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eMERGE & Beyond: The Future of Electronic Medical Records (EMR) and Genomics
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Executive Summary

Introduction

The Electronic Medical Records and Genomics (eMERGE) network conducts genomic discovery and clinical implementation research by leveraging data from large biorepositories linked to electronic medical records. On October 30th, NHGRI hosted a workshop entitled "eMERGE and Beyond: The Future of Electronic Medical Records (EMR) and Genomics." The workshop reviewed the current goals and accomplishments of eMERGE and suggested future directions for a possible continuation of eMERGE. It was attended by external experts in key areas of genomic medicine, including members from the National Advisory Council for Human Genome Research, the eMERGE Network, and the private healthcare sector. The workshop was divided into 4 special topics for discussion: (1) Electronic Phenotyping for Genomic Research, (2) Evidence Generation for Genomic Medicine, (3) EMR Integration of Genomic Results and Automated Decision Support, and (4) Novel and Disruptive Opportunities in Genomic Medicine. To identify concepts and gaps of interest in the field of Genomic Medicine that a potential new phase of eMERGE could address, each section included an eMERGE presentation, a reaction, discussion, and a consensus summary of how a new phase of eMERGE could address critical gaps. The bulleted list below is based on these discussions, and provides recommendations for each of the aforementioned special topics.

Electronic Phenotyping for Genomic Research

- Focus on developing better phenotyping methods and technologies, such as:
  - Increasingly automated phenotyping
  - Longitudinal phenotyping
  - Continuum of disease severity rather than binary disease absent/present
  - Incorporating information on the time course of conditions to create more accurate phenotypes
  - Alternate approaches to manual phenotype validation
- Focus on fewer phenotypes and experiment with alternative phenotyping strategies to improve speed and efficiency
- Find more efficient ways to pool, normalize and analyze data across all eMERGE sites

Evidence Generation for Genomic Medicine

- Improve capture of standardized family history data across all sites; develop apps for collecting family history and incorporate the information into the EMR
- Standardize or synthesize different study designs including ROR decisions because currently there is a wide spectrum of study designs across the different institutions of eMERGE, which reduces sample size and impairs joint analysis
- Seek appropriate balance between standardization and experimentation with different study designs since there is not yet enough information to standardize many aspects
- Develop and document best practices from the studies already completed in eMERGE

- Create data standards for new types of genomic medicine “data objects”, such as genome sequencing data VCF formats, which incorporate quality metrics
- Study the value and impact of reporting negative results and study the definition of negative results in different contexts
- Assess longer-term outcomes of testing and results reporting

EMR Integration of Genomic Results and Automated Decision Support

- Develop tools and standardized displays to synthesize and present information at the point of care so that physicians do not have to hunt for information to make a decision
- Develop user-centered designs through both display-based and event-based eCDS
- Build foundations that promote shareable eCDS, which includes the knowledge representation of complex CDS and enhancement of existing knowledge repositories
- Narrow the scope of eMERGE to developing CDS in a few specific, high-priority clinical areas to avoid spreading resources too thin
- Develop closed-loop CDS, which contains automated outcome assessment tools and allows determination of whether users followed the guidance to assess utility
- Explore patient-specific factors that might increase or decrease the importance of genomic information in the CDS to improve relevance of alerts to specific patients
- Evaluate the effect of standard and nonstandard approaches to delivering results on physician-patient relationships including what information can be disseminated using alternative technologies vs. what needs to be conveyed through a genetic counselor
- Address scaling of phenotyping/interpretation/reporting as its own research problem
- Develop roadmap for naïve adopters of genomic data/CDS implementation in EMR
- Develop standard extract of EMR for research

Novel and Disruptive Opportunities in Genomic Medicine

- Incorporate genomic data derived or inferred from external sources, such as patient-contributed data, environment (geocoding), direct-to-consumer data, social media, “peer-to-peer phenotyping” (patients sharing their clinical manifestations via social media), family history, and online disease-focused patient communities
- Perform real time variant interpretation that incorporates patient data as well as matches publicly available knowledge sources to the patient’s variants
- Develop methods to efficiently re-interpret genomic results over time
- Develop methods to automate interpretation via analysis pipelines
- Enhance clinical methods for assessing pathogenicity and variant penetrance
- Link EMR-derived phenotypes with other classes of -omics data
- Apply deep learning techniques to the characterization of VUS, drug targets, and toxicity predictions associated with primary genomic data
- Encourage patient-centered data governance and develop or encourage development of apps for patient self-phenotyping
- Develop innovative ways to present sequence information to general physicians, especially those early in training, by identifying problems that physicians are facing and creating pragmatic solutions
- Evaluate the legal and ethical implications of directly contacting relatives of patients potentially harboring deleterious variants rather than having to go through the patient
- Assess crowdsourcing of variant interpretation

The workshop presentation slides and video recordings have been posted online at https://www.genome.gov/27569445/.