



**Future Opportunities for Genome Sequencing and Beyond:
A Planning Workshop for the National Human Genome Research Institute**

July 28-29, 2014

*Bethesda North Marriott Hotel and Conference Center
Bethesda, Maryland*

AGENDA

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	<i>Salons A & B</i>
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 (<i>on your own</i>)	
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:** *Salons A & B*
**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*** *Linden Oak*
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 *Brookside A*
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

Forest Glen

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

Brookside B

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

Salons A & B

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*)

Salons A & B

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)*
Breakout Group Co-Chairs

Salons A & B

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

Salons A & B

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