Foundational biomedical informatics research in the clinical and translational science era: a call to action

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ABSTRACT

Advances in clinical and translational science, along with related national-scale policy and funding mechanisms, have provided significant opportunities for the advancement of applied clinical research informatics (CRI) and translational bioinformatics (TBI). Such efforts are primarily oriented to application and infrastructure development and are critical to the conduct of clinical and translational research. However, they often come at the expense of the foundational CRI and TBI research needed to grow these important biomedical informatics subdisciplines and ensure future innovations. In light of this challenge, it is critical that a number of steps be taken, including the conduct of targeted advocacy campaigns, the development of community-accepted research agendas, and the continued creation of forums for collaboration and knowledge exchange. Such efforts are needed to ensure that the biomedical informatics community is able to advance CRI and TBI science in the context of the modern clinical and translational science era.

Over the past decade the health and life sciences communities have experienced a marked and dramatic shift toward translational and team science-based approaches to both basic and applied research.1,2 This transition is due in part to policy and funding initiatives at the national level, such as the clinical and translational science award (CTSA) program. A common theme spanning this evolution is recognition of the critical need to apply biomedical informatics theories and methods to enable the collection, exchange, management, analysis and dissemination of multidimensional datasets and knowledge collections. For example, complex clinical phenotype data describing large populations must be integrated with similarly large amounts of genomic data in order to support the identification of clinically relevant phenotype-genotype correlations. These types of needs have catalyzed an explosion of informatics research and development targeting the clinical and translational research domains. Such efforts have enabled numerous advancements in clinical and translational research informatics knowledge and practice. However, at the same time, the maturation of clinical research informatics (CRI)1 and translational bioinformatics (TBI)3 is at risk of failing to meet expectations if commensurate foundational research in those same areas is not conducted.

‘LIVING LABORATORIES’ FOR CLINICAL AND TRANSLATIONAL RESEARCH INFORMATICS

Applied CRI and TBI research is being performed in a variety of ‘living laboratory’ settings, as represented by the following exemplary instances:

- The National Cancer Institute’s caBIG program was launched in 2004 with the goal of developing an informatics infrastructure for multisite research and data sharing.4 In response to this objective, informatics researchers involved in caBIG have developed a variety of service-oriented data-sharing platforms, research data management tools and knowledge management systems.

- The National Center for Research Resources (NCRR) CTSA program began in 2006, with the aim of establishing a network of academic health centers, each with a scholarly ‘home’ for clinical and translational science.5 From the outset, biomedical informatics expertise and resources have been considered critical components of each site’s efforts to achieve such an aim. The informatics programs of most CTSA sites focus on activities such as: informatics training; consultative services; database design/hosting; data warehousing; data sharing infrastructure and the execution of complex data analyses.

- Throughout the National Institutes of Health (NIH) funding portfolio, there is a variety of single and multisite programmatic research initiatives, focusing on specific disease areas. These programs regularly include informatics and data management teams that develop and support clinical and translational research informatics infrastructures. Examples of such efforts include the National Cancer Institute-funded Chronic Lymphocytic Leukemia Research Consortium, National Center for Research Resources-funded rare diseases clinical research network and NIH-funded osteoarthritis initiative.

Additional examples of such ‘living laboratories’ include the NIH-funded National Centers for Biomedical Computing and Biomedical Information Science and Technology Initiative Programs. The defining characteristic of the aforementioned applied informatics activities is a major emphasis on: the development of production-grade data-sharing and management tools that address well-defined end-user requirements; and short-term or indirect (e.g., science-focused) funding cycles that prioritize technical deliverables.

THE BIOMEDICAL COMMUNITY: A CALL TO ACTION

We believe that the preceding examples of ‘living laboratories’ for CRI and TBI research raise a number of re-occurring concerns. Directed by funding and policy agendas, these efforts have largely focused on solving immediate, pressing and application-oriented needs, without a complementary focus on enhancing underlying foundational
CRI and TBI knowledge and methods. Such an issue is particularly problematic in settings in which the primary emphasis is on specific disease domains, which often relegate informatics to a purely service-oriented role. Such phenomena has the potential to lead to several problematic outcomes, including: the creation of informatics ‘silos’ that correspond to sources of funding or scientific endeavor, rather than enabling foundational research; and the restriction of such efforts to a subset of universities and centers. Both of these concerns lead to limitations on the ability of informaticians to ask and answer a wide array of foundational research questions relevant to the clinical and translational sciences.

Given the preceding concerns, we feel that the informatics community should and must critically evaluate the state of foundational CRI and TBI research, engage with the larger research community, and take a number of steps to catalyze the foundational research needed to advance the state of CRI and TBI science. Such steps would ideally include:

1. A comprehensive and rigorous campaign of advocacy to ensure that advancing foundational CRI and TBI knowledge and practice is both recognized and supported as a core objective of transformational changes occurring in the clinical and translational research domains. Such advocacy should seek to engage informaticians as equal partners in the planning and execution of such efforts, rather than as service providers responding to a predetermined mandate. Targets of this campaign would include academic and professional leaders, policy makers, funders and scientific communities with whom such partnerships would ideally be conducted.

2. Community mobilization in order to refine and promote a national-scale agenda focused on the critical challenges and opportunities facing the CRI/TBI domains, and to shape the trajectory of related research, development, funding and training activities. This agenda should prospectively guide the CRI/TBI community, rather than continuing our current state of primarily being reactive to emergent funding or policy agendas.

3. Forums must be established to ensure that the preceding activities are not limited to those few institutions, investigators, or thought leaders who are funded by high profile initiatives. The dialog surrounding the advancement of CRI and TBI should be conducted in a manner that is open to all informaticians and researchers with relevant interests or expertise, without regard to institutional affiliations or resources. The recently concluded joint AMIA research summits are a good step in this direction.

Ultimately, we believe that these recommendations provide the core path toward establishing a robust, broad-based and productive CRI and TBI community in the emergent clinical and translational science era. It is our hope that this commentary will serve as a call to action to energize our community and colleagues to achieve such a laudable aim.

Funding The efforts of PROP in the preparation of this editorial commentary were supported in part by grants from the NIH/NCI (P01-CA081534, R01-CA134232) and NIH/NCRR (UL1-RR025755). The efforts of PJE in the preparation of this editorial commentary were supported in part by grants from the NIH/NLM (R01-LM009533) and the NIH/NCRR (UL1-RR026314). The efforts of JN in the preparation of this editorial commentary were supported in part by contracts/grants from the NIH/NCI (1435-04-04CT-73980, Subcontract 79208CBS10) and NIH/NCRR (U42 RR017673).

Provenance and peer review Not commissioned; externally peer reviewed.

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