Diffusion and Use of Genomic Innovations in Health and Medicine: Workshop Summary

Lyla M. Hernandez, Rapporteur, Roundtable on Translating Genomic-Based Research for Health

ISBN: 0-309-11677-5, 116 pages, 6 x 9, (2008)

DIFFUSION AND USE OF **GENOMIC INNOVATIONS** IN HEALTH AND MEDICINE

WORKSHOP SUMMARY

Lyla M. Hernandez, *Rapporteur*

Roundtable on Translating Genomic-Based Research for Health

Board on Health Sciences Policy

INSTITUTE OF MEDICINE
OF THE NATIONAL ACADEMIES

THE NATIONAL ACADEMIES PRESS
Washington, D.C.
www.nap.edu

THE NATIONAL ACADEMIES PRESS 500 Fifth Street, N.W. Washington, DC 20001

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This project was supported by contracts between the National Academy of Sciences and American College of Medical Genetics (Unnumbered contract); American College of Physicians (Unnumbered contract); American Medical Association (Unnumbered contract); AstraZeneca Pharmaceuticals, Inc. (Unnumbered contract); Blue Cross/Shield Association (Unnumbered contract); Centers for Disease Control and Prevention (CDC) (Contract No. 200-2005-13434); College of American Pathologists (Unnumbered contract); Department of Veterans Affairs (VA) (Contract No. V101(93) P-2238); Eli Lilly and Company (Contract No. LRL-0028-07); Food & Drug Administration (Contract No. 223012460); Genetic Alliance (Unnumbered contract); Genomics Health, Inc. (Unnumbered contract); GlaxoSmithKline, Inc. (Unnumbered contract); Health Systems Research, Inc. (Contract No. 07-H0116); National Human Genome Research Institute (Contract No. N01-OD-4-2139, TO#189); National Institute of Child Health and Human Development (Contract No. N01-OD-4-2139, TO#189); National Society of Genetic Counselors (Unnumbered contract); Secretary's Advisory Committee on Genetics, Health and Society (Contract No. N01-OD-4-2139, TO#189); and United Health Care (Unnumbered contract). Any opinions, findings, conclusions, or recommendations expressed in this publication are those of the author(s) and do not necessarily reflect the views of the organizations or agencies that provided support for the project.

International Standard Book Number-13: 978-0-309-11676-3
International Standard Book Number-10: 0-309-11676-7

Additional copies of this report are available from the National Academies Press, 500 Fifth Street, N.W., Lockbox 285, Washington, DC 20055; (800) 624-6242 or (202) 334-3313 (in the Washington metropolitan area); Internet, http://www.nap.edu.

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Suggested citation: IOM (Institute of Medicine). 2008. *Diffusion and use of genomic innovations in health and medicine: Workshop summary.* Washington, DC: The National Academies Press.

"Knowing is not enough; we must apply.
Willing is not enough; we must do."
—Goethe

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This report has been reviewed in draft form by individuals chosen for their diverse perspectives and technical expertise, in accordance with procedures approved by the National Research Council's Report Review Committee. The purpose of this independent review is to provide candid and critical comments that will assist the institution in making its published report as sound as possible and to ensure that the report meets institutional standards for objectivity, evidence, and responsiveness to the study charge. The review comments and draft manuscript remain confidential to protect the integrity of the deliberative process. We wish to thank the following individuals for their review of this report:

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Although the reviewers listed above have provided many constructive comments and suggestions, they were not asked to endorse the final draft of the report before its release. The review of this report was overseen by

Bradford H. Gray, Ph.D., Editor, *The Milbank Quarterly,* Principal Research Associate, The Urban Institute. Appointed by the Institute of Medicine, he was responsible for making certain that an independent examination of this report was carried out in accordance with institutional procedures and that all review comments were carefully considered. Responsibility for the final content of this report rests entirely with the author and the institution.

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The sequencing of the human genome has generated excitement about the potential of genomic innovations to improve medical care, preventive and community health services, and public health. Until fairly recently, genetic information was used primarily in the diagnosis of relatively rare genetic diseases, such as cystic fibrosis and Huntington's Disease, but a transformation in the use of genetic and genomic information is under way.

Genetic markers of increased risk for such chronic diseases as diabetes and coronary artery disease have been identified. Research on how genes influence the effects of drugs holds promise for helping physicians individualize drug therapy. Tests designed to help providers make treatment decisions based on variations in a patient's genome are being developed. The Department of Health and Human Services has launched a Personalized Health Care Initiative, one goal of which is to "link clinical and genomic information to support personalized health care"[2] (DHHS, 2007). It is anticipated that "genetic prediction of individual risks of disease and responsiveness to drugs will reach the medical mainstream in the next decade or so" (Collins and McKusick, 2001). To date, however, few of these promising discoveries have resulted in actual applications in medicine and health (Burke et al., 2006).

[1]The planning committee's role was limited to planning the workshop, and the workshop summary has been prepared by the workshop rapporteur as a factual summary of what occurred at the workshop.

[2]Personalized health care, as defined by the Department of Health and Human Services, refers to medical practices that are targeted at individuals based on their specific genetic code in order to provide a tailored approach (www.hhs.gov/myhealthcare/glossary/glossary.html).

In 2007 the Institute of Medicine established the Roundtable on Translating Genomic-Based Research for Health. The purpose of the Roundtable is to foster dialogue and discussion that will advance the field of genomics and improve the translation of research findings to health care, public health, and health policy. As a first step in examining issues of translation of genomic innovations, the Roundtable decided to hold a workshop to gather information on three questions below. Information obtained from the workshop was then used to further discussion and exploration of the answers to these questions:

1. Are there different pathways by which new scientific findings move from the research setting into health care?
2. If so, what are the implications of those different pathways for genomics?
3. What can we learn from the translation of other new technologies as we seek to understand the translation of genome science into health care?

The December 4, 2007, workshop was moderated by Wylie Burke, chair of the Roundtable, and consisted of panel presentations in four areas: the process of translation of innovations, practical incentives and barriers to translation, translation of genomic technology at the clinical level, and opportunities and constraints for translation both within the United States and globally. A discussion period followed each panel. At the conclusion of the meeting Burke offered a summary of the day's presentations. While various types of genomic innovation were discussed, a number of presentations focused primarily on genomic testing technologies. The complete agenda can be found in Appendix A, and biographical sketches of the speakers are in Appendix B.

The following report summarizes speaker presentations and discussions. Any conclusions reported should not be construed as reflecting a group consensus, rather they are the statements and opinions of presenters and participants.

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Translation of Innovations

A BROAD PERSPECTIVE

Robert M. Califf, M.D., MACC
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Biomedical science is advancing at an amazing rate, yet the translation of that science into better health outcomes has not kept pace. Much of this lag is due to non-technological reasons, including financing, regulation, and cultural issues. Another factor is that the rewards for researchers who promote innovation are increasingly disconnected with the healthcare needs of society at large.

Translation is a fragmented and uncoordinated process that, with few exceptions, takes 25 to 30 years from initial scientific discovery to the delivery of a therapy to the people who benefit most (Figure 2-1). While basic discoveries occur predominantly in academic medical centers funded by the National Institutes of Health (NIH), the process of translating these discoveries almost always begins in the medical products industry, where a basic discovery is followed up with a period of specifically directed preclinical activity intended to test whether the putative therapeutic target is indeed viable. The next step is determined by a decision-making process that comprises multiple steps and includes assessments that link financial support with the probability of success; if the decision is to move forward, then the next stage of development is undertaken by clinical research orga-