National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)

Genes, Environment, and Health Initiative: Translating Whole Genome Association Data into Clinical Practice

Natcher Conference Center, Building 45, National Institutes of Health (NIH)
March 10-11, 2008

Agenda

Monday, March 10, 2008

7:30 a.m. – 8:30 a.m. **Registration**

Session I
Chair: *Muin Khoury,* Centers for Disease Control and Prevention

8:30 a.m. – 10:00 p.m. **Introductions and Presentation of Meeting Format/Goals**
Griffin Rodgers, NIDDK
Brenda Weis, National Institute of Environmental Health Sciences

From GWA to Health Applications: The Promise
Francis Collins, National Human Genome Research Institute

From Health Applications to Population Health Impact: The Translation Challenge
Muin Khoury, Centers for Disease Control and Prevention

10:00 a.m. – 10:30 a.m. **Break**

10:30 a.m. – 12:00 p.m. **Case Study 1: Moving a GWAS Discovery into a Therapeutic Intervention**

Inflammatory Bowel Disease Genetics
Judy Cho, Yale University

Refining and Translating Genomics for Disease Sub Setting and Coordinated Target Discovery for More Effective Therapeutic Clinical Trial Design
Stephan Targan, Cedars-Sinai Health System

Short Presentation: Determinants of HIV Response
David Goldstein, Duke University

Predicting Unmodifiable Disease Risks: Emotional and Behavioral Responses
Theresa Marteau, King's College London

Following case study presentations, participants are encouraged to participate in a group discussion of issues, including:

1. What level of "proof" of causation is required before a GWAS finding is sufficiently reliable to be deemed ready for translational considerations?
2. How can genetic information be used effectively when there is no proven treatment?

12:00 p.m. – 1:00 p.m. ***Lunch*** *(on your own)*

Session II

1:00 p.m. – 2:30 p.m. **Case Study 2: Putting Together a Picture of Risk for a Complex Genetic Trait for Prognostic Testing**
Chair: *Hakon Hakonarson,* The Children's Hospital of Philadelphia

Thinking Big: Using Genome Wide Association Meta-Analysis to Identify Additional Loci Influencing Type 2 Diabetes, Obesity, and Height
Mark McCarthy, Oxford Centre for Diabetes, Endocrinology and Metabolism

Translating Type 2 Diabetes Whole Genome Association Studies
Alan Shuldiner, University of Maryland School of Medicine

Providing Information on Genetic Risk for Common Disease in the Context of Environmental Risk Factors
Colleen McBride, National Human Genome Research Institute

Following case study presentations, participants are encouraged to participate in a group discussion on questions, including:

1. How do we assemble an accurate assessment of genetic risk?
2. What goals are realistic for the near-term for complex diseases like diabetes: risk assessment and, based on risks, medical surveillance and lifestyle modification, or therapeutic development?

2:30 p.m. – 3:00 p.m. **Break**

3:00 p.m. – 4:30 p.m. **Case Study 3: Cancer Genetics and Genomics: Evidence-Based Guidelines for Gene-Based Testing**

Evaluation of Genomic Applications in Practice and Prevention, BRCA1 Testing
Al Berg, University of Washington

Implications of Germline Variation for Breast Cancer Treatment
Mark Robson, Memorial Sloan-Kettering Cancer Center

Genetic Counseling Challenges with Genetic Risk for Cancer
Jill Stopfer, University of Pennsylvania

Molecular Diagnosis of Kidney Failure
Matthias Kretzler, University of Michigan

Application of Molecular Information to Primary Diagnosis of Breast Cancer for Targeted Treatment Decisions and Improved Patient Outcome
John Sninsky, Celera

Following case study presentations, participants are encouraged to participate in a group discussion on issues, including:

1. Based on what we have learned from the breast cancer work, what are the barriers to widespread genetic testing based on genotype or expression for complex traits?
2. How do we develop evidence-based guidelines and how do we translate them into practice?

Tuesday, March 11, 2008

Session III
Chair: *Joan Scott,* Genetics and Health Policy Center

8:30 a.m. – 10:00 a.m. **Incorporating Genetic Information into Clinical Practice**
Wylie Burke, University of Washington

Commercial Development of Genetic Tests
Brad Popovich, Sirius Genomics

10:00 a.m. – 10:30 p.m. ***Break***

10:30 a.m. – 12:00 p.m. **Case Study 4: Pharmacogenomics and Translation in General Practice**

Pharmacogenetics and GWAS
Hakon Hakonarson, Children's Hospital of Philadelphia

Pharmacogenetic Clinical Trials
Nik Schork, Scripps Research Institute

Warfarin Dosing and Genetic Variation
Allan Rettie, University of Washington

Economic Considerations in the Use of Pharmacogenomics
David Veenstra, University of Washington

Following case study presentations, participants are encouraged to participate in a group discussion of issues, including:

1. What are the challenges in designing therapeutic studies based on genotype?
2. What are the optimal conditions for translating pharmacogenomics applications into practice?
3. What are the barriers to widespread use of pharmacogenomic data?

12:00 p.m. – 1:00 p.m. ***Lunch*** *(on your own)*

Session IV: **Setting a Translation Research Agenda**

1:00 p.m. – 3:00 p.m. Participants will discuss emerging themes and questions about translating genetic data from GWAS into clinical research and applications and identify key questions for future research.