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

Genome TV - http://www.genome.gov/GenomeTV/ WFRCAST.

Bench

Genomic Medicine Meeting

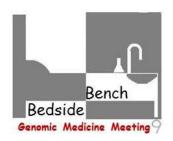
Bedside

TWITTER:	#GenomicMed9	<u>ov/Genomerv/</u>
7:30 AM	Registration and Breakfast	Registration Table
	Session 1: Introduction, Background Dan Roden, Moderator	
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	
	Session 2: Vexing Clinical Problems Needing Basic Input Laura Rodriguez, Moderator	
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	

Genomic Medicine IX: NHGRI's Genomic Medicine Portfolio

April 19-20, 2016

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)

Genomic Medicine IX: NHGRI's Genomic Medicine Portfolio

Wednesday, April 20th

ADJOURN

4:00 PM

7:30 AM	Breakfast	Bedside Genomic Medicine Meeting	9
	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 Integrated Bult, Moderator	ration	
10:15 AM	Translating Human to Models and Back Again: Phenotype Ontologies for Data Integration and Discovery	Melissa Haende	l
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:	Robert Nussbau	m
https://dul	<u>kemed.webex.com/dukemed/j.php?MTID=m3e8636840d17de</u> Meeting number: 738 378 332 Meeting password: GM904202016	<u>8e1eaf15051d925</u>	<u>5752</u>
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 Rubin Barbara Stranger	stein,	
2:00 PM	 Summary Discussion: Promoting Bedside-Back-To-Bench Rese Prioritizing Variants for Functional/Mechanistic Investigation Incentivizing Demonstrations of Benignity Engaging Industry 		
3:30 PM	Next Steps	Carol Bult Teri Mano	