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/](http://www.genome.gov/GenomeTV/) 
**TWITTER:** #GenomicMed9 

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
<th>Speaker(s)</th>
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</thead>
<tbody>
<tr>
<td>7:30 AM</td>
<td>Registration and Breakfast</td>
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<tr>
<td>8:30 AM</td>
<td><strong>Session 1: Introduction, Background</strong></td>
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<td></td>
<td>Welcome, Introductions, and Goals of the Meeting</td>
<td>Carol Bult</td>
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<td>Teri Manolio</td>
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<td>Eric Green</td>
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<td>8:50 AM</td>
<td>Convincing Clinicians to Use Functionalized Genomic Information</td>
<td>Howard Jacob</td>
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<tr>
<td>9:05 AM</td>
<td>Magnitude of the Problem – Basic Science Perspective On Need for Integration</td>
<td>Monte Westerfield</td>
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<tr>
<td>9:30 AM</td>
<td>Magnitude of the Problem – Clinical Perspective on Need For Integration</td>
<td>Gail Herman</td>
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<tr>
<td>9:55 AM</td>
<td>Discussion</td>
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<tr>
<td>10:40 AM</td>
<td>Break</td>
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<tr>
<td>11:00 AM</td>
<td><strong>Session 2: Vexing Clinical Problems Needing Basic Input</strong></td>
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<td>Speeding Functional Assessment to Benefit Patients</td>
<td>Stephen Kingsmore</td>
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<tr>
<td>11:20 AM</td>
<td>De Novo Variants that Inform Clinical Phenotypes</td>
<td>Christine Seidman</td>
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<tr>
<td>11:40 AM</td>
<td>Discussion</td>
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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)
Session 5: Functionalizing VUS's
Howard McLeod, Moderator

8:30 AM Massively Parallel Functional Analysis of Missense Mutations Lea Starita
In BRCA1 for Interpreting Variants of Uncertain Significance

8:50 AM CRISPR-Cas9 Mediated Mouse Model Creation and Haoyi Wang
Transcription Regulation

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 Melissa Haendel
Ontologies for Data Integration and Discovery

10:35 AM Data Integration: Genome X Transcriptome X EMR Nancy Cox

10:55 AM Discussion

[Webex link: https://dukemed.webex.com/dukemed/j.php?MTID=m3e8636840d17de8e1eaf15051d925752]
Meeting number: 738 378 332
Meeting password: GM904202016

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 Carol Bult
- Prioritizing Variants for Functional/Mechanistic Investigation Teri Manolio
- Incentivizing Demonstrations of Benignity
- Engaging Industry

3:30 PM Next Steps Carol Bult
4:00 PM ADJOURN

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