Objectives:

1. Discuss the scientific questions and opportunities that can be substantially addressed by large-scale genomics studies, starting with genome sequencing but also considering other genomic technologies.

2. Consider options for future NHGRI programs that would address these questions and opportunities.

Monday, July 28, 2014

8:30 a.m. - 9:00 a.m. Welcome and Charge for the Workshop
Eric Green, M.D., Ph.D.
National Human Genome Research Institute, NIH

9:00 a.m. - 9:15 a.m. Background and Orientation
Adam Felsenfeld, Ph.D.
National Human Genome Research Institute, NIH

Part 1: The Science of the Current NHGRI Genome Sequencing Program

9:15 a.m. - 9:45 a.m. Discovering Variants Conferring Risk for Common Diseases
Michael Boehnke, Ph.D.
University of Michigan

9:50 a.m. - 10:20 a.m. Discovering the Genomic Bases of Mendelian Diseases
Roderick McInnes, M.D., Ph.D.
Lady Davis Institute for Medical Research

10:25 a.m. - 10:55 a.m. Genome Sequencing for Clinical Care
Dan Roden, M.D.
Vanderbilt University

11:00 a.m. - 11:20 a.m. Break (on your own)

11:20 a.m. - 11:50 a.m. Functional Genomics at Scale
Joseph Ecker, Ph.D.
Howard Hughes Medical Institute and Salk Institute for Biological Studies
11:50 a.m. - 12 noon  Setting the Stage for the Discussion  
Adam Felsenfeld, Ph.D.  
National Human Genome Research Institute, NIH

12 noon - 1:15 p.m.  Lunch (on your own)

1:15 p.m. - 2:15 p.m.  Discussion and Refinement of Opportunities  
Ewan Birney, Ph.D.  
European Molecular Biology Laboratory

Part 2: How Should Scale Evolve?

2:15 p.m. - 2:45 p.m.  Introduction to Breakout Groups:  
What Are the “Big Challenges” That NHGRI Should Pursue in the Next ~5 Years?  
William M. Gelbart, Ph.D.  
Harvard University  
Adam Felsenfeld, Ph.D.  
National Human Genome Research Institute, NIH

3:00 p.m. - 5:00 p.m.  Breakout Groups (2 hours each)  

Group 1: Understanding the Genetic Architecture of Health and Disease at Scale  
Eric Boerwinkle, Ph.D.  
The University of Texas Health Science Center at Houston  
Mike Bamshad, M.D.  
University of Washington  
Adam Felsenfeld, Ph.D.  
National Human Genome Research Institute, NIH

Group 2: Integrating Genomic Variant Discovery with Function  
Richard Myers, Ph.D.  
HudsonAlpha Institute for Biotechnology  
Mark Gerstein, Ph.D.  
Yale University  
Elise Feingold, Ph.D.  
National Human Genome Research Institute, NIH  
Mike Pazin, Ph.D.  
National Human Genome Research Institute, NIH
Group 3: Clinical Genome Sequencing at Scale

Heidi Rehm, Ph.D.
Partners Healthcare

Sharon Emma Plon, M.D., Ph.D.
Baylor College of Medicine

Lucia Hindorff, Ph.D., M.P.H.
National Human Genome Research Institute, NIH

Carolyn M. Hutter, Ph.D.
National Human Genome Research Institute, NIH

Group 4: Comparative and Evolutionary Genomics

Andrew Clark, Ph.D.
Cornell University

Evan E. Eichler, Ph.D.
University of Washington

Michael W. Smith, Ph.D.
National Human Genome Research Institute, NIH

5:15 p.m. - 5:30 p.m. Reconvene: Issues Arising, Logistics, Evening Session

Adam Felsenfeld, Ph.D.
National Human Genome Research Institute, NIH

5:30 p.m. - 7:30 p.m. Dinner (on your own)

7:30 p.m. - 9:00 p.m. Challenge Talks (10-minute talk/5-minute question)

7:30 p.m. - 7:45 p.m. Peptide Display and T-Cell Recognition Project

David Haussler, Ph.D.
University of California, Santa Cruz

7:45 p.m. - 8:00 p.m. Using the Negative Correlation Between Polygenic and Rare Variant Burdens for Common Disease To Improve Study Efficiency and Power

Nancy Cox, Ph.D.
The University of Chicago

8:00 p.m. - 8:15 p.m. The Human Cell Atlas

Aviv Regev, Ph.D.
Broad Institute

8:15 p.m. - 8:30 p.m. Measuring the Functional Consequences of Very Large Numbers of Human Genetic Variants

Jay Shendure, M.D., Ph.D.
University of Washington
8:30 p.m. - 8:45 p.m. Not Your Father’s PDF: New Forms of Knowledge Representation for Genomic Sequence
Daniel Masys, M.D.
University of Washington

9:00 p.m. Breakout Group Co-Chairs Meet To Coordinate Summaries
Breakout Co-Chairs and Staff Only

Tuesday, July 29, 2014

**Part 3: Moving From Ideas to Implementation**

7:30 a.m. - 8:30 a.m. Staff Meets with "Scenarios" Co-Chairs To Develop Discussions
(Breakout Co-Chair Coordination if Needed)

8:30 a.m. - 10:10 a.m. Breakout Reports From Day 1 (15-minute report/10-minute discussion)
Salons A & B
Breakout Group Co-Chairs

8:30 a.m. - 8:55 a.m. Group 1: Understanding the Genetic Architecture of Health and Disease at Scale

8:55 a.m. - 9:20 a.m. Group 2: Integrating Genomic Variant Discovery with Function

9:20 a.m. - 9:45 a.m. Group 3: Clinical Genome Sequencing at Scale

9:45 a.m. - 10:10 a.m. Group 4: Comparative and Evolutionary Genomics

10:15 a.m. - 10:30 a.m. Break (on your own)

10:30 a.m. - 10:45 a.m. Introduction: "Scenarios"
Adam Felsenfeld, Ph.D.
National Human Genome Research Institute, NIH

William M. Gelbart, Ph.D.
Harvard University

10:45 a.m. - 1:30 p.m. Alternate "Scenarios" (5-minute introduction/25-minute discussion)

10:45 a.m. - 11:15 a.m. Element 1: Understanding the Genetic Architecture of Health and Disease at Scale
Len Pennacchio, Ph.D.
Lawrence Berkeley National Laboratory

Adam Felsenfeld, Ph.D.
National Human Genome Research Institute, NIH
11:15 a.m. - 11:45 a.m.  
**Element 2: Integrating Genomic Variant Discovery with Function**  
Carlos Bustamante, Ph.D.  
Stanford University  

Mike Pazin, Ph.D.  
National Human Genome Research Institute, NIH  

Elise Feingold, Ph.D.  
National Human Genome Research Institute, NIH  

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

1:00 p.m. - 1:30 p.m.  
**Element 3: Clinical Genome Sequencing at Scale**  
James Philip Evans, M.D., Ph.D.  
The University of North Carolina at Chapel Hill  

Lucia Hindorff, Ph.D., M.P.H.  
National Human Genome Research Institute  

1:30 p.m. - 2:30 p.m.  
**Discussion: Combining Strategic and Tactical Considerations**  
Rex Chisholm, Ph.D., M.S.  
Northwestern University  

Adam Felsenfeld, Ph.D.  
National Human Genome Research Institute, NIH  

Jeffery A. Schloss, Ph.D.  
National Human Genome Research Institute, NIH  

2:30 p.m. - 3:00 p.m.  
**Wrap-up**  
Eric Green, M.D., Ph.D.  
National Human Genome Research Institute, NIH  

3:00 p.m.  
**Adjournment**