

Computational Models for Analyzing Genotype-Phenotype Associations in Rare Diseases

Agenda
July 24-25, 2008
Room 9100, Rockledge II

08:30 AM **Welcome**
Drs. Shurin and Gopal-Srivastava

Introductions and Charge
Meeting Co-Chairpersons & NHLBI Staff

Rare Diseases in Heart, Lung, and Blood

09:00 AM **Rare Diseases in Heart, Lung and Blood**

A number of monogenetic and polygenetic rare diseases are known in the areas of heart lung and blood. In addition, these diseases often express a wide array of symptoms in patients with the same disease. Some of these diseases and their symptoms will be discussed in relation to phenotypes

9:00 Heart Jeffrey Towbin MD, Texas Heart Institute, Baylor College of Med.

“Rare Disease of the Heart: Cardiomyopathies”

9:20 Heart Arthur Moss MD, University of Rochester

“New Insights Into the Long QT Syndrome: A Genetic Channelopathy”

9:45 Lung Benjamin Rybicki PhD, Case Western Reserve University

“Genetic Dissection of Sarcoidosis Phenotypes”

10:00 Lung Sessions Cole MD, Washington University, St. Louis

“Genetic Disruption of Pulmonary Surfactant Metabolism”

10:30 AM **Break**

10:45 Blood Jeffrey Lipton MD, Albert Einstein College of Medicine

“The Lack of Robust Phenotypic Data: A Correctable Lesion in the Analysis of Genotype-Phenotype Correlations in Rare Diseases”

11:10 Blood Ellis Neufeld MD, Children’s Hospital Boston

“Genetic analyses in “simple” and complex blood disorders”

11:30 Open discussion

12:00 noon **Lunch**

1:00 PM **Defining and Collecting Phenotype Information**

What and how much phenotypic information should be collected? How well are phenotypes defined and what metrics should be used? Which phenotypic data are most suitable for genotype association studies?

1:00 Barry Collier MD, Rockefeller University

“Ontology-driven Phenotyping Instruments to Pool Data Across Sites: The Electronic Research Record”

- 1:20 Edwin Silverman MD, Brigham and Women's Hospital
"Rare Genetic Determinants of COPD"
1:40 Open Discussion

2:00 PM Genotyping and Genetic Epidemiology

What are the most appropriate methods for genotyping? Genome-wide analyses vs. candidate gene analyses – which are better and when to use them?

- 2:00 Emily Harris PhD, National Human Genome Res. Inst.
"Genome-wide Association Success Stories: What Can They Tell Us for Rare Diseases?"
2:20 Gail Jarvik MD, University of Washington
"Areas of Study Design, Analysis Issues and the Role of the Independent Investigator"

2:40 PM Break

- 3:00 Terri Beaty, Johns Hopkins University
"How to detect interaction between genes and environmental exposures"
3:20 Steve Sherry, National Library of Medicine
"The NCBI dbGaP database of genotypes and phenotypes provides resources for genome-wide association studies"
3:40 Open Discussion

4:00 PM Summary of Days' Discussions

4:30 PM Adjourn

-- DAY TWO --

8:30 AM Computational Models and Challenges in analyzing genotype-phenotype data

- 8:30 Mike Province PhD, Washington University St. Louis
"Mining for Gold Dust: Methods to Detect the Cumulative Impact of Small Effect Gene Variants"
8:50 Chris Amos PhD, MD Anderson Cancer Center, Houston
"Practical Issues in Performing Large Scale Meta-analyses for Complex Diseases"
9:10 Ellen Sidransky, MD, National Human Genome Res. Inst.
"Genotype Phenotype Correlation in Gaucher Disease"
9:30 Idan Menashe, PhD, National Cancer Institute
"PGA: Power Calculator for Case-control Genetic Association Analyses"
9:50 Paola Sebastiani, PhD, Boston University
"Leveraging pleiotropy to boost the power of genome wide association studies"
10:10 Open discussion

10:30 Break

BREAK OUT SESSIONS

10:45 Identification of Needs of Research Communities

Drs. Towbin, Rybicki, Lipton, Harris, Sherry, Province, Sidransky, Menashe, Moss, Cole, Neufeld, Silverman, Jarvik, Beaty, Amos, and Sebastiani

12:00 Working Lunch

1:00 Final Discussions and Recommendation Synthesis

2:00 Adjourn