Clinical Bioinformatics: new challenges for human-centered decision-support systems

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Abstract. The panel deals with the challenges related to building new decision-support systems for clinicians in the genomic medicine era. The definition of new tools is becoming mature as we seek to integrate the diverse and disparate sources of information available to clinicians (clinical data, -omics information, literature search, knowledge repositories) in such a way that decisions are effectively improved. The complexity of the problem needs an interdisciplinary approach, whereby artificial intelligence, information integration, and cognitive sciences should cooperate to structure information and knowledge, to help reasoning, and to improve decision-making capabilities at the point of care.

Keywords. Clinical bioinformatics, decision support, data integration, interoperability, cognitive science

Introduction

Clinical Bioinformatics (CBI) has recently emerged as “the clinical application of bioinformatics-associated sciences and technologies to understand molecular mechanisms and potential therapies for human diseases” [1]. CBI thus emphasized the clinical impact that molecular medicine may potentially have if the necessary information is properly made available for decision-making. In this sense, CBI truly deals with the challenge of integrating molecular and clinical data to accelerate the translation of knowledge discovery into effective treatment and personalized medicine.

Observers have suggested that CBI should assist clinicians in a variety of contexts, including clinical genomics (biomarker discovery), genomic medicine (identification of genotype/phenotype correlations), pharmacogenomics, and genetic epidemiology at the point of care [2]. CBI thus needs to merge bioinformatics with decision-support systems and automated reasoning - areas where biomedical informatics and artificial
intelligence in medicine have a widely recognized and long-standing expertise. Moreover, the design of CBI-based systems to be used by clinicians and practitioners needs to take into account the cognitive and socio-technical aspects of users. CBI is therefore at the confluence of different disciplines, and may foster the definition of a comprehensive framework to deal and manage all kinds of biomedical data, supporting their transformation into meaningful information and knowledge. Such work is increasingly identified as translational science, spanning the applications of informatics from the molecular and genomic levels to clinical levels and population health [3]. The panel accordingly has the goal to describe the key role of decision-support systems in CBI applications, looking at this problem from different angles, focusing on data analysis and integration, information sharing and interoperability, clinical decisions, cognitive science, and knowledge structures.

1. Clinical Bioinformatics – decision support through knowledge and data integration (Riccardo Bellazzi)

The first presentation in the panel will deal with the main ingredients of CBI, namely clinical data, molecular data, -omics databases, and the biomedical literature, thus introducing the current challenge of properly integrating such a vast amount of information into a decision-support system. Within this context, the role of different methodological and technological tools will be discussed, including, in particular, information retrieval, literature-based discovery, case-based reasoning and workflow management.

2. Bioclinical datawarehouses and translational research  (Patrice Degoulet)

The second presentation in the panel will deal with the design, development and practical use of bioclinical datawarehouses integrating heterogeneous patient-related data when considering their source (e.g., different clinical information systems) or their nature (e.g., clinical or omics data).

Application domains include genetic epidemiology, selection of patients for translational research but also the evaluation of decision-support systems before their integration into the production systems.

3. The Data Representation Challenge: Encapsulate and Bubble-up (Amnon Shabo)

Effective utilization of raw omics data in clinical environments depends on alignment of data representations. There is a continuum starting from raw & mass genomic data, e.g., whole-exome sequencing, and ending in summarized data, e.g., lab orgeneticist reports. In-between the two edges we need room for 'key data' as an intermediate representation with two layers: (1) certain data sets (e.g., biomarkers) are extracted as a result of analyses identifying the most significant data sets to the patient at the point of care; and (2) these key data sets are further analyzed in order to extract ('bubble-up') those data items that can be directly associated with phenotypic data and
thus make up clinical-omics statements (COS), e.g., this somatic mutation in the tumor tissue led to resistance to that drug. Such COS can be better 'understood' by decision support applications because semantics is explicitly represented, as opposed to common data representations where each type of data is represented separately and the genotype-phenotype associations are merely implied. To that end, it is important to use clinical-omics data standards that are capable of encapsulating raw omics key data and representing bubbled-up data in COS that eventually can be incorporated into the patient EHR. Encapsulated key omics data sets should be represented in common bioinformatics formats and they need to (1) point to the mass genomic data from which they have been extracted and (2) be referenced by bubbled-up data items that in turn are associated with phenotypic data. Such design has been used in the development of the HL7 Clinical Genomics specifications (e.g., as in the EC FP7 Hypergenes project).


The genomics revolution has introduced multiple new kinds of data that may be measured on individual patients and thereby need to be incorporated into clinical decision-support systems. Although the challenges encountered in offering decision support are similar with the use of both clinical and genomic data, the complexity and novelty of genetic information offers new challenges to clinicians as they attempt to offer personalized care. Thus our conventional notion of decision support needs to be expanded to consider the special needs of clinicians when they consider complex data from multiple sources. Cognitive research is accordingly relevant since it considers knowledge structures and mental processes underlying such performance, thereby informing the design and assessment of decision-support systems. Existing EHRs will need to be adapted to deal with the integration of genomic data regarding individual patients [4]. Interpreting an individual’s genomic data in conjunction with his corresponding clinical data will require new kinds of cognitive work with multiple information sources, new methods for knowledge organization, and new ways of visualizing the information that is available [5]. The creation of effective decision-support functionalities will accordingly require cognitive studies of expert decision making and an analysis of the resulting decision models. Cognitive science is accordingly a key element in the development of decision-support tools in the era of clinical bioinformatics and personalized medicine.

References