

agenda:

Steering Committee Meeting

Thursday, June 4th, 2020

2:00 pm – 5:00 pm EST

- 2:00-2:15 p.m. NHGRI program official report | Robb Rowley (NIH/NHGRI)
- 2:15-2:25 p.m. Meeting goals & announcements | Rex Chisholm (SC Chair, Northwestern)
- 2:25-2:35 p.m. CSG & FHIR update | Hana Zouk (Partners/Broad), Eric Venner (Baylor), & Mullai Murugan (BCM)
- 2:35-3:00 p.m. Phenotyping workgroup natural language processing update & eMERGE III final deliverables | Wei-Qi Wei (VUMC) & Chunhua Weng (Columbia)
- 3:00-3:25 p.m. Outcomes workgroup outcomes analyses & eMERGE III final deliverables | Josh Peterson (VUMC), John Connolly (CHOP), Christin Hoell (Northwestern)
- 3:25-3:30 p.m. MeTree Supplement Update | Georgia Wiesner (VUMC)
- 3:30-3:45 p.m. ROR/ELSI workgroup eMERGE III final deliverables | Ingrid Holm (BCH) & Iftikhar Kullo (Mayo)
- 3:45-4:00 p.m. EHRI workgroup eMERGE III final deliverables | Sandy Aronson (Partners/Broad) & Casey Overby Taylor (JHU/Geisinger)
- 4:00-4:15 p.m. PGx eMERGE III final deliverables | Cindy Prows (CCHMC) & Laura Rasmussen-Torvik (Northwestern)
- 4:15-4:30 p.m. Genomics workgroup eMERGE III final deliverables | David Crosslin (KP/UW), Patrick Sleiman (CHOP), & Megan Roy-Puckelwartz (Northwestern)
- 4:30-4:45 p.m. Clinical Annotation workgroup eMERGE III final deliverables | Gail Jarvik (KP/UW) & Heidi Rehm (Partners/Broad)
- 4:45-5:00 p.m. Overall lessons learned from eMERGE III and recommendations for eMERGE IV
- 5:00 p.m. Closing Remarks | Rex Chisholm (SC Chair, Northwestern)

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