JAMIA Call for Papers: Special Focus Issue on Electronic Health Records-Driven Phenotyping

Guest Editors

- Jyotishman Pathak (Mayo Clinic, Rochester, MN, USA)
- Joshua C. Denny (Vanderbilt University, Nashville, TN, USA)
- Abel N. Kho (Northwestern University, Chicago, IL, USA)

Description of the Special Issue

The identification of patient cohorts for clinical and genomic research is a costly and time-consuming process. This bottleneck adversely affects public health by delaying research findings, and in some cases by making research costs prohibitively high. To address this issue, the leveraging of electronic health records (EHRs) to identify patient cohorts has become an increasingly attractive option. With the rapidly growing adoption of EHR systems due to Meaningful Use, and linkage of EHRs to research biorepositories, evaluating the suitability of EHR data for clinical and translational research is becoming ever more important, with ramifications for genomic and observational research, clinical trials, healthcare delivery research and comparative effectiveness studies.

A key component for identifying patient cohorts in the EHR is to define inclusion and exclusion criteria that algorithmically select sets of patients based on stored clinical data. This process, commonly referred to as EHR-driven phenotyping, allows the definition of phenotypes over structured data (e.g., demographics, diagnoses, medications, and laboratory measurements) as well as unstructured clinical text (e.g., radiology reports, encounter notes, discharge summaries). In general this process can be quite complex, involving heuristics encoded as rules or machine learning algorithms. The goal of this special issue of the Journal of American Medical Informatics is to provide a forum for presenting methodologies, tools, and algorithms to enable high-throughput phenotype extraction from EHR data. Submissions from outside the USA describing generalizable experiences are highly encouraged.

Topics of Interest

Possible topics include, but are not limited to:

- Novel architectures for facilitating high-throughput phenotyping from EHRs
- Natural language processing (NLP) and text mining methods for phenotype extraction
- Cost-benefit analyses of electronic phenotype identification in healthcare settings
- Evaluation of transportability of phenotyping methods and reuse of existing phenotype algorithms across different healthcare settings, EHR systems, and component systems (e.g., different NLP systems when applied to EHR data)
- Standards-based representation and modeling of phenotyping algorithms
- Machine learning approaches to developing and validating phenotyping algorithms
- Data mining of EHR data for clinical discovery
- Methods and platforms for semi-automated execution of phenotyping algorithms
- State-of-the-art surveys in phenotyping methods, tools and techniques
- Secondary uses of EHR data for genomics, epidemiology, comparative effectiveness research, healthcare quality and deliver research, clinical decision support, etc.
- Evaluation of EHR data extraction accuracy (e.g., accuracy of structured billing codes) and data cleaning methods
- Quality of patient-reported outcomes that are incorporated into the EHR, data harmonization across various healthcare institutions

- Comparison of different EHR systems in terms of how various coding systems can be accommodated, how flexible the systems are in allowing users to structure clinical information, etc.
- Ethical aspects of utilizing EHR for research, informed consent practices, standardized data use agreements, differences in regulations across institutions, states and countries

Authors should make sure to place their work in the context of biomedical research or healthcare, and to carefully review the relevant literature. *Research articles, case studies*, and *brief communications* should describe clear evaluation strategies and quantitative or qualitative results, and discuss how results could be generalized to other settings. *Reviews* should be systematic. *Perspectives* should provide consensus of a group of experts who are highly experienced in the topic, and should demonstrate command of the existing literature. Open-source software code and data should be submitted, as well as data when appropriate.

Important Dates

~~March 31, 2013~~ April 15, 2013: Manuscript submission deadline
May 31, 2013 (expected): Initial decisions sent to authors
June 30, 2013 (expected): Revised manuscript submission deadline
August 31, 2013 (expected): Final decisions sent to authors

Submission and Peer Review Process

To ensure consideration in the special issue, authors should note in a cover letter that their submission is for the "Special Issue on Electronic Health Records-driven Phenotyping". Detailed information for online submission to JAMIA is available via http://jamia.bmj.com.

All manuscripts will be subject to the rigorous JAMIA peer-review process. Manuscripts that are considered within scope and meet quality expectations will be reviewed by a minimum of two experts for scientific merit. Assistance of a native English speaker is highly recommended prior to submission. Authors should format and structure their manuscripts according to the guidelines specified at: http://jamia.bmj.com/site/about/guidelines.xhtml. Accepted articles may appear in print or in an online JAMIA issue.

Questions Regarding the Issue

Please direct any questions regarding the special issue to jamia-phenotyping@googlegroups.com.