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# Journal of Biomedical Informatics

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Call for Papers

## Special Issue on Methodologies for Translational Bioinformatics

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 Submission Deadline: August 1st, 2009

In keeping with the shift in bioinformatics focus for JBI to translational bioinformatics, we are pleased to solicit papers for a special issue of the journal that will appear in 2010. The issue will focus on original methodological research papers in the area of translational bioinformatics. We define translational bioinformatics broadly as in the AMIA Strategic Plan [1]. Papers submitted to the special issue accordingly should include methods for dealing with (i) molecular-level data (genes, gene products, macromolecules, chromosomes or other subcellular components in human cells) and/or (ii) clinical-level data (clinical signs and symptoms, diseases, clinical procedures or medications).

With the completion of human and model organism genomes, and the increasing utilization of biomedical computational methods, translational bioinformatics is evolving rapidly into a complex multidisciplinary field overlapping with nearly all areas of biological, biomedical and clinical research. Indeed, key factors have synergistically brought about momentous opportunities for computations in the era of molecular medicine [2]: (i) the availability and cost reduction of molecular measurements that are improving at an accelerated pace, (ii) the public availability of large datasets of molecular measurements in diseases states (e.g., GEO), (iii) the culture of sharing molecular data and tools, (iv) the increasing expectation for clinicians critically to interpret discoveries in molecular medicine that appear in clinical journals, and (v) the increase in research funding in translational bioinformatics (e.g., NIH Roadmaps, Clinical and Translational Science Awards). Biomolecular and clinical informatics plus statistical genetics and genomic medicine – the key areas of translational informatics research – are thus jointly poised to play an increasing role in accelerating the translation of knowledge discovery from genome scale studies to effective treatment and tailored disease management or prevention. Further, this is truly an extraordinary time for translational biomedical informatics research and development because some discoveries will lay the foundations of molecular medicine for decades to come. Undeniably, translational bioinformatics may even lead to a pivotal moment in history with the fundamental discovery of the molecular underpinnings of diseases, which will in turn identify novel targets for therapies that could manage complex diseases. The past success of biomedical informatics in transforming medical research and patient care is merely a glimpse into the remarkable promises that the field holds in an era of accelerated advancements in molecular medicine.

This issue is opportune to exemplify the research efforts and to capture the progress in the application of translational bioinformatics research methods and tools in molecular medicine, genetics and translational clinical trials. In an effort to accelerate translational bioinformatics research and applications, the American Medical Informatics Association, in partnership with International Society for Computational Biology, has been holding an annual Summit on Translational Bioinformatics since 2008. This special issue will further highlight the multidisciplinary nature of this research field and provide a unique opportunity to bring together novel methodological proposals in translational bioinformatics from researchers in the fields of biomolecular and clinical informatics, statistical genetics, computational biology, and computer science. We hope that this special issue will serve as a catalyst to foster subsequent multidisciplinary research and scientific exchanges in translational bioinformatics.

Here are examples of responsive submissions to this special issue:

1. *Informatics methods for the analysis of molecular and clinical measurements.* Novel modalities for molecular measurements, including those for epigenetics, interactions, and proteins, continue to be introduced each year. These submissions should address how the development of novel analytic methods for these molecular measurements are applied to disease, how molecular measurements can be stored and retrieved in electronic health records, and how novel or existing analytic methods can now be applied to clinical measurements.
2. *Computational approaches to finding molecular mechanisms and therapies for disease.* DNA sequencing, gene expression microarrays, and other tools measuring outputs of the genome have been proposed to enable the discovery of biomarkers for disease and novel types of therapeutics. The submissions should focus on how data-driven, knowledge-driven, and physics-driven approaches can be applied to facilitate medication and biomarker discovery.

3. *Informatics concepts, tools, and techniques to enable integrative translational research.* Integrative translational research projects make coordinated use of molecular (genomic, epigenetic, proteomic, etc.) as well as clinical information (imaging, EMR, etc.) to understand pathophysiology and to understand and predict responses to therapeutic interventions. Studies of this kind are core components of many NIH Roadmap projects and aim to relate molecular and imaging parameters to the resulting pathophysiology and clinical parameters. These interdisciplinary submissions can propose informatics methods or tools in the semantic modeling, grid, natural language processing, information integration, and information warehouse areas that have potential for facilitating integrative translational research. System descriptions are welcome if they illustrate and substantiate the underlying methodology that is the principal focus of the report.
4. *Relating and representing molecular or subcellular phenotypes with relevance to the clinical levels (disease, clinical procedures and medications).* Phenotypes broadly describe the unique traits of an organism, some of which are related to disease. Improved measurement technologies and ontologies have enabled investigators to obtain and represent large collections of phenotypes, but relating these to clinical and health remains a challenge. Though electronic health records are being increasingly adopted, much of the useful phenotypic and clinical descriptors remain in free-text. Submissions should address these challenges, with representations geared towards enabling novel prognostic, diagnostic, and therapeutic applications of molecular and genetic medicine.
5. *Dissecting disease through the study of organisms, evolution, and taxonomy.* Over a quarter million different species have had some genetic sequence obtained. While animal and cellular models have long been studied as a proxy for human disease, this type of paper will address the challenge of building informatics methods to relate experimental, molecular or genetic findings and phenotypes from animal models to human disease, and methods that take advantage of the evolutionary scale of sequenced genomes. Such papers will also cover modeling of the spread of infectious disease.
6. *Informatics methods in genetics discoveries and clinical practice.* In the past two years, over 250 disease genes have been discovered and characterized from genetic studies, the majority of which are associated with complex diseases such as diabetes, hypertension, cancer, etc. Consequently, molecular bioinformatics methods are increasingly considered for the identification of additional disease gene candidates. Further, with the reducing cost of genome-wide sequencing or assaying, new opportunities are emerging to provide personalized genome maps and their interpretation. Responsive submissions could focus on (i) the development of bioinformatics methods for disease gene discoveries in genetic studies, or (ii) the translation and evaluation of post-genomic knowledge in decision making and support, electronic records and other genetic applications to clinical practice and personal health management.

#### Peer-Review Process

All submitted papers will go through a rigorous peer-review process with at least two reviewers. The acceptance process will focus on those papers that address innovative methods in translational bioinformatics. All submissions should follow the guidelines for authors available at the *Journal of Biomedical Informatics* web site (<http://www.elsevier.com/locate/yjbin>). JBI's editorial policy is also outlined on that page and will be strictly followed by special issue reviewers.

#### Submission Process

Authors must submit their paper via the online Elsevier Editorial System (EES) at <http://ees.elsevier.com/jbi>. Authors can register and upload their text, tables, and figures as well as subsequent revisions through this website. Potential authors may contact the Publishing Services Coordinator in the journal's editorial office ([jbi@elsevier.com](mailto:jbi@elsevier.com)) for questions regarding this process.

#### References

- [1] American Medical Informatics Association. AMIA Strategic Plan. 2006. <http://www.amia.org/inside/stratplan/>; accessed Mar 1, 2009.
- [2] Butte, AJ. Translational Bioinformatics: Coming of Age. *J Am Med Inform Assoc.* 2008;15(6):709–714.