exploration of these important topics. In addition, we will consider barriers to moving the findings of basic genome science into translational research; challenges in education and public engagement; and innovative approaches to partnership. We look forward to continuing the work of the Roundtable and helping to identify the pathways, strategies, and partnerships that can help progress the field toward the promise of genomics.

Sincerely,

**Wylie Burke, M.D., Ph.D.**

Professor and Chair

Department of Bioethics & Humanities

University of Washington

# Reflecting Back

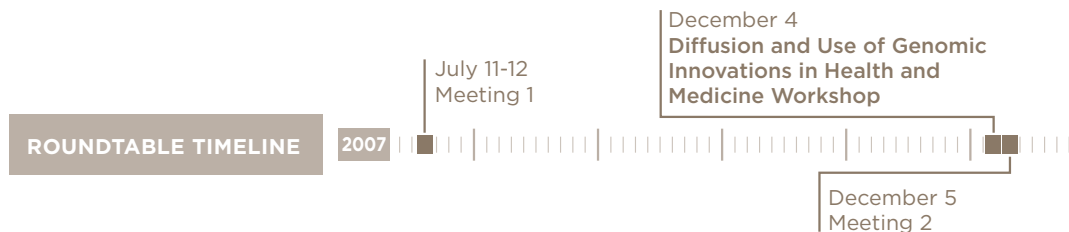
## Roundtable Activities in 2011

### Roundtable Meetings

The Roundtable members met three times in 2011 to continue their discussions on emerging and enduring issues to advance the field of genomics. Meeting topics discussed during the February 22-23 meeting included (1) the economics of individualized medicine; (2) integrating large-scale genomic information into health information technology systems; and (3) the role of interdisciplinary communication in facilitating the use of large-scale genomic information in clinical practice. The topic addressed during the July 20 Roundtable meeting was regulatory science and its role in the translation of research findings and drug and diagnostic device development. The November 16 meeting featured the following three discussions: (1) the Patient-Centered Outcomes Research Institute: A Conversation with Joe Selby; (2) the Presidential Commission for the Study of Bioethical Issues—Genes to Genomes: Collection, Use, and Governance of Human Genome Sequence Data; and (3) ethical concepts in genomics research.

### Integrating Large-Scale Genomic Information into Clinical Practice: A Workshop (July 19, 2011)

As DNA sequencing technology becomes more refined, costs continue to drop, and clinical applications increase for genomic and genetic analyses, the integration of this new technology into practice is inevitable. However, the large amount of patient-specific data that are generated through large-scale genomic analysis pose a number of questions and challenges for integrating this information into the current health care system. Solutions will need to be devised regarding the storage, access, and portability of these genetic data. Relevant information will need to be extracted and deposited into the medical record in a clinically comprehensible manner. The analysis of this information, and re-analysis as new findings emerge, will put further constraints and burdens on health care practitioners in a system that currently lacks a mechanism



to reimburse for these activities. Additionally, there is a lack of understanding and training for patients and health care providers on the potential impact genetic and genomic discoveries can have for treatment course and health. Ethical issues such as informed consent and stewardship over this information will also have to be considered as integration moves forward. With these questions and issues in mind, the Roundtable hosted a workshop to explore and examine potential solutions for integrating large-scale genomic information into clinical practice with respect to the analysis, interpretation, and delivery of genomic information. Stakeholders, including bioinformaticists, clinical investigators, health care providers, patient advocacy groups, payers, policy makers, and public health professionals presented their perspectives.

**Facilitating the Development and Utilization of Genome-Based Diagnostic Technologies: A Workshop (November 15, 2011)**

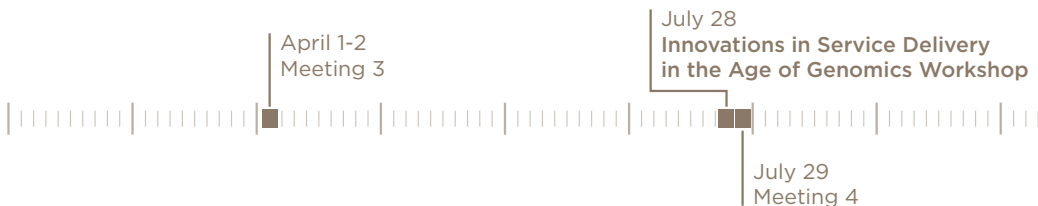
The sequencing of the human genome and the identification of links between specific genetic variants and disease formation have led to an explosion of genome-based diagnostic tests that have the potential to direct therapeutic interventions or predict the onset of disease. However, the current adoption of genomic diagnostic tests into practice by providers has been limited due to a lack of evidence of clinical utility. Thus, health funders and practitioners lack the data necessary to distinguish which tests can improve practice or the clinical settings in which tests will provide the greatest value. In part, this lack of evidence and usage has led to what has been termed a “vicious cycle” of undervaluation, with test developers producing either no or low evidence of clinical utility, followed by limited usage by providers and little reimbursement by payers. This undervaluation by all groups results in limited resources for developers to produce the evidence desired by various stakeholders and a perpetual continuation of the cycle. The Roundtable hosted a workshop to identify challenges and potential opportunities to advance the development and use of genomic diagnostic tests through research, reimbursement, and regulatory policy. Stakeholders, including diagnostic test developers, payers, policy makers, health care providers, patient advocacy groups, investors, and others presented their perspectives.

2008

April 1-2  
Meeting 3

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

July 29  
Meeting 4



# Looking Forward

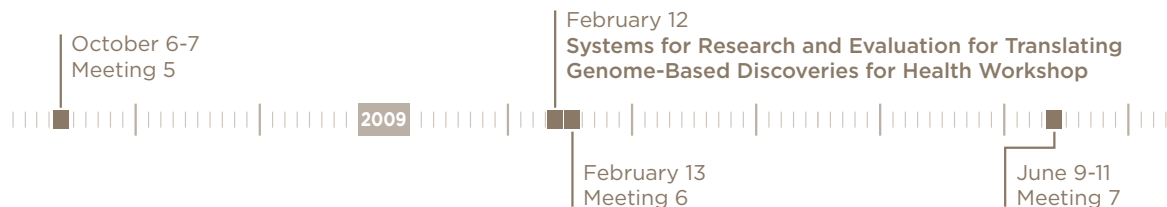
## Roundtable Activities in 2012

### Roundtable Meetings

The Roundtable members will meet on March 22, July 18, and December 4, 2012. Meeting topics to be discussed include ongoing efforts to develop a genetic variant database, aligning basic genomic research for more efficient and effective translation, and challenges for targeted therapeutics development.

### New Paradigms in Drug Discovery: How Genomic Data Are Being Used to Revolutionize the Drug Discovery and Development Process: A Workshop (March 21, 2012)

New drug approvals have remained reasonably steady for the past 50 years while at the same time the total spending on health-related research and development has tripled since 1990. There are many suspected causes for this trend, including increased regulatory barriers, the rising costs of scientific inquiry, and the lack of production models that have successfully incorporated new technology. Regardless of the cause, this trajectory is not economically sustainable, and in response many companies are turning toward collaborative models of drug development. The Roundtable will host a public workshop to investigate how genetic and genomic information is currently being used and will be used in the future to improve the



process of drug discovery and development. The goal of the workshop is to examine the current investment in, use, and utility of genetic and genomic data to improve these processes. The presentations and discussions will explore various aspects of the issue, including target identification, clinical trial design, strategic partnerships, and the use of emerging technologies, such as next generation sequencing. The workshop will also investigate the economic drivers, incentives, and models that use genomics in drug development and will feature an array of stakeholders, including leaders from academia, industry, and governmental organizations.

#### **Assessing the Economics of Genomic Medicine: A Workshop (July 17-18, 2012)**

The sequencing of the human genome and the identification of links between specific genetic variants and disease formation have led to an explosion of genomics-based innovation and technology and the advent of genomic medicine. These advancements have the ability to direct patient treatment toward more effective or less harmful options and potentially to reduce health care costs caused by unnecessary or ineffective treatments. However, this is not a certainty and some stakeholders are concerned that genomic technologies will simply be cost additive to the health care system without providing significant benefit to patients. In order to better forecast the costs and benefits of integrating genetic and genomic information into clinical practice, the Roundtable will host a workshop that will bring economists, regulators, payers, and providers together to analyze three scenarios—preconception care, a deep vein thrombosis and pulmonary embolism event, and directing cancer care—where genomic information can be utilized for clinical decision making. The various factors that influence costs will be identified and discussed along with alternative models of implementation. The goal of the workshop will be to advance discussions around the clinical integration of genomic applications.

#### **Improving the Efficiency and Effectiveness of Genomic Science Translation: A Workshop (December 3, 2012)**

The anticipated health care developments that were predicted at the completion of the Human Genome Project have not, for the most part, culminated in products or processes that are used in medical settings. This lack of translation between basic genome science and clinical research has frustrated many scientists, health care practitioners, investors, policy makers, and patients. Recognizing the growing divide among these sectors, the Roundtable will hold a workshop to examine how basic science can best be positioned to foster successful translation of early discoveries.

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

2010

November 16-17  
Meeting 8

March 22  
The Value of Genetic and Genomic  
Technologies Workshop

March 23  
Meeting 9



# Working Groups

Four working groups have been established within the Roundtable to examine various topics and aspects of translating genomic information into appreciable health care applications. Each group meets regularly by teleconference as well as in person at Roundtable meetings. Together they discuss issues of mutual interest to the group as well as suggest, plan, and complete Roundtable workshops, seminars, and commissioned papers.

## Clinical Practice and Public Health

This working group focuses on issues related to the development and implementation of genetic and genomic technologies in clinical and public health practice. Members examine issues such as the economic impact of genomic medicine, the educational needs of the health care workforce, and the intersection between bioinformatics and clinical practice. The group is currently planning a workshop to examine the various factors that influence the economics of integrating genetic and genomic information into clinical practice so that benefits can be maximized.





### Drug Development Informed by Genetics and Genomics

The Roundtable established a working group to examine the drug discovery and development process. This group focuses on topics such as using collaborative research paradigms to advance drug development, data sharing, biospecimen resources, and genomic and genetic strategies for drug discovery and development. The group is currently planning a workshop to investigate the economic drivers, incentives, and models that use genomics in drug development.

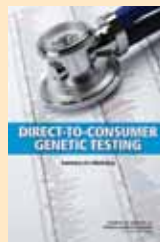
### Diagnostic Applications

The Roundtable established the Diagnostic Applications working group to discuss issues related to the development and use of molecular diagnostics. This group has focused its discussions on three key elements in the development and adoption of clinically useful genome-based diagnostic applications: evidence gaps, regulatory policy, and reimbursement for genomic tests.

### Overview Group

This working group was established by the Roundtable to examine issues that are overarching and crosscutting for all the members and groups. Members also coordinate timely responses to emerging issues within the field where input from the Roundtable would be of significant value to larger public discussions. Currently, the members of this group are planning the December 2012 workshop that will attempt to identify how the translational process between basic genome science and medicine can be improved.

## Reports Released in 2011



**Direct-to-Consumer Genetic Testing: Summary of a Workshop**



**Generating Evidence for Genomic Diagnostic Test Development: Workshop Summary**



**Integrating Large-Scale Genomic Information into Clinical Practice: Workshop Summary**

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

# Roundtable Members

(as of December 2011)

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

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

**Paul R. Billings, M.D., Ph.D.**  
Life Technologies Corporation

**Bruce Blumberg, M.D.**  
Kaiser Permanente

**Denise E. Bonds, M.D., M.P.H.**  
National Heart, Lung, and  
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**C. Thomas Caskey, M.D., FACP**  
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**Sara Copeland, M.D.**  
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**W. Gregory Feero, M.D., Ph.D.**  
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**Andrew N. Freedman, Ph.D.**  
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**Richard Hodes, M.D.**  
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**Sharon Kardia, Ph.D.**  
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**Mohamed Khan, M.D., Ph.D.**  
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**Debra Leonard, M.D., Ph.D.**  
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Office of Rare Diseases Research

**Elizabeth Mansfield, Ph.D.**  
Food and Drug Administration

**Garry Neil, M.D.**  
Johnson & Johnson

**Robert L. Nussbaum, M.D.**  
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School of Medicine

**Michelle Ann Penny, Ph.D.**  
Eli Lilly and Company

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

**Victoria M. Pratt, Ph.D., FACMG**  
Quest Diagnostics Nichols Institute

**Ronald Przygodzki, M.D.**  
Department of Veterans Affairs

**Allen D. Roses, M.D.**  
Duke University

**Kevin A. Schulman, M.D.**  
Duke University

**Joan A. Scott, M.S., C.G.C.**  
National Coalition for Health  
Professional Education in Genetics

**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 and Genomics

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

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

## Roundtable Staff

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**Sean David, M.D., D.Phil.**  
James C. Puffer, M.D./American Board  
of Family Medicine Fellow, Stanford  
University School of Medicine

**Sarah H. Beachy, Ph.D.**  
Christine Mirzayan Science and  
Technology Policy Graduate Fellow  
(August to November 2011)

**Claire Giammaria, M.P.H.**  
Research Associate

**Tonia Dickerson**  
Senior Program Assistant

## Board on Health Sciences Policy Staff

**Andrew M. Pope, Ph.D.**  
Director

March 21

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

July 17-18

**Assessing the Economics of  
Genomic Medicine Workshop**

December 3

**Improving the Efficiency and  
Effectiveness of Genomic Science  
Translation Workshop**

2013

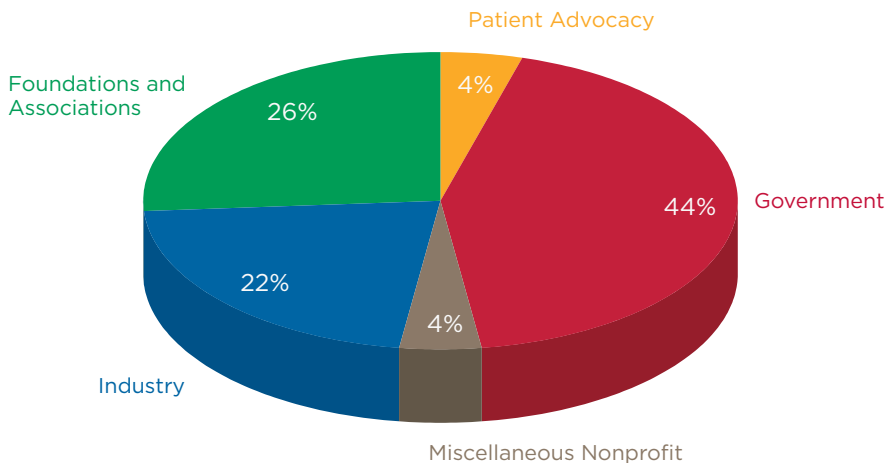
March 22  
Meeting 16

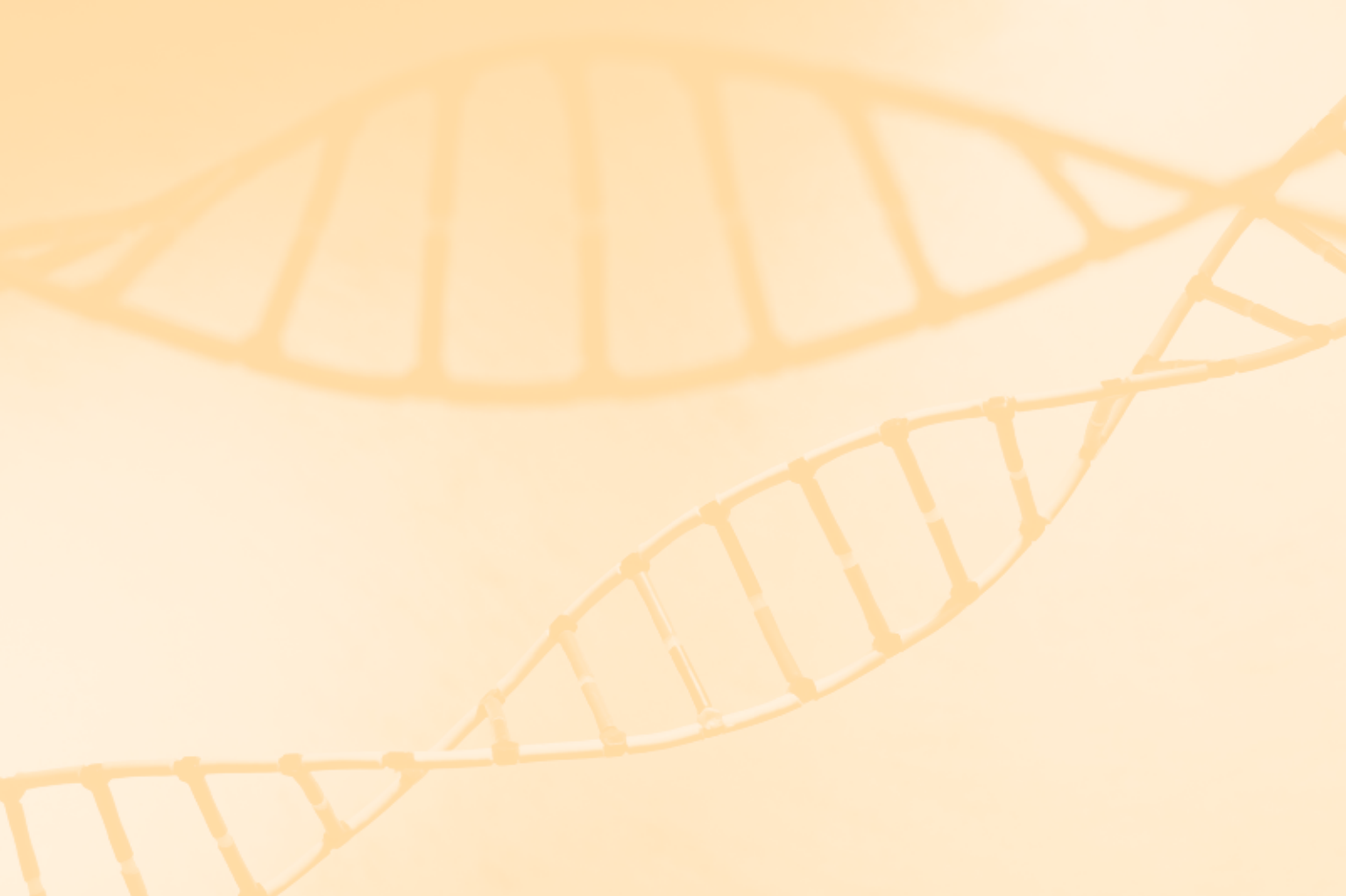
July 18  
Meeting 17

December 4  
Meeting 18

# Roundtable Sponsors

- American College of Medical Genetics and Genomics
- 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
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- Life Technologies Corporation
- National Cancer Institute
- National Coalition for Health Professional Education in Genetics
- National Heart, Lung, and Blood Institute
- National Human Genome Research Institute
- National Institute of Mental Health
- National Institute on Aging
- National Society of Genetic Counselors
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