National Human Genome Research Institute

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 GTEx LINCS TCGA/ICGC KOMP2/IMPC	Daniel Gilchrist
4:00 p.m.	**Proposals for Future Directions** ENCODE PIs' Vision for Functional Genomics	Joseph Ecker
	Recommendations related to genome function from NHGRI's Planning Workshop on the Future Opportunities for Genome Sequencing and Beyond	Mark Gerstein Richard Myers
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 understanding functional elements in the genome	Laurie Boyer
	8:00 p.m. – 8:15 p.m. Genomics at the "quantum" level: new directions for genomic data generation and functional validation	William Greenleaf
8:15 p.m.	**Topic #1 Discussion**	
9:00 p.m.	**Adjourn**	

<u>Wednesday, March 11, 2015</u>

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 genotype-phenotype association studies	Eric Boerwinkle

	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 genomic analyses: past successes and future challenges	Karen Adelman
	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**