

Meeting Agenda

Hyatt Regency Bethesda • 1 Bethesda Metro Center • Bethesda, MD 20814

Wednesday, February 1, 2017

7:30 – 8:30 a.m.	Networking Breakfast	Regency Foyer
8:30 – 8:40 a.m.	Welcome, Opening Remarks, General Updates <i>Rongling Li</i>	Regency I/II
8:40 – 8:50 a.m.	Announcements, Opening Remarks <i>Rex Chisholm</i>	Regency I/II
8:50 – 10:05 a.m.	Workgroup Breakout Session <ul style="list-style-type: none">• Genomics Workgroup & PGx Subgroup• EHR Integration Workgroup	Cabinet Judiciary
10:05 – 10:15 a.m.	Networking Break	Regency Foyer
10:15 – 10:40 a.m.	Undiagnosed Genetic Disorders in Adults with Chronic Kidney Disease (CKD) <i>Ali Gharavi</i>	Regency I/II
10:40 – 11:55 a.m.	Workgroup Breakout Session <ul style="list-style-type: none">• Outcomes Workgroup• Clinical Annotation Workgroup	Cabinet Judiciary
11:55 – 12:15 p.m.	Working Lunch	Regency Foyer
12:15 – 1:05 p.m.	Sequencing and Genomic Data Update <i>Heidi Rehm, Richard Gibbs & David Crosslin</i>	Regency I/II
1:05 – 2:20 p.m.	Workgroup Breakout Session <ul style="list-style-type: none">• ROR/ELSI Workgroup• Phenotyping Workgroup	Cabinet Judiciary
2:20 – 2:50 p.m.	Networking Break and Report-Out Preparation	Regency Foyer
2:50 – 3:05 p.m.	Meharry Medical College (Site Update) <i>Sam Adunyah & Phil Lammers</i>	Regency I/II
3:05 – 3:30 p.m.	Genomics Workgroup & Geocoding Report-Out <i>Megan Roy-Puckelwartz, Patrick Sleiman & David Crosslin</i>	Regency I/II
3:30 – 3:45 p.m.	EHR Integration Workgroup Report-Out <i>Sandy Aronson</i>	Regency I/II
3:45 – 4:05 p.m.	PGx Workgroup Report Report-Out <i>Laura Rasmussen-Torvik</i>	Regency I/II
4:05 – 4:20 p.m.	Outcomes Workgroup Report-Out <i>Hakon Hakonarson, Josh Peterson & Marc Williams</i>	Regency I/II
4:20 – 4:35 p.m.	Clinical Annotation Workgroup Report-Out <i>Gail Jarvik & Heidi Rehm</i>	Regency I/II
4:35 – 5:00 p.m.	ROR/ELSI Workgroup & HCP Report-Out <i>Ingrid Holm & Iftikhar Kullo</i>	Regency I/II
5:00 – 5:15 p.m.	Phenotyping Workgroup Report-Out <i>George Hripcsak & Peggy Peissig</i>	Regency I/II
5:15 – 5:30p.m.	Closing Remarks <i>Rex Chisholm</i>	Regency I/II
5:30 p.m.	Adjourn	

Toll-Free: 1-888-936-7423 OR Long Distance: +1 (510) 365-3231

Access Code: 486-477-721

<https://attendee.gototraining.com/r/960687669438928130>

eMERGE & CSER: The Convergence of Genomics and Medicine

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Thursday, February 2, 2017

7:30 – 8:25 a.m.	<p>Networking Breakfast</p> <p>CSER Tumor Working Group Breakfast <i>Will Parsons & Dan Robinson</i></p> <p>Topic: Discussing AMP Standards and Guidelines for the Interpretation and Reporting of Sequence Variants in Cancer</p>	Regency Foyer Cabinet / Judiciary
8:25 – 8:30 a.m.	<p>Opening Remarks</p> <p>CSER Program Officer <i>Lucia Hindorff (NHGRI)</i> eMERGE Program Officer <i>Rongling Li (NHGRI)</i></p>	Regency I/II
8:30 – 8:50 a.m.	<p>Top 5 Consortium-Wide Achievements from CSER & eMERGE</p> <p>CSER SC Chair <i>Gail Jarvik (U. of Washington)</i> eMERGE SC Chair <i>Rex Chisholm (Northwestern)</i></p>	Regency I/II
8:50 – 9:35 a.m.	<p>Session 1: Revision of ACMG Gene List</p> <p>Moderator <i>Gail Jarvik (U. of Washington)</i></p> <ul style="list-style-type: none"> • ACMG Incidental Findings List 2.0 <i>Wendy Chung (Columbia)</i> • VUS or GUS? Variants or genes with weak or uncertain evidence <i>Sharon Plon (Baylor)</i> • ACMG Gene Lists, Secondary Findings and Children <i>Ian Krantz (CHOP)</i> 	Regency I/II
9:35 – 9:45 a.m.	<p>Networking Break</p>	Regency Foyer
9:45 – 10:30 a.m.	<p>Session 2: Family Cascade Testing</p> <p>Moderator <i>Richard Sharp (Mayo)</i></p> <ul style="list-style-type: none"> • Building a Family Network <i>Kathleen Leppig (Group Health Cooperative)</i> • eMERGE Familial Implications of ROR Subgroup <i>Janet Williams (Geisinger)</i> • Challenges Related to Family Involvement in Clinical Whole-Genome Sequencing: Views of Non-Genetics Providers <i>Leila Jamal (Baylor)</i> 	Regency I/II
10:30 – 11:15 a.m.	<p>Session 3: Opportunities for Healthcare Quality Measurement in Genomics</p> <p>Moderator & Introduction <i>Marc Williams (Geisinger)</i></p> <ul style="list-style-type: none"> • Opportunities for Healthcare Quality Measurements in Genomics <i>John Bernot (National Quality Forum)</i> 	Regency I/II
11:15 – 11:25 a.m.	<p>Networking Break</p>	Regency Foyer

Livestream Link: www.genome.gov/GenomeTVLIVE

*eMERGE Members are invited to stay and attend CSER's Steering Committee meeting, beginning at 2:15pm in Regency I/II

11:25 – 12:25 p.m.

Group 1: EHR Group

**Cabinet /
Judiciary**

Discussion Chairs | *Brian Shirts (CSER) & Sandy Aronson (eMERGE)*

Relevant EHR Workgroups

CSER: EHR
eMERGE: EHR Integration

Discussion Topics

- Design & potential of future DIGITize-style CDS implementation Guides
- Technical & legal implications of large genomics information being accessible via EHR

Return of Results & Clinical Annotation Workgroups – choose Group 2 or 3 from the list below to attend

Relevant ROR & Clinical Annotation Workgroups

CSER: ACT-ROR, Informed Consent & Governance, Genetic Counseling, Pediatrics, Practitioner Education
eMERGE: ROR/ELSI, Clinical Annotation

Group 2: ACMG59 List

Regency III/IV

Discussion Chairs | *Gail Jarvik (CSER) & Wendy Chung (eMERGE)*

Discussion Topic

- Discuss current ACMG gene-disease list and consider nominations to the list

Group 3: Clinician Education

**Old
Georgetown**

Discussion Chairs | *Sharon Plon (CSER) & Ingrid Holm (eMERGE)*

12:25 – 1:00 p.m.

Working Lunch

Regency Foyer

1:00 – 2:00 p.m.

Group 1: Outcomes

**Cabinet /
Judiciary**

Discussion Chairs | *David Veenstra (CSER) & Josh Peterson (eMERGE)*

Relevant Outcomes Workgroups

CSER: Outcomes & Measures
eMERGE: Outcomes, Phenotyping

Discussion Topics

- eMERGE workgroup process/outcomes measures
- CSER psychosocial outcomes measures

Group 2: Genomics

**Old
Georgetown**

Discussion Chairs | *Will Salerno (CSER) & Megan Roy-Puckelwartz (eMERGE)*

Relevant Genomics Workgroups

CSER: Sequencing Standards, Tumor
eMERGE: Genomics

Discussion Topics

- Determine best methods & recommendations for CNVs/SV identification, interpretation, and inclusion on clinical reports
- Best format for sharing of known curated variants across groups

Group 3: ELSI

Regency III/IV

Discussion Chairs | *Barbara Koenig (CSER) & Kyle Brothers (eMERGE)*

Discussion Topics

- Future ELSI issues in translational genomic research, and the roles our consortia play in addressing these challenges

2:00 p.m.

Joint eMERGE/CSER Meeting Adjourns