

# From Genome Function to Biomedical Insight: ENCODE and Beyond

# March 10-11, 2015 Natcher Conference Center, Rooms E1/E2 National Institutes of Health

## Agenda

### Objectives:

- 1. Discuss the scientific questions and opportunities for better understanding genome function and applying that knowledge to basic biological questions and disease studies through large-scale genomics studies.
- 2. Consider options for future NHGRI projects that would address these questions and opportunities.

#### Tuesday, March 10, 2015

1:00 p.m.	Welcome and Setting the Context	Eric Green
1:15 p.m.	Purpose of Workshop: Background and Planning Process for Future Initiatives	Elise Feingold
1:35 p.m.	From Genome Function to Biomedical Insights: Defining the Scientific Challenges	Ewan Birney
2:05 p.m.	Discussion	
2:30 p.m.	Break	
3:00 p.m.	Genome Function Circa 2016: Updates from Related Projects Moderator: Daniel Gilchrist	
	ENCODE	Michael Snyder
	REMC/IHEC/BLUEPRINT PsychENCODE Genomics of Gene Regulation (GGR) 4D Nucleome FunVar	Michael Pazin

FANTOM Daniel Gilchrist

GTEX LINCS TCGA/ICGC KOMP2/IMPC

4:00 p.m. **Proposals for Future Directions** 

ENCODE PIs' Vision for Functional Genomics Joseph Ecker

Recommendations related to genome function from NHGRI's Mark Gerstein Planning Workshop on the Future Opportunities Richard Myers

for Genome Sequencing and Beyond

5:00 p.m. **General Discussion** 

6:00 p.m. Working Dinner

7:00 p.m. **Topic #1: Identifying and characterizing functional elements** 

Moderator: Carol Bult

7:00 p.m. – 7:30 p.m.

The regulatory landscape: where are the gaps? Ross Hardison

7:30 p.m. – 7:45 p.m.

Creating a framework for mechanistic studies B. Franklin Pugh

7:45 p.m. – 8:00 p.m.

ENCODE 2.0: improving the syntax for Laurie Boyer

understanding functional elements in the genome

8:00 p.m. - 8:15 p.m.

Genomics at the "quantum" level: new directions for William Greenleaf genomic data generation and functional validation

8:15 p.m. **Topic #1 Discussion** 

9:00 p.m. Adjourn

Wednesday, March 11, 2015

8:00 a.m. Topic #2: Using genomic assays of function to interpret the role of genetic variation in

disease

Moderator: Eric Boerwinkle

8:00 a.m. – 8:25 a.m.

Leveraging whole genome annotation for Eric Boerwinkle genotype-phenotype association studies

8:25 a.m. - 8:50 a.m.

Hirschsprung disease consequent to mutations in

the RET gene regulatory network

Aravinda Chakravarti

8:50 a.m. - 9:05 a.m.

Genetically predicted endophenotypes:

getting to the next level in understanding how genome variation drives disease

Nancy Cox

9:05 a.m. - 9:20 a.m.

Identification of regulatory variation important for

maternal metabolism during pregnancy

William Lowe

9:20 a.m. **Topic #2 Discussion** 

10:05 a.m. *Break* 

10:30 a.m. Topic #3: Using genomic assays of function to study basic biological questions

Moderator: Aviv Regev

10:30 a.m. - 10:50 a.m.

Understanding basic biology using functional

genomics: solving the genotype-phenotype problem

**Brenda Andrews** 

10:50 a.m. – 11:10 a.m.

Fundamental insights into gene regulation from

Karen Adelman

genomic analyses: past successes and future challenges

11:10 a.m. – 11:30 a.m.

Analyzing cytosine modifications in genomic DNA

Anjana Rao

11:30 a.m. – 11:50 a.m.

Ask not what you can do for ENCODE – ask what ENCODE can do for you

John O'Shea

11:50 a.m. **Topic #3 Discussion** 

12:30 p.m. *Lunch* 

1:30 p.m. **Discussion** 

Moderators: Eric Boerwinkle, Carol Bult, Aviv Regev

2:30 p.m. Final Recommendations, including priorities and balance of activities

Moderators: NHGRI

3:30 p.m. Adjourn