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Preface

Towards Clinical Bioinformatics

Once again, the editors of the Yearbook of Medical Informatics 2004 have assembled an impressive collection of papers that capture the state of our field. The papers provide a focused set of intellectual contributions ranging widely from the precise mathematical models required for signal and image processing to the symbolic knowledge representations required to capture complex biomedical concepts. However, taken as a whole, the papers also provide a tantalizing summary of the challenges facing biomedical informatics in 2004. These challenges include internal challenges to the field, and external challenges from the biomedical enterprise.

The field of biomedical informatics has seen an explosion of interest in the last decade, and that has led to an explosion of sub-disciplines. The diversity across the sections of this Yearbook is striking, both in the intellectual problems that they approach, the methods that they use, and the communities which benefit from their successes. Gone are the days when a motivated investigator could keep track of the entire field, and have a good understanding of the technical challenges in each sub-discipline. Investigators must now be content with detailed knowledge of

their own field, and often must lose track of those that are not “critical path” in their own work. In this context, the Yearbook becomes more important because it offers a carefully selected collection of papers that allow today’s investigator to conveniently assess the challenges and progress made. The editors have thus given those of us who still want to be “informatics generalists” an opportunity to see how things are going in closely-related disciplines. Bringing together these papers in one volume provides a special service both to those of us who remember the days when the field was small, as well as to newcomers who benefit from seeing the intellectual context in which the individual sub-disciplines now exist.

The external challenges to the field are reflected in the categories chosen for the sections. Many of the sections could be predicted as long-standing “bread and butter” topics in medical informatics. I am struck, however, by two categories which would not have appeared ten years ago: “Towards Clinical Bioinformatics” and “Education and Consumer Informatics.” I believe that these two sections owe their existence to two important and relatively recent developments: the rise of bioinformatics and the rise of the world wide web.

Bioinformatics has exploded onto the informatics scene as a result of high throughput data collection techniques that create large datasets beyond the manual processing capability of humans. These techniques include DNA sequencing, micro-array expression measurements, proteomic techniques and others. The excitement of basic biological discovery has drawn the attention of many young scientists, and has led to the rapid development of new databases, search techniques, pattern matching algorithms and knowledge representations. These technologies have already accelerated our understanding of basic biology. But with the completion of the human genome, there is now a mandate to translate the basic science of genomics into clinically useful tools for diagnosis and treatment. In many cases, traditional clinical informatics practitioners have joined forces with their bioinformatics colleagues to approach the problem of supporting “genomic medicine” with informatics. These collaborations for the most part have been extremely rewarding for both sides, and have shown our non-informatics colleagues in biology and medicine that the methodological focus of informatics provides a common core that allows informatics researchers to more fluidly move between biomedical disciplines. Thus, I am excited to see a section on clinical bioinformatics, as evidence that we are looking beyond traditional application-domain boundaries to do what is needed in advancing clinical care.

The world-wide-web is hardly news, but the emergence of consumer informatics can clearly be traced to the popularity of the WWW as a source of health information. Many studies have shown that health care information is one of the most common uses of the web, and there is a growing literature about the challenges in helping patients manage complex technical information. There is also a need to help consumers separate high quality information from information designed to sell things. Many predictions about medicine in the future are dominated by an assumption that consumers will use web resources to generate health questions and to make health decisions. The more radical scenarios have healthcare turned upside-down with a healthcare market in which purchase decisions for individual medical services are made through a competitive bidding process similar to the online auction sites that exist for goods. No matter how things turn out, it is clear that the web as a source of information and as a method for communication about healthcare is here to stay. Thus, it is entirely appropriate that the medical informatics community develop a high quality literature on the challenges and technologies that facilitate web-based health education and decision-making.

But it is the combination of these two informatics threads that intrigues me most. If medical information is abundant and complex now, can we imagine the challenges facing consumers as genomic medicine becomes a

reality? My patients come to me now because they have a headache and they are worried about a brain tumor. I evaluate them, and most often wind up explaining basic probabilities, differential diagnosis, and the value of watching symptoms over time. What will I do when my patients come to me because they have had their genome sequenced, and they are worried that they are at risk for brain tumors, due to a genetic polymorphism that they saw related to brain tumors on a web site? The conversation will be substantially the same, but will be much more manageable if both clinical informatics and consumer informatics can provide 1) accurate knowledge of the basic biology associated with different genetic backgrounds, and 2) good techniques for helping inform health-care consumers about rational decision making using this information. It is likely that all the other areas of informatics represented in this Yearbook (I have only highlighted two) will also directly impact that conversation with my patients. Thus, it is with a great sense of satisfaction that I introduce the Yearbook of Medical Informatics 2004, which provides us, as informatics generalists, an encouraging snapshot of intellectual progress within medical informatics.

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