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 Coller 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