

# Wednesday

October 9

7:00 am - 9:00 am

## Breakfast Provided

Ballroom Foyer

8:00 am - 8:50 am

## All Project 3/ELSI Meeting

Salon A - C

Project 3 Investigators

8:30 am - 12:00 pm

## Concurrent Sessions I - Morning Session

### Large-Scale Analysis Centers (LSACS)

Salons D-E

8:30 am - 9:30 am

#### Preview of Plenary Session & Discussion

LSAC PI's

9:30 am - 9:45 am

#### Break

9:45 am - 10:30 am

#### Center 1;

*Program Description, Purpose, Relation to D2020, Budget Allocations, Discussion*

10:30 am - 11:15 am

#### Center 2;

*Program Description, Purpose, Relation to D2020, Budget Allocations, Discussion*

11:15 am - 12:00 pm

#### Center 3;

*Program Description, Purpose, Relation to D2020, Budget Allocations, Discussion*

### Clinical Sequencing Exploratory Research Steering Committee (CSER)

Salons A-C

9:00 am - 9:15 am

#### Welcome & Announcements

Jim Evans

9:15 am - 9:30 am

#### Possible CSER Project & Products

Wylie Burke

##### Details:

- Variant sign-out best practices and stakehold paper (Possible Sequencing or ROR work group project.
- EMR vendors and other stakeholder (possible EMR work group project)
- Need for orthogonal validation of next-gen results (Sequencing work group project)
- Other ideas, priorities, feedback?

9:30 am - 10:30 am

#### CSER U & R Grant Consent Form Analysis Project

Gail Henderson

10:30 am - 10:45 am

#### Break

10:45 am - 11:45 am
<p><b>"One challenge or lesson from our site"</b></p> <p><b>Format:</b> 10 minute presentation from each site individually, followed by a 5 minute Q&amp;A</p> <p><b>Presentations:</b></p> <ul style="list-style-type: none"> <li>• Jonthan Berg, UNC</li> <li>• Will Parsons, BCM</li> <li>• Nancy Spinner, CHOP</li> <li>• Ben Wilfond, Kaiser</li> </ul>
11:45 am - 1:15 pm
<p><b>Working Lunch</b></p> <p>CSER Working Groups</p> <p><b>Locations:</b></p> <ul style="list-style-type: none"> <li>• Salon A   Act-ROR</li> <li>• Salon B   EMR</li> <li>• Salon C   Informed Consent &amp; Governance</li> <li>• Salon E   Genetic Counselors</li> <li>• Russell   Pediatrics</li> <li>• Dirksen   Analysis &amp; Phenotype Measures</li> <li>• Calhoun   Sequencing Standards</li> <li>• Hart   Outcomes &amp; Measures</li> </ul>

**11:45 am - 1:15 pm**

**Lunch Provided**

*Ballroom Foyer*

*CSER: eat in breakouts (see above right);*

*LSAC: eat in Rockville & Chevy Chase*

*CSER lunch begins at 11:45 am;*

*LSACS lunch begins at 12:00 pm*

**1:15 pm - 4:00 pm**

**Concurrent Sessions I - Afternoon Session**

**Large-Scale Analysis Centers (LSACS)**

1:15 pm - 2:15 pm
<b>Summary of Center &amp; Discussion of SAP</b>
2:15 pm - 3:15 pm
<b>What are the Critical Scientific Questions &amp; Technology Goals that are best Addressed by LSACs?</b>
3:15 pm - 4:00 pm

**Clinical Sequencing Exploratory Research Steering Committee (CSER)**

1:15 pm - 2:00 pm
<b>Coordinating Center Update</b>
Gail Jarvik
<b>Presentations:</b>
<ul style="list-style-type: none"> <li>• Joseph Salama on the CSER Website</li> <li>• David Crosslin on the MEGA variant annotation database</li> </ul>
2:00 pm - 2:45 pm

**Setting Specific Deliverables for the Next 12 Months; What is Most Important to Accomplish before October 2014?**

**Program Overview: CRVR/U41**

**Format:**  
20 minute talk, followed by a 25 minute discussion

- Speakers:**
- Jonthan Berg, UNC
  - Heidi Rehm, BWH/Harvard
  - Sharon Plon, BCM

2:45 pm - 4:00 pm

**Variant sign-out Panel**

**Format:**  
15 minute overview from each panelist

- Panelists:**
- Yaping Yang, BCM
  - Heidi Rehm, BWH
  - Michael Dorschner, UW
  - Nick Wagle, DFCI

- Moderator:**
- Nick Wagle, DFCI

**4:00 pm - 5:00 pm**

**Break**

**5:00 pm - 6:30 pm**

**Dinner & Social**

*Dinner provided; individuals to pay hotel directly \$36/person  
Dirksen/Hart/Calhoun Rooms*

**7:00 pm - 8:30 pm**

**Plenary Session; Scientific Presentations**

*Salons A-D*

7:00 pm - 7:45 pm

**Scientific presentation; Complex Diseases**  
Mark McCarthy

7:45 pm - 8:30 pm

**Scientific presentation; Cancer/somatic sequencing; translation to medicine**  
Ken Offit

# Thursday

October 10

7:00 am - 8:15 am

## Breakfast Provided

*Ballroom Foyer*

8:00 am - 12:15 pm

## Plenary Session; Program Presentations

*Salons A-D*

8:00 am - 9:05 am

### Large-Scale Analysis Centers Program Presentation

**Format:** 45 minute talk and 20 minute Q&A

TBD

9:05 am - 9:55 am

### Centers for Mendelian Genomics Program Presentation

**Format:** 35 minute talk and 15 minute Q&A

David Valle

9:55 am - 10:20 am

### Break

10:20 am - 11:25 am

### Clinical Sequencing Exploratory Research Program Presentation

**Format:** 45 minute talk and 20 minute Q&A

**Speakers:**

Jim Evans

Wylie Burke

11:25 am-12:15 pm

### Genome Sequencing Informatics Tools Program Presentation

**Format:** 35 minute talk and 15 minute Q&A

Gabor Marth

12:15 pm - 1:20 pm

## Lunch Provided

*Ballroom Foyer*

**1:20 pm - 3:30 pm****Plenary Session; *Unifying Topics****Salons A-D*

1:20 pm-2:25 pm

**Variant Annotation and Database Session****Panelists:**

- Mark Gerstein, Non-coding variant annotation
- Daniel MacArthur, Loss of function variants and their annotation
- Sharon Plon, New variant databases

**Moderator:** Debbie Nickerson

2:25 pm-3:30 pm

**Interplay between Genomic Technologies and Informatics****Panelists:**

- Elaine Mardis, Human Re-sequencing projects
- Jay Shendure, Haplotype-resolved genome sequencing and assembly
- Jim Knowles, Emerging Technologies and analysis strategies-single-cell RNA-seq

**Moderator:**

Li Ding

**3:30 pm - 4:00 pm****Break****4:00 pm - 6:00 pm****CMG and GS-IT Joint Session***Salon A-D*

4:00 pm - 4:20 pm

**Emerging Bioinformatics Tools**

4:20 pm - 4:40 pm

**What are we missing? Examples from the CMG Programs**

4:40 pm - 5:40 pm

**Discussion/Brainstorming - How can the CMG and GS-It groups work together?**

5:40 pm - 6:00 pm

**Action Items with Li Ding****6:00 PM****Adjournment***Dinner on your own.*

# Friday

October 11

7:00 am - 8:00 am

## Breakfast Provided

Ballroom Foyer

8:00 am - 12:00 pm

## Concurrent Sessions II - Morning Session

### Centers for Mendelian Genomics

Salons A-B

8:00 am - 10:00 am

#### Welcome & Technology (or other vignette)

**Format:**

30 minute presentation from each site individually with a 10 minute Q&A

**Moderated by Debbie Nickerson**

**Presentation Order:**

- Jim Lupski, BCM-Johns Hopkins
- Dianna Milewicz, University of Washington
- Murat Gunel, Yale University

10:05 am - 10:20 am

#### Break

10:20 am - 11:30 am

#### Discussion

*Synchronizing Quarterly Reports & Data Release - Priorities*

**Moderator**

Mike Bamshad

### Genome Sequencing Informatics Tools Program

Salons C-D

8:00 am - 8:30 am

#### Networking with CSER

Heidi Rehm

8:30 am - 10:10 am

#### Project Progress

**Format:**

15 minute presentation from each site individually with a 5 minute Q&A

**Progress Update Order:**

- Li Ding & Jim Eldred, Turnkey System for HTP Sequence Data Analysis
- Goncolo Abecasis & Gabor Marth, Variant Identification & Functional Assessment
- Eric Banks, GATK Genome Analysis Toolkit
- Steve McCarroll & Bob Handshaker; Genome STRiP: Structural Variation
- Ali Torkamani, SG-Advisor: Annotation & Variant Interpretation
- Jim Knowles & Eva Deelman, Workflows for mRNA & Genome Re-sequencing

10:10 am - 10:25 am

#### Break

10:25 am - 11:25 am

#### iSeq Tools Portal

Gabor Marth & Brian D'Astous

11:25 am - 11:40 am
<b>GS-IT Deliverables</b> Li Ding
11:40 am - 12:00 pm
<b>Unifying Topics;</b> <i>Presentors:</i> <ul style="list-style-type: none"> <li>• Geraldine Van der Auwera</li> <li>• Steve McCarroll</li> </ul> <i>Moderator:</i> <ul style="list-style-type: none"> <li>• Li Ding, Chair</li> </ul>

<b>11:30 am - 12:00 pm (CMG)</b>	<b>12:00-1:00pm (GS-IT)</b>
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**Lunch Provided**

Ballroom Foyer

CMG working lunch begins at 11:30 am;

GS-IT lunch begins at 12:00 pm

<b>12:00 - 2:00 pm (CMG)</b>	<b>1:00-3:00pm (GS-IT)</b>
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**Concurrent Sessions II - Afternoon Session**

**Centers for Mendelian Genomics**

Salons A-B

12:00 pm - 12:30 pm
<b>Value of Mendelian Program</b> Rick Lifton
12:30 pm - 1:00 pm
<b>Sample Solicitation Update</b> Kati Buckingham
1:00 pm - 1:45 pm
<b>Thinking Ahead</b>
1:45 pm - 2:00 pm
<b>Wrap up</b>

**Genome Sequencing Informatics Tools Program**

Salons C-D

1:00 pm - 1:20 pm
<b>Unifying Topics;</b> <i>Next-Gen Outreach: semantic zoom &amp; other visualizations, Software Carpentry, Use of Cloud, GATK Forum, video tours, workshops, and social media</i> <ul style="list-style-type: none"> <li>• Brian D'Astous</li> <li>• Ewa Deelman</li> <li>• Goncolo Abecais</li> <li>• Geraldine Van der Auwera</li> </ul>
1:20 pm - 1:35 pm
<b>Unifying Topics;</b> <i>Shared Resources: Phone home, BioStar, workflows, data formats</i> <ul style="list-style-type: none"> <li>• Eric Banks</li> <li>• Jim Knowles</li> <li>• Jim Elred</li> <li>• Bob Handshaker</li> <li>• Ali Torkamani</li> </ul>

1:35 pm - 1:50 pm

**Unifying Topics;**

*Public Private Interface: Commercial vs. research, license models*

- Eric Banks
- Ali Torkamani
- Li Ding, etc

1:50 pm - 2:10 pm

**Unifying Topics;**

*Variation: SNV, Indel, Structural Variation*

- Mike Wendel
- Kai Ye
- Bob Handshaker
- Steve McCarroll
- Gabor Marth
- Eric Banks

2:10 pm - 2:25 pm

**Unifying Topics;**

*Variation Detection & Annotation*

- Ali Torkamani
- Mike Wendl
- Hyun Min Kang
- Kai Ye
- Eric Banks
- Alistair Ward

2:25 pm - 2:40 pm

**Unifying Topics;**

*Pipelines & workflows, Use of Cloud*

- Ewa Deelman
- Gabor Marth
- Goncolo Abecasis
- Li Ding, et al

2:40 pm - 3:00 pm

**Unifying Topics;**

*Clinical Data: CSER Perspective*

- Jonathan Berg

**3:00 PM**

**Adjournment**