eMERGE Network: Summary of the Steering Committee Meeting

February 1, 2017 in Bethesda, MD

The winter Year Two, Phase III eMERGE Steering Committee was held on February 1st, 2017 in Bethesda, MD. In order to ensure that the Network continues on a productive note midway through our second year, please find highlights from the Meeting below. Day 2 of the meeting (February 2nd) was joint with the CSER consortium.

Presentation slides are available here (login required).

Full Session

Welcome, Opening Remarks, General Updates | *Rongling Li (NIH/NHGRI)*

- Rongling Li, on behalf of the NHGRI eMERGE Team, provided the Program Official Report for the 29th eMERGE Steering Committee Meeting. It is noted that Carolyn Hutter, Ph.D. is now the Acting Director of the Division of Genomic Science at the NHGRI.
- The Network reviewed the eMERGE Phase III Timeline, specifically noting the inclusion of an Interim dbGaP Data Submission after July 2017 at which time at least a portion of the sequencing data will be available from all sites. The Interim dbGaP Data Submission will serve as a work product of the first half of eMERGE Phase III and position the Network favorably for a potential eMERGE Phase IV continuation.
- A high level overview of eMERGE's data contribution to the scientific community at large as of 1/27/2017 was provided, specifically noting 1617 approved data access requests. In addition, eMERGE data availability was reviewed, including data type, size (n), DNAnexus date, and dbGaP submission.
- The Program's goals for the meeting were outlined, which are included below:
    - Update on genomic sequencing status and dataflow
    - Report on eMERGE data imputation
    - Refine timelines for sequencing, phenotyping, network data management, and dbGaP data submission
    - Set up site-specific timeline for RoR
    - Update on ongoing scientific projects
    - Propose new network-wide projects/studies
    - Identify gaps in genomic medicine that can be filled continuously by combining biorepositories and EMRs
    - Suggest possible directions for potential eMERGE IV initiatives
- Questions for workgroups to address in their breakout sessions are listed below. Any questions unable to be addressed in the allotted time today can be discussed within the workgroups monthly meetings or at the next Steering Committee meeting in June.
    - What are the deliverables in eMERGE III?
    - What is the timeline for completing these deliverables?
    - What are the barriers that are hindering the ability to complete the goals of the workgroup?
    - How will the workgroup products build the foundation for a possible eMERGE IV?

Announcements, Opening Remarks | *Rex Chisholm (Northwestern/SC Chair)*

- In response to feedback from the Network, the agenda has been structured in a format that allows for more time to be spent in workgroup breakout discussions.
- The following day is the second joint CSER/eMERGE meeting conducted since CSER's inception. Participation was encouraged as there are many synergies between the two Networks.

- Upcoming Steering Committee & ESP meetings and the publication status were reviewed, as well as developments since the prior Steering Committee/ESP meeting (October 2016), which are included below.
    - eMERGE received a favorable ESP report. The ESP suggested the Network collect outcomes and study impact of ROR for PGx, which the Network addressed by elevating PGx to full workgroup status and acquiring an additional co-chair (Cindy Prows of CCHMC). The ESP also suggested initiation of more collaborative projects amongst the Network members. Rex pointed towards the 1800 available whole genomes (CHOP and NU) and the exomes from Columbia as potential areas for collaboration.
    - Imputation of eI/eII/eIII array data on the Michigan Imputation Server (MIS) is in process with intended completion by March 2017.
    - The CSGs have begun return of sequencing results and clinical reports to sites. To date, 6800 samples received, 3800 sequenced, and 293 clinical reports issued.
    - PGx dataset is finalized, and the dbGaP upload is in process.

Addressing Ethical Challenges in Networked Biorepositories | *Kyle Brothers (CC/U. of Louisville) & Aaron Goldenberg (Case Western)*

- This project's goals are: 1) to characterize the ethical and regulatory challenges created by current networked biorepositories, and 2) to identify policy and practice solutions to address these challenges. It is funded through the ELSI program of the NHGRI, *NHGRI 1R01HG008988-01A1*. The project's website: www.BiobankNetworks.org
- Specific aims of the project include:
    - Examine the ethical and regulatory challenges in existing networked biorepositories and the solutions they use to address them ("Bio-network" Survey and Interviews)
    - Longitudinal observations of the governance, oversight, and problem-solving processes utilized by networked biorepositories (Participant Observations): Mayo Clinic Biobanks, Kentucky Cancer Registry, eMERGE Network, Sage Bionetworks, NBSTRN Virtual Dried Bloodspot Repository, Others?
    - Create best practices or other recommendations to address consent, privacy and data security, data access, governance, and oversight in networked biorepositories (Translational Aim): National Symposium/Workgroup on /Networked Biorepositories

Undiagnosed Genetic Disorders in Adults with Chronic Kidney Disease (CKD) | *Ali Gharavi (Columbia)*

- This work focused on the relationship between Chronic Kidney Disease and genetic disorders. Specifically, 1.5% of adults with CKD have a diagnosable genomic disorder that may have implications for medical management. Exome sequencing offers the opportunities to define late onset complications of known disorders, identify new mechanisms of injury, and establish their genetic basis.
- Ali presented a study that showed enrichment for 5 genomic syndromes with specific associations. In addition, his study indicated a global association between CKD and neurocognitive phenotypes.
- eMERGE is developing CKD and autoimmunity phenotypes in eIII, as well as developing network wide concept sheets to identify CNV associations with CKD and kidney developmental phenotypes and GWAS/CNV analysis of autoimmunity traits. Further there are three genes on the eMERGE-Seq panel that will serve to confirm the penetrance of known phenotypes and discover new associations.

Sequencing and Genomic Data Update | *Heidi Rehm (Partners/Broad), Eric Venner (Baylor) & David Crosslin (CC/UW)*

- Sequencing Center Update
    - Sequencing Centers oriented the group to the Network's current status in the sequencing ecosystem and provided metrics/summary statistics for samples that have already been sequenced. In addition, sequencing centers addressed sample requirements, and their plan to complete CAP mandated proficiency testing.

        - Partners/Broad has sequenced 1221 samples from the KPW/UW and 91 samples from Geisinger. An example of the variant curation form and CNV output files was presented and will be circulated to sites. The variant curation form will include ALL variants assessed with annotations and interpretation notes. The CNV output file will include all CNVs (note that likely pathogenic/pathogenic CNVs ≥ 3 exons will be reported clinically). Partners/Broad reviewed plans for PGx reporting. Case studies of secondary/incidental findings were presented. eMERGE's VariantWire application was approved, allowing eMERGE sites to view interpreted variants in that network.
        - 406 samples are completely through the Baylor pipeline. The group discussed site specific reporting requirements, which will be accommodated as allowed by assay design, with additional tables added to clinical reports.
- Genomic Data Update
    - eMERGE Imputed dataset: All eMERGE array date including legacy data is being re-imputed against the HRC reference using the Michigan Imputation Server. The total merged set (pre-QC) has a sample size of 85,150. CC will provide the following deliverables to the network:
        - A merged PLINK file with posterior probabilities at .7, .9 or both.
        - A merged VCF file
        - A merged posterior probability file
    - PGRNSeq re-alignment: All ID issues have been resolved and BAMs have been re-aligned to the same reference. The new annotated, QC'd dataset with individual BAMs and VCFs will be uploaded to dbGAP and made available to the network (SPHINX) imminently. The Genomics Workgroup will address privacy concerns and finalize SPHINX private/public side updates by April 2017.

Meharry Medical College (Site Update) | *Samuel Adunyah (Meharry) & Phil Lammers (Meharry)*

- Drs. Adunyah and Lammers presented an overview of the Meharry Translational Research Center (MeTRC), including MeTRC's goal for its current cycle (2014-2019), leadership/governance structure, and background information about Meharry Medical College, specifically noting that it is a historically black medical college and focuses on providing care to underrepresented minorities. In addition, they detailed MeTRC's marketing and recruitment strategies, timeline for recruitment/enrollment/ROR/data analysis and reporting, and current progress to date for achieving their eMERGE goal.
- MeTRC's role within eMERGE is to recruit and obtain blood samples for DNA extraction for germ-line sequencing from up to 500 African American participants with cancer or at high-risk for cancer. Results will be returned to physicians and patients via EMR, and sequencing data will also be shared across the eMERGE sites. As part of their protocol, MeTRC will obtain both RNA and protein from blood samples for additional studies in various projects by Meharry and external investigators.
- Recruitment population: Breast cancer, prostate cancer, colorectal cancer, lung cancer. The goal is to have equal cohorts in each risk sub-category (affected cohort and high-risk cohort).
- Demographics: Age 22-44 represents 42% of their total ~226k population, and age 45-64 represents 30%. Of the total population, 74% identify as Not Hispanic of Latino.

Closing Remarks | *Rex Chisholm (Northwestern/SC Chair)*

- Steering Committee is reminded of the goal to generate eMERGE/CSER joint projects.
- Interim dbGaP submission: The group raised no concerns, therefore it is agreed that the interim dbGaP submission in late summer or early fall is acceptable. The 109 genotype panel will be submitted.
- Marylyn Ritchie (Geisinger) will circulate a concept sheet to run an eMERGE-wide GWAS submission for the Global Lipids Genetics Consortium's next round of global GWAS meta-analyses.

Workgroup Report-Outs

Genomics Workgroup & Geocoding Supplement | *Megan Roy-Puckelwartz (Northwestern), Patrick Sleiman (CHOP) & David Crosslin (CC/UW)*

- Genomics:
  - Deliverables/Timeline for completion:
    - Imputed genotype data: Imputation/merging will be complete March 2017. The group may want to re-visit calling indels in the future, as they will not be included in this dataset.
    - PGRNSeq realignment: The re-aligned dataset will be available to the Network in February 2017.
    - eMERGE-Seq Data: Sequencing will be complete in 2018
    - HLA region investigations: This is a collaboration/group effort and is expected to yield eMERGE deliverables.
    - DNAnexus Tools: Geisinger has already made some tools available and will make project data (input/tools used/output) available for testing. The group will develop more tools for use in the eMERGE analysis pipeline.
    - WGS: 1800 diverse samples are available for analysis, and will be made available to HRC for enhancement of that dataset. The dataset is currently in dbGaP, with the goal of moving it to DNAnexus.
    - SPHINX: The SPHINX resource will be expanded to include realigned PGRNSeq and eMERGE-Seq data. Expanded annotation and the addition of phenotype data are being discussed.
  - Challenges with data generation and analysis have been overcome
  - Foundation for eMERGE IV: eMERGE III will focus on discovery/lessons learned. A tremendous amount of data has been/will be deposited into dbGaP, and eMERGE IV could continue to explore the richness of this data. Developing a core dataset of common demographics and covariates, and generating best practices on implementing a common data model could also be addressed in eMERGE IV.
- Geocoding:
  - Patrick outlined the methods, variables, and address format the Geocoding group will use to produce its deliverables.
    - Centralized vs distributed method. The group is investigating using DNAnexus as a web-based, centralized method of generating and analyzing geocoded data.
    - Environmental variables have been prioritized, and as many as possible will be incorporated.
  - Next steps include defining and testing the analysis pipeline, disseminating the SOP to the group, and depositing demographics/exposure data in a central repository (DNAnexus).

EHR Integration Workgroup | *Sandy Aronson (Harvard/Partners)*

- The EHRI workgroup remains focused on fulfilling the goals of their charter. They have established a core network. Data is flowing into the core repositories, and data is almost at the point of flowing to the individual sites. This flow of data is mediated by a common file format working across the network for moving results.
- The workgroup's next step is to move data into the sites, and ensure that it is properly digested and used at the site-level. Because the data flow will vary across sites, the workgroup is now focused on determining internal workflows and how to best accomplish fulfillment of establishing end-to-end data flow across the network. It is noted that each site will have to utilize a parser and has different clinical/research workflows into the EHR system.
- Science: The workgroup is writing a manuscript on inter-institutional network capabilities thus far; tracking milestones with anticipation of writing a manuscript on the sites' experiences with establishing infrastructure based on structured files; developed a subgroup focused on infobuttons.

- Community: The workgroup is involved with the HL7 Clinical Genomics Working Group, the CSER EHR Working Group (DIGITizE Lynch Syndrome Collaboration, Evaluating Cost of CDS), and Infobutton Work (multiple synergies including with ClinGen)
- Next steps for the workgroup include evaluating site digestion processes and estimating timelines. Members will also discuss eMERGE Phase IV on their next workgroup call.

PGx Workgroup | *Laura Rasmussen-Torvik (Northwestern) & Cindy Prows (CCHMC)*

- The PGx workgroup participated in Clinical Annotation, Genomics, and Phenotyping workgroup breakout sessions. For the report out, Laura gave a brief summary of what was discussed in the other workgroup sessions (described in their respective sections), and gave an update on phenotype and non-phenotype projects being developed by group members.
- Ongoing PGx workgroup activities include tracking CDS activities, collecting additional outcomes data, and developing a PGx analysis pipeline.
- Barriers: Group participation will help overcome barriers.
- PGx reporting in eIII: Sequencing centers will provide signed CLIA reports. The group discussed what individual sites will do with the information on the reports as this could be an opportunity to expand PGx work with varied responses. This depends, in part, on format standards used and if they are compatible with site EMRs. Sequencing centers will provide mock reports to the group when available. The group discussed whether the report is clinical or research oriented and what responsibility/obligation the network has to returning results to patients. This discussion will be continued.
- Information on the overlap of participants in PGx/eI-III will be updated and circulated.

Outcomes Workgroup | *Josh Peterson (CC/Vanderbilt), Hakon Hakonarson (CHOP) & Marc Williams (Geisinger)*

- As a reminder, the Outcomes Workgroup will develop cross-site outcomes to track implementation and impact of eMERGE III sequencing. The workgroup will focus on answering the overarching question of whether returned eMERGE III-generated genomic results impact health care utilization and outcomes of importance to patients and families.
- The workgroup is in Stage 3/3 in its progress towards preparing for outcomes studies, which is to define specific outcomes projects. To facilitate this stage, the workgroup is in the process of developing data collection instruments. The workgroup is focused on prioritizing higher frequency phenotypes across sites to collect outcomes, and will talk further with Columbia about prioritizing Chronic Kidney Disease (CKD).
- The outcome assessment workflow is split between Variant Positive Results (Case, N=~1000) and Variant Negative Results (Control, N=~24000). Variant Positive Results will go through a detailed manual and EMR-based abstraction, whereas Variant Negative Results will run through automated extraction from EHR. Most of this will occur around 6 months after ROR that way there will be patient and EHR data available at the same time point to cross-analyze. Many of these outcomes assessment forms, which are phenotype-specific, will be formatted in REDCap.
- Familial Implications of ROR Subgroup: The subgroup aims to evaluate outcomes related to cascade genetic testing, and working on clear challenges related to consent and accessibility of data on family of a proband. In addition, the subgroup is collaborating with the ROR/ELSI workgroup on and ancillary R01 studies to survey patients.
- Economics: The economics subgroup aims to focus on analysis of standardized costs attached to differences in health care utilization between Variant Positive and Variant Negative cohorts. The challenge is assessing which process measures are attributable to ROR. The economics subgroup also aims to analyze projected savings over time for when health outcomes are expected to change as a result of ROR.
- Pediatrics: The pediatrics subgroup aims to expand its pediatric asthma exacerbation frequency study to a larger cohort, match findings with other eMERGE cohorts and write a manuscript on its current findings within ~470 asthma cases.

Clinical Annotation | *Gail Jarvik (UW) & Heidi Rehm (Partners/Broad)*

- Heid and Gail presented the group's work interpreting and developing return of results protocol for secondary/incidental findings, which have been found in about 3% of each sequencing center's samples.
- The group has developed a feedback loop to interpret variants and finalize return plans for borderline or unanticipated results. Two case studies were presented to illustrate the process and its outcome.
- Most of the group's deliverables have been completed. The group is interfacing with the RoR/ELSI group to formalize eMERGE's role in informing ClinGen priorities and the ACMG list. The group is also providing input into PGx reporting plans.
- Processes and best practices for panel-based actionability determination, variant interpretation, and return of results are expanded products that could inform eIV work.

ROR/ELSI Workgroup & HCP Supplement | *Ingrid Holm (BCH) & Iftikhar Kullo (Mayo)*

- Disclosure of ROR project: Georgia Wiesner (Vanderbilt) and Kathy Leppig (Kaiser/UW) are leading an effort to survey site return of results processes. This will help inform the HCP survey and will also enable compare/contrast of ROR processes across sites.
- Participant Survey Subgroup: This subgroup is trying to harmonize participant surveys across sites in the hopes that sites will be able to collect similar data in order to enable compare/contrast studies.
- IRB Perspectives Project: This project, led by Robyn Fossey (Mayo) aimed to collate interactions with IRBs across sites and formulate lessons learned. A manuscript is in circulation for review amongst the group, and will be submitted shortly thereafter.
- Potential Workgroup Projects for 2017: Fragmented nature of family communication is a topic of interest for the group. Further additional projects include: qualitative analyses of Provider-Participant ROR encounter; Audio vs Videotaping; MyResults.Org expansion; re-contact by phenotype; collaboration with the Networked Biorepositories Project by Kyle Brothers and Aaron Goldenberg.
- Collaborations: The workgroup is aiming to collaborate with the Clinical Annotation Workgroup to address the meaning that participants attribute to results and if this will inform future ROR. Also with the Clinical Annotation Workgroup, the workgroup aims to address how patients and providers react to variant reclassification. The workgroup also aims to collaborate with the Outcomes Workgroup on the time of ROR, costs and healthcare utilization, and personal costs that the patients might incur as a result of ROR. With CSER, the workgroup aims to harmonize surveys with the CSER network as well as clinical education, and also provide input on the ACMG59.
- Health Care Provider (HCP) Supplement: This is a one-year supplement funded by the NHGRI ELSI Branch. The aims are 1) to develop a survey that will elicit the preferences of HCPs who are receiving genomic information as part of eMERGE Phase III, and 2) to pilot test the survey in a subset of HCPs at two of the eMERGE III sites, analyze that data, and finalize the survey for administration in the eMERGE III Network. Methods include literature reviews, interview of HCP to inform survey, survey development, cognitive interviews and piloting the study. The group has developed domains to structure survey and a timeline. Since the group was only funded to develop, and not implement, the survey, the group is working on submitting an R01 for implementation funding.

Phenotyping Workgroup | *George Hripcsak (Columbia) & Peggy Peissig (Marshfield)*

- The group reviewed the current status of Phenotype development and reimplementation. While progress was made, the group is concerned about the number and complexity of the phenotypes left to develop and implement.
    - Recommendation: each site develops 3 eIII phenotypes and implements 5-7 PGx Phenotypes.
        - The group will re-prioritize remaining phenotypes and generate a new timeline. Genomics workgroup representation at Phenotyping meetings is encouraged to assist in identifying common project interests.
        - The group will consider a new non-sequential and iterative workflow.

        - The group will simplify phenotypes and data dictionaries.
        - The group will develop a core set of covariates
        - Commitment to timelines is essential. The group expects to complete phenotype development by August of 2018 and will complete implementation in early 2019.
- Deliverables/timelines:
    - Phenotypes (Ongoing)
    - dbGaP Submissions (July 2017 and May 2019)
    - CardioCore resource (2018)
    - SPHINX and eRC data updates (ongoing)
    - Manuscripts
- Barriers:
    - Expanding scope of phenotypes (quantity and complexity)
    - Prioritization
    - Commitment to timelines
- Foundation for eIV:
    - Common Data Model validation and best practices. This could be accomplished with other funding.
    - New phenotype development workflow.
- How to make one consistent view of clinical conditions (phenotype) across case/control, covariate, and/or outcome status.

Summary of Action Items

1. Sequencing centers will complete CAP proficiency testing in March 2017.
2. Sequencing centers will complete pharmacogenomic pipeline validation/report format and begin issuing reports by April 2017.
3. Baylor will finalize their pipeline for submitting variants to ClinVar.
4. The Genomics workgroup will complete the imputed genotype dataset by March/April 2017 and make it available to the Network.
5. The Genomics Workgroup will complete QC on the re-called PGRNSeq dataset and make it available to the Network by March 2017.
6. The Genomics workgroup will finalize the data management pipeline for the eMERGE-Seq dataset by June 2017.
7. The Genomics workgroup will continue to develop the data analysis pipeline and tools available in DNAnexus (no timeline, continuous as needed development).
8. The Genomics workgroup will finalize SPHINX enhancement plans by the end of first quarter.
9. The Geocoding subgroup will define and test and gene x environment analysis pipeline.
10. The Geocoding subgroup will disseminate standard operating procedures for gene x environment analyses.
11. The Geocoding subgroup will deposit a dataset of demographic/environmental exposure in a central repository accessible by the Network.
12. The EHRI workgroup will complete its paper (NT184) on inter-institutional network capabilities that have been established thus far by May 2017.
13. The EHRI workgroup will track milestones in anticipation of formulating a manuscript by February 2018 depicting the experience of establishing infrastructure based on structure file based result transfer to sites.
14. The EHRI workgroup will continue collaboration with CSER's EHR working group on DIGITizE Lynch Syndrome, with an anticipated completion date of December 2017.

15. The EHRI workgroup will continue collaboration with CSER's EHR working group on evaluating cost of CDS, with an anticipated completion date of December 2017.
16. The PGx workgroup will work with the Phenotyping workgroup to integrate and prioritize PGx related phenotypes in the overall phenotype prioritization list and timeline. (Done)
17. The PGx workgroup will work with the Clinical Annotations workgroup and sequencing centers to develop a pipeline for return of PGx results in eIII (including providing input on what is on reports, coordinating return plans across the network).
    a. Laboratory representatives will provide updates and receive input on pipeline and reporting mechanisms. This began with 2/21/17 workgroup call.
        i. Both sequencing centers expect to begin delivering PGx data to sites in April 2017.
    b. Sites' ROR plans initially discussed during 2/21/17 workgroup call.
    c. Will present draft tracking sheet for sites' ROR plans at March 2017 meeting
    d. Will implement tracking sheet during Spring 2017 that sites can update as their PGx projects evolve
18. The PGx workgroup will create an analysis pipeline for common/rare PGx variant analysis on DNAnexus.
    a. Discussed during 2/21/17 workgroup call. Geisinger has an analysis structure for common and rare variants already developed in DNAnexus and can open it up to rest of the network.
        i. To provide demonstration at future call – targeting workgroup session before next SC
    b. Will discuss testing once PGRNseq realignment of BAMs and recalling of VCFs completed (anticipate testing to begin in Summer 2017)
19. The Outcomes workgroup will evaluate outcomes related to cascade genetic testing, specifically working on clear challenges related to consent and accessibility of data on family of a proband. This will be facilitated through their Familial Implications of ROR Subgroup.
20. The Outcomes workgroup will collaborate with the ROR/ELSI workgroup and ancillary R01 studies to survey patients.
21. The Outcomes workgroup will address analysis of standardized costs attached to differences in healthcare utilization between variant positive and variant negative cohorts. This will be facilitated through their Economics Subgroup.
22. The Outcomes workgroup will address analysis of projected savings over time for when health outcomes are expected to change as a result of ROR. This will be facilitated through their Economics Subgroup.
23. The Outcomes workgroup will replicate a pediatric asthma exacerbation frequency project on a larger cohort and match findings with other eMERGE cohorts. This will be facilitated through their Pediatrics Subgroup.
24. The Outcomes workgroup will finalize outcome assessment tools within next 3 months.
25. The Outcomes workgroup will conduct phenotype specific analysis in the next 12-24 months.
26. The Outcomes workgroup will conduct global outcome analyses in 24 months.
27. The Clinical Annotation and PGx Workgroups will work with sequencing centers to ensure pharmacogenomics reporting approaches are consistent with site needs.
28. The Clinical Annotation and PGx workgroups will seek consensus for returning structured pharmacogenomic data to sites so that it is consumable and conforms to informatics standards.
29. The Clinical Annotation workgroup will interface with the RoR/ELSI group (develop feedback loop from RoR studies to inform criteria/genes lists, formalize role of eMERGE and CSER in informing ClinGen priorities and ACMG list).
30. The ROR/ELSI workgroup will survey healthcare providers across sites to assess the impact of ROR on HCPs post-disclosure. This will be facilitated through their HCP Supplement Subgroup.
31. The ROR/ELSI workgroup will complete surveys of participants at baseline, post-disclosure and 6 months after disclosure. This will be facilitated through their Participant Survey Subgroup.
32. The ROR/ELSI workgroup will coordinate with the Outcomes Workgroup via the Familial Implications of ROR subgroup to address familial implications from the participant perspective.

33. The ROR/ELSI workgroup will complete a manuscript on IRB Perspectives across eMERGE.
34. The ROR/ELSI workgroup will complete a manuscript on the various process of ROR across eMERGE sites.
35. The ROR/ELSI workgroup will collaborate with the Clinical Annotation workgroup to address the meaning that participants attribute to results.
36. The ROR/ELSI workgroup will collaborate with the Clinical Annotation workgroup to address how patients and providers react to variant reclassification.
37. The ROR/ELSI workgroup will collaborate with the Outcomes workgroup to address the timing of ROR, costs and healthcare utilization, and personal costs.
38. The ROR/ELSI workgroup will assess the feasibility of potential additional projects discussed at the in-person meeting.
39. The Phenotyping workgroup will develop an overall phenotype prioritization list (including eIII, PGx, and eI/II phenotypes) and timeline for completion by the end of April 2017.
40. The Phenotyping Workgroup will develop a coordinated set of phenotypic variables that will form the base set of analysis variables used for all electronic algorithms. Data dictionaries will be kept succinct with few if any additional variables. This is anticipated to be defined/implemented by the June 2017 Steering Committee Meeting.
41. The Phenotyping Workgroup will define the phenotypic variables, led by David Crosslin, for inclusion in an interim dbGaP submission in July 2017.
42. The Phenotyping Workgroup will support SPHINX and eRC quarterly data refresh efforts.

Next Meeting: June 8th and 9th, 2017 in Boston, MA

emerge network
ELECTRONIC MEDICAL RECORDS & GENOMICS