

Genomic Medicine IX: NHGRI's Genomic Medicine Portfolio

April 19-20, 2016



Sheraton Silver Spring
8777 Georgia Avenue
Silver Spring, MD 20910
(301) 468-1100

www.sheratonsilverspring.com

Tuesday, April 19th

LOCATION: Sheraton Silver Spring – Magnolia Ballroom
WEBCAST: Genome TV - <http://www.genome.gov/GenomeTV/>
TWITTER: #GenomicMed9

7:30 AM	Registration and Breakfast	Registration Table
	<i>Session 1: Introduction, Background</i> <i>Dan Roden, Moderator</i>	
8:30 AM	Welcome, Introductions, and Goals of the Meeting	Carol Bult Teri Manolio Eric Green
8:50 AM	Convincing Clinicians to Use Functionalized Genomic Information	Howard Jacob
9:05 AM	Magnitude of the Problem – Basic Science Perspective On Need for Integration	Monte Westerfield
9:30 AM	Magnitude of the Problem – Clinical Perspective on Need For Integration	Gail Herman
9:55 AM	Discussion	
10:40 AM	Break	
	<i>Session 2: Vexing Clinical Problems Needing Basic Input</i> <i>Laura Rodriguez, Moderator</i>	
11:00 AM	Speeding Functional Assessment to Benefit Patients	Stephen Kingsmore
11:20 AM	De Novo Variants that Inform Clinical Phenotypes	Christine Seidman
11:40 AM	Discussion	

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Tuesday, April 19th...continued



12:25 PM LUNCH

Session 3: From Variant to Disease Mechanisms – Specific Examples of How Model Systems have used Genetic / Genomic Approaches to Lend Insight into Human Disease that had Clinical Relevance
Howard Jacob, Moderator

1:30 PM Integrating Model Organism Data around Clinical Genomics Calum MacRae

1:50 PM Leveraging Congenital Heart Disease Mouse Model Findings to Improve Clinical Outcome Cecilia Lo

2:10 PM Discussion

3:00 PM BREAK

Session 4: Computational Approaches to Variant Function Prediction Methods for Predicting Functional Consequences of Variants
Marc Williams, Moderator

3:20 PM Leveraging Massive-Scale Databases of Human Genetic Variation Daniel MacArthur

3:40 PM Empowering Variant Effect Prediction with Large Scale Mutagenesis Data Douglas Fowler

4:00 PM Discussion

4:45 PM **PANEL 1: Topics to be Defined Based on Day 1 Sessions**
Teri Manolio, Moderator
Les Biesecker, Greg Cooper, Cat Lutz, Erin Ramos, and Liz Worthey

5:45 PM First Day Summary and Discussion Carol Bult
Teri Manolio

6:00 PM ADJOURN

On Your Own for Dinner (see map in registration materials)

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Wednesday, April 20th

7:30 AM	Breakfast	
	Session 5: Functionalizing VUS's Howard McLeod, Moderator	
8:30 AM	Massively Parallel Functional Analysis of Missense Mutations In BRCA1 for Interpreting Variants of Uncertain Significance	Lea Starita
8:50 AM	CRISPR-Cas9 Mediated Mouse Model Creation and Transcription Regulation	Haoyi Wang
9:10 AM	Discussion	
9:55 AM	BREAK	
	Session 6: Biomedical Phenotype Ontologies and Data Integration Carol Bult, Moderator	
10:15 AM	Translating Human to Models and Back Again: Phenotype Ontologies for Data Integration and Discovery	Melissa Haendel
10:35 AM	Data Integration: Genome X Transcriptome X EMR	Nancy Cox
10:55 AM	Discussion	
11:35 AM	Providing Clinical Utility: Who Is the Judge and Jury? Webex link: https://dukemed.webex.com/dukemed/j.php?MTID=m3e8636840d17de8e1eaf15051d925752 Meeting number: 738 378 332 Meeting password: GM904202016	Robert Nussbaum
11:50 AM	Discussion	
12:00 PM	WORKING LUNCH	
1:00 PM	Panel 2: Topics to be Defined Based on Day 2 Sessions Rex Chisholm, Moderator Calum MacRae, Jose Ordovas, Peter Robinson, Wendy Rubinstein, Barbara Stranger	
2:00 PM	Summary Discussion: Promoting Bedside-Back-To-Bench Research <ul style="list-style-type: none"> ▪ Prioritizing Variants for Functional/Mechanistic Investigation ▪ Incentivizing Demonstrations of Benignity ▪ Engaging Industry 	Carol Bult Teri Manolio
3:30 PM	Next Steps	Carol Bult Teri Manolio
4:00 PM	ADJOURN	