

## **Cohorts for Heart and Aging Research in Genetic Epidemiology Biobank Workshop**

We are pleased to announce a day-long workshop on the use of electronic health record data and health-system based biobanks for genetic epidemiology research. The workshop will be held the Wednesday prior to the 2019 CHARGE meeting in St. Louis and bring together experts in the field from a number of large academic health system-based biobanks. They will discuss the opportunities and challenges inherent in the use of electronic health record (EHR). The meeting is aimed to be an accessible introduction to those new to the use of EHR data for genetic analyses, as well as be of interest to those with considerable experience in the area.

### **AGENDA**

#### **8:30 Welcome and Introductions**

#### **8:45 Landscape of academic biobanks (4 minutes each)**

Penn Medicine Biobank – Scott Damrauer

Colorado Biobank – Leslie Lange

Michigan Genomics Initiative – Lillias Maguire

BioMe – TBD

McCode – Anne Justice

Partners Healthcare Biobank – Krishna Aragam

Taichung Biobank – Wayne Sheu (30 minutes)

#### **9:45 Break**

#### **Questions we face when working in a biobank: phenotyping and study design**

- 10:00 Lillias Maguire, University of Michigan. How do I decide who has what disease? Use of structured data and rule based phenotyping
- 10:30 Girish Nadkarni, Mount Sinai School of Medicine. What do I do with all the unstructured data? NLP and other approaches to EHR data
- 11:00 Jinbo Chen, University of Pennsylvania. What happens if I get the phenotype wrong? Misclassification bias at the population scale
- 11:30 Anne Justice, Geisinger Health System. What's different about biobank data? Harmonizing biobank and cohort study data

#### **12:00 Lunch**

#### **Questions that are well suited to asking in a biobank: Gene-first and multi-trait analysis**

- 1:00 Ron Do, Mt. Sinai School of Medicine. What are the real-world implications of carrying a genetic variant? Gene-first analyses of Transthyretin Amyloid Cardiomyopathy
- 1:30 Christopher Gignoux, University of Colorado. How does population structure affect health and disease? Population health-based approaches to genetics

- 2:00 Pradeep Natarajan, Massachusetts General Hospital. Do polygenic risk scores really tell us anything? Health system-based analysis of genetic risk
- 2:30 Anuarg Verma, University of Pennsylvania. What is a PheWAS? Analytic pipelines, strengths and limitations
- 3:00 Marylyn Ritchie, University of Pennsylvania. Can PheWAS tell us anything novel? The value of unbiased analyses
- 3:35 Wrap up