agenda: Steering Committee Meeting

emerge network

Wednesday, February 19th, 2020

Venue: Hyatt Regency Bethesda, Bethesda MD

Meeting Room: Regency I/II

8:00-8:30 a.m.	**Light breakfast**
8:30-8:45 a.m.	NHGRI program official report | Robb Rowley (NIH/NHGRI)
8:45-9:05 a.m.	Meeting goals & announcements | Rex Chisholm (SC Chair, Northwestern)
9:05-9:15 a.m.	eMERGE data migration | David Crosslin (KPW/UW)
9:15-9:30 a.m.	Data utilization on the AnVIL | Robert Carroll (VUMC)
9:30-10:00 a.m.	Reclassifications | Hana Zouk (Partners/Broad) & Eric Venner (Baylor)
10:00-10:20 a.m.	**Networking Break**
10:20-11:20 a.m.	Lessons Learned: Penetrance Panel| Dan Roden (VUMC), Iftikhar Kullo (Mayo), Wendy Chung (Columbia), Gail Jarvik (KP/UW)
11:20-11:40 a.m.	Evaluation of breast cancer polygenic risk scores using eMERGE data | Cong Liu (Columbia)
11:40-12:00 p.m.	PheMap: a multi-resource knowledgebase for high-throughput phenotyping within electronic Health Records | Neil Zheng (VUMC)
12:00 – 12:30 p.m.	**Working Lunch**
12:30-1:40 p.m.	Breakout session • Milestone One: Expand the understanding of penetrance (Regency I/II) | *Clinical Annotation & Genomics* • Milestone Three: Standardize genomic clinical decision support (Regency III/IV) | *EHRI & PGx*
1:40-2:00 p.m.	Use of ClinVar for variant selection for real-time genetic diagnosis of epilepsy | Nephi Walton (Geisinger)
2:00-3:10 p.m.	Breakout session • Milestone Two: Impact of return of results on participant outcomes (Regency I/II) | *Outcomes* • Milestone Four: Develop natural language processing for five phenotypes (Regency III/IV) | *Phenotyping*
3:10-3:30 p.m.	**Networking Break**
3:30-3:50 p.m.	An implementation science framework to develop a CDS tool for Familial Hypercholesterolemia | Hana Bangash (Mayo)
3:50-5:00 p.m.	Breakout session • Milestone Five & Six: At risk family members & institutional impact of ROR (Regency I/II) | *ROR* • Milestone 7 | Independent working time for MCS teams (NT296, NT306, NT352 NT357) (Regency III/IV) | MCS Teams
5:00-6:00 p.m.	**eMERGE III Milestone Discussion | Light refreshments** **Goal:** Establish remaining needs to maximize scientific impact of eMERGE III (Regency Foyer) • Remaining workgroup and milestone barriers & expected needs for completion of work • Outstanding manuscript concept sheets and barriers to completion
6:00 p.m.	Closing remarks | Rex Chisholm (SC Chair, Northwestern)

Dial-In Information

Toll-Free: 1-866-901-6455 | United States 1-562-247-8421
Attendee Access Code: 888-196-492
Web-link: https://attendee.gotowebinar.com/register/5392020819915708941

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Thursday, February 20th, 2020

Venue:	Hyatt Regency Bethesda, Bethesda MD
Meeting Room:	Regency I/II
8:00-8:30 a.m.	**Light breakfast**
8:30-8:35 a.m.	Opening remarks | Robb Rowley (NIH/NHGRI)
8:35-9:15 a.m.	Six-month post return Outcomes analyses | Josh Peterson (Coordinating Center), Iftikhar Kullo (Mayo), Wendy Chung (Columbia), & Les Kirchner (Geisinger)
9:15-9:30 a.m.	Natural language processing phenotyping | Wei-Qi Wei (VUMC) & Chunhua Weng (Columbia)
9:30-9:55 a.m.	**Networking break & group photo**
9:55-10:15 a.m.	Return of Results: Experiments of nature | Ingrid Holm (BCH) & Julia Wynn (Columbia)
10:15-10:30 a.m.	XML FHIR update & progress | Mullai Murugan (Baylor)
10:30-10:45 a.m.	Genomics overall lessons | David Crosslin (KP/UW), Patrick Sleiman (CHOP), & Megan Roy-Puckelwartz (Northwestern)
10:45-11:00 a.m.	Pharmacogenomics lessons from eMERGE III | Cindy Prows (CCHMC) & Laura Rasmussen-Torvik (Northwestern)
11:00-12:00 p.m.	eMERGE III final deliverables | Rex Chisholm (SC Chair, Northwestern)
12:00 p.m.	**Adjourn**

Network Milestones

1. Expand the understanding of penetrance by describing the lessons learned from eMERGE, for example the sample size and age at onset required for penetrance analysis. Conduct penetrance analysis in conditions with sufficient data in the eMERGE cohort to assess impact on clinical outcomes.

2. Determine the impact of return of genetic results (RoR) on patients' immediate outcomes, 6 months and when available 12 months after RoR for variants with sufficient prevalence and data, which includes identifying Modification of clinical care (such as changes in prescriptions, lab tests ordered, etc.) and Outcomes related to processes of care, clinical utility, family utility, provider utility, and patients' psychosocial factors

3. Improve and/or standardize genomic clinical decision support (CDS) for return of clinically relevant genetic or incidental results directly to physicians, including initial assessment of impact on relevant process outcomes.

4. Develop a natural language processing (NLP) component for a maximum of five high-priority phenotypes, agreed upon by the phenotyping WG, the Steering Committee and NHGRI.

5. Explore the challenges involved in identifying at-risk family members and informing them of their potential risk as well as collect the responses of the family members.

6. Estimate the institutional impact of RoR.

7. Disseminate lessons learned on the various aspects of genomic medicine implementation by activities such as publishing articles that propose the key elements for effectively returning genomic results to providers and patients and comparing the impact different methods of RoR have on patient and physician care across all sites

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