Integrating Large-Scale Genomic Information into Clinical Practice

July 19, 2011

The Keck Center of the National Academies
500 Fifth St., N.W.
Washington, DC 20001

Workshop Objective:

- To highlight and identify the challenges and opportunities in integrating large-scale genomic information into clinical practice.

Workshop Assumptions:

- Sequencing technology will advance enough to produce clinically meaningful results.
- Whole genome sequencing (WGS) will be cost-effective and comparable to other diagnostic tests.

7:45 A.M. **Working Breakfast**

8:30 A.M. **Welcoming Remarks**

Wylie Burke, *Roundtable Chair*
Professor and Chair
Department of Bioethics and Humanities
University of Washington

8:35 A.M. **Charge to Workshop Speakers and Participants**

Bruce Blumberg, *Workshop Co-Chair*
Institutional Director of Graduate Medical Education
Northern California Kaiser Permanente
The Permanente Medical Group

Catherine A. Wicklund, *Workshop Co-Chair*
Director, Graduate Program in Genetic Counseling
Past President, National Society of Genetic Counselors
Associate Professor, Department of Obstetrics and Gynecology
Northwestern University

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8:45 A.M. **Keynote Address: The Realization of Genomic Medicine**

Leslie G. Biesecker
Chief and Senior Investigator, Genetic Disease Research Branch
National Human Genome Research Institute

SESSION I: WORKFORCE

Session Focus:

- What are the necessary knowledge and skill sets required for analyzing, interpreting, and utilizing genomic information? From a laboratory perspective? From a clinical perspective?
- Given the range of skills needed to turn genomic information into clinically actionable medical practice, what are the training needs for an individual and what is not being addressed?
- What is needed to translate genomic information from the lab to the provider? Will collaborative medicine be needed to interpret genomic information?

Session Moderator: Michael S. Watson, American College of Medical Genetics

9:15 A.M. **Requisite Knowledge and Skill Sets for Large-Scale Genomic Data Utilization**

Bruce R. Korf
Wayne H. and Sara Crews Finley Chair in Medical Genetics
Professor and Chair, Department of Genetics
Director, Heflin Center for Genomic Sciences
University of Alabama at Birmingham

SESSION II: ANALYSIS

Session Focus:

- What are the standards and criteria for updating database information with novel discoveries? Can this be done in a unified approach?
- Will new knowledge be incorporated into previously analyzed data?
- Who will be responsible for re-analysis of genomic data? Where does the responsibility lie for re-contacting patients? What is a model for analysis?

Session Moderator: Debra G. Leonard, Weill Cornell Medical Center

9:45 A.M. **Defining Actionable Novel Discoveries, Annotating Genomes, and Reanalysis – A Laboratory Perspective**

Federico Monzon
Associate Professor of Pathology
Weill Cornell Medical College / The Methodist Hospital
Director, Research Pathology and Molecular Diagnostics Cores
The Methodist Hospital Research Institute

Madhuri Hegde
Associate Professor, Department of Human Genetics
Scientific Director, Emory Genetics Laboratory
Emory University School of Medicine

Defining Actionable Novel Discoveries, Annotating Genomes, and Reanalysis – A Clinical Perspective

Heidi Rehm
Assistant Professor of Pathology, Brigham & Women's Hospital and Harvard Medical School
Chief Laboratory Director, Laboratory for Molecular Medicine
Partners Healthcare Center for Personalized Genetic Medicine

Michael Christman
President and CEO
Coriell Institute for Medical Research

11:05 A.M. **Discussion with Speakers and Attendees**

11:50 A.M. BREAK (collect lunch and return to seats)

SESSION III: INTERPRETATION

Session Focus:

- What will the emerging model of interpretation look like?
- Will there be separate vendors and interpretation engines of WGS data? What is the regulatory environment for private interpretation services? How will results be verified? Who will be the authority to consult for a second opinion?
- How will other clinical data and past medical records be pooled to make these interpretations more meaningful? How will future (e.g., epigenetic) information be added?
- How can population-based data be correctly applied in an individual patient setting? How will the absence of population-based data affect interpretation?

Session Moderator: Muin Khoury, Centers for Disease Control and Prevention

12:05 P.M. **Interpretation Models and Context**

Medical Practice

Robert Nussbaum
Holly Smith Chair and Professor of Medicine
Chief, Division of Medical Genetics
Member, Institute of Human Genetics
Professor of Neurology
University of California, San Francisco School of Medicine

Pathology

Mark Boguski
Associate Professor, Center for Biomedical Informatics
Harvard Medical School

Bioinformatics

Nicholas J. Schork
Director of Biostatistics and Bioinformatics
Professor, Molecular and Experimental Medicine
The Scripps Translational Science Institute

1:20 P.M. **Discussion with speakers and attendees**

SESSION IV: DELIVERY OF INFORMATION

Session Focus:

- How will the data be shared?
- How will genomic information be delivered at point of care? What is the setting for delivery? What happens to the information once it is delivered?
- What is the accountability for stewarding the information?
- What are the practical issues for integrating genomic information into practice and how can they be overcome? What is the necessary infrastructure and what are the needed procedures?

Session Moderator: W. Gregory Feero, National Human Genome Research Institute

2:15 P.M. **Innovative Mechanisms for Delivery**

Integral Delivery Systems and Active Monitoring of Data

Mary V. Relling
Chair, Pharmaceutical Department
St. Jude Children's Research Hospital

Granting Access to Personal Health Information

Robert H. Shelton
Founder and CEO
Private Access, Inc.

2:55 P.M. **Discussion with Speakers and Attendees**

3:40 P.M. BREAK

SESSION V: ETHICAL AND LEGAL ISSUES

Session Focus:

- How do we protect privacy while maximizing usability of the information? How is an individual's autonomy protected?
- What are the legal issues involved in recontact and delivery of information? What is the liability for holding information from whole genome sequencing which is not acted upon? How can these issues be resolved?
- What are the legal implications and precedence for tracking patients in order to supply updated information as it becomes available? What is the best approach for doing so?
- How do the ethical and legal issues reflect off each other?

Session Moderator: Martha Turner, American Nurses Association

3:55 P.M. **Ethical and Legal Implications for Integrating Large-Scale Genomic Data into Clinical Practice**

Henry T. Greely
Deane F. and Kate Edelman Johnson Professor of Law
Professor, by courtesy, of Genetics
Director, Center for Law and the Biosciences
Stanford University

SESSION VI: PANEL DISCUSSION

Session Focus:

- What is the vision for how this field will emerge? What will the scope of practice look like?
- What will the WGS health partnership look like and how will it function?
- What new technology/information is foreseeable in the near future and what is needed in place to deploy and make use of it?

Session Moderator: Bruce Blumberg, Kaiser Permanente
Catherine A. Wicklund, National Society of Genetic Counselors

Overview and Panel Objectives:

4:25 P.M. **Envisioning the Future**

Panelists

Mark Boguski, Harvard Medical School

Michael Christman, Coriell Institute for Medical Research

Henry T. Greely, Stanford University

Madhuri Hegde, Emory University

Bruce R. Korf, University of Alabama at Birmingham

Federico Monzon, Weill Cornell Medical College / The Methodist Hospital

Robert Nussbaum, University of California, San Francisco School of Medicine

Heidi Rehm, Harvard Medical School

Nicholas J. Schork, The Scripps Translational Science Institute

Robert H. Shelton, Private Access, Inc.

SESSION VII: FINAL REMARKS

5:55 P.M. **Concluding Remarks**

Bruce Blumberg, *Workshop Co-Chair*
Institutional Director of Graduate Medical Education
Northern California Kaiser Permanente
The Permanente Medical Group

Catherine A. Wicklund, *Workshop Co-Chair*
Director, Graduate Program in Genetic Counseling
Past President, National Society of Genetic Counselors
Assistant Professor, Department of Obstetrics and Gynecology
Northwestern University

6:10 P.M. **Adjourn**