emerge network

genda: Steering Committee Meeting

Thursday, October 3rd, 2019

Venue: The Hilton, 1750 Rockville Pike, Rockville, MD, 208

Roosevelt/Madison Meeting Room:

Lite breakfast 8:00-8:30 a.m.

8:30-8:50 a.m. NHGRI program official report | Robb Rowley (NIH/NHGRI)

8:50-9:10 a.m. Announcements, opening remarks | Rex Chisholm (SC Chair, Northwestern)

9:10-9:30 a.m. Integrating and harmonizing EHR data from multiple institutions across mother and child dyads | Brett

Harnett (CCHMC)

9:30-10:00 a.m. Sequencing Center & Genomic Data updates

FHIR, SV analysis & Year 5 updates | Eric Venner (BCM/HGSC)

Re-analysis of variants identified in eMERGEseq | Hana Zouk (Partners Broad)

Network datasets | David Crosslin (UW/CC)

AnVIL & eMERGE data access | Ken Wiley (NHGRI) 10:00-10:45 a.m.

10:45-11:00 a.m. **Networking Break**

Workgroup breakout session one* 11:00-12:20 p.m.

Harmonization, ROR, and impact on clinical outcomes (Roosevelt/Madison) | Outcomes, ROR, Clinical

Annotation, EHRI

Natural language processing & phenotype discovery (Jefferson) | Phenotyping, Genomics, PGx

12:20-12:50 p.m. **Working Lunch**

12:50-1:10 p.m. Choices, expectations, and attitudes about genomic screening in Latino and Ashkenazi Jewish

individuals | Julia Wynn (Columbia)

1:10-2:30 p.m. Workgroup breakout session two*

Preliminary analysis of eMERGEseq data (Roosevelt/Madison) | Clinical Annotation, Genomics,

Outcomes, Phenotyping

EHRI, CDS, & FHIR standards (Jefferson) | EHRI, PGx, ROR

2:30-2:50 p.m. Results of comparative study of participant reported adherence with adherence reported in the EHR

Alanna Kulchak Rahm (Geisinger)

Networking Break 2:50-3:10 p.m.

3:10-4:30 p.m. Workgroup breakout session three*

Genomic datasets and discovery (Roosevelt/Madison) | Genomics, PGx, CSG, Phenotyping

ROR Recommendations: informing return across large networks (Jefferson) | ROR/ELSI, Outcomes,

Clinical Annotation, EHRI

4:30-4:50 p.m. Genomic medicine for Chronic Kidney Disease: lessons and opportunities from eMERGE | Ali Gharavi

(Columbia)

Making work visible for electronic phenotype implementation: Lessons learned from the eMERGE 4:50-5:10 p.m.

Network | Ning Sunny Shang (Columbia)

5:10-5:15 p.m. Closing remarks | Rex Chisholm (SC Chair, Northwestern)

5:15 p.m. **Adjourn**

Dial-In Information Toll-Free: 1-877-568-4108 | United States 1-213-929-4221 Attendee Access Code: 523-304-296

Web-link: https://attendee.gotowebinar.com/register/3884028524235584513

^{*}Please note that investigators are invited to attend any breakout session. Relevant Workgroups are suggested for reference.

emerge network

genda: Steering Committee Meeting

Friday, October 4th, 2019

The Hilton, 1750 Rockville Pike, Rockville, MD, 20852 Venue:

Meeting Room: Roosevelt/Madison

Lite breakfast (open) | Executive session with ESP (Closed session; **Library**) 8:00-8:30 a.m. 8:30-8:45 a.m. Opening remarks | Robb Rowley (NIH/NHGRI) & Teri Manolio (NIH/NHGRI)

Comments from ESP Chair | Howard McLeod (Moffitt Cancer Center) 8:45-8:55 a.m.

8:55-9:15 a.m. eMERGE Network overview: Priorities, goals, progress and ESP recommendations | Rex

Chisholm (SC Chair, Northwestern)

9:15-10:20 a.m. Panel: Discovery in eMERGE data

GWAS, Polygenic Risk Scores & PheWAS demonstrate a polygenic determination of

vesicoureteral reflux | Miguel Verbitsky (Columbia)

• Phenotype Associations of LPA variants differ by race: A phenome wide association study

Benjamin Satterfield (Mayo)

The clinical utility of predicted family histories for Mendelian and genetically complex

forms of disease | Scott Hebbring (Marshfield)

10:20-10:40 a.m. **Networking Break**

10:40-11:55 a.m. Lessons learned panel: Impact of ROR on downstream analyses

Informing and harmonizing variant interpretation | Heidi Rehm (Partners/Broad), Iftikhar

Kullo (Mayo), & Adam Gordon (Northwestern)

Return of results pathways, barriers, and harmonizing across sites |Ingrid Holm (BCH) &

Iftikhar Kullo (Mayo)

• Impact of ROR process on Outcomes assessment | Josh Peterson (VUMC/CC) & Marc

Williams (Geisinger)

11:55-12:30 p.m. **Working Lunch**

Interim Data Analysis: Clinical Outcomes | Wendy Chung (Columbia), Christin Hoell (NU), 12:30-1:10 p.m.

Iftikhar Kullo (Mayo)

Status of eMERGE FHIR Specification and Implementation Project | Larry Babb (P/B) 1:10-1:35 p.m.

1:35-2:25 p.m. Input/Feedback from the ESP, general discussion

2:25-2:30 p.m. Closing remarks | Rex Chisholm (SC Chair, Northwestern)

2:30 p.m. **Adjourn**

Executive session with ESP (Closed session; Roosevelt/Madison) 2:30-3:15 p.m.

Dial-In Information Toll-Free: 1-877-568-4108 | United States 1-213-929-4221 Attendee Access Code: 523-304-296

Web-link: https://attendee.gotowebinar.com/register/3884028524235584513