

Chapter XI

Clinical and Biomolecular Ontologies for E-Health

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ABSTRACT

This chapter mainly focuses on biomedical knowledge representation and its use in biomedicine. It first illustrates the existent more relevant bioinformatics resources and why they need to be better integrated. Then it describes what the main problems that machines can encounter in processing the factual biomedical knowledge are, what terminologies, classifications and ontologies are, and why they could help in better organizing and exploiting the bioinformatics resources available online. The authors hope that a concise perspective of the field and a list of selected resources, commented with their scope and usability, may help interested people in quickly understanding the main principles of knowledge representation in biomedicine and its high relevance for modern biomedical research and e-health.

INTRODUCTION

In the current post-genomic era, molecular medicine is increasingly gaining relevance both in health care research and practice. Availability

of the complete sequence of the human genome and of new nanotechnology approaches in molecular biology allows quickly and simultaneously studying thousands of genes and their expression levels. Advancements in information technologies

and biomedical informatics are providing tools and techniques to manage the amount of data produced, and are making more easily accessible several different databanks of biomolecular information and many methods for their analysis. With the increasing biomolecular and biomedical informatics progresses, many healthcare sites are progressively more offering several genetics tests at relatively low costs. In the near future, biomolecular tests and screenings are expected to revolutionize the diagnosis of inherited diseases in a similar way as imaging tests from different techniques (e.g. computer tomography (CT), magnetic resonance (MR), positron emission tomography (PET), single photon emission computer tomography (SPECT), ultra sounds (US)) have transformed the diagnostic practice of several illnesses and the diagnostic services offered by healthcare providers.

Although genetics tests can now be easily and routinely performed thanks to the automatic or semiautomatic procedures developed, management and interpretation of the data they produce, in particular of the results of more advanced biomolecular exams, still present a number of issues. In fact, they generate a great amount of data that need to be efficiently stored and statistically analyzed in order to identify, among all genes or proteins studied in each test, those significantly altered in the tested conditions. Moreover, to correctly interpret such test results, the known structural, functional, and phenotypic information about the identified genes and protein products need to be further analyzed. Such information - which include presence of specific sequence characteristics and protein domains; cytogenetic localization; expression in different cellular tissues and organs; and involvement in particular biological processes, molecular functions, biochemical pathways, genetic diseases or phenotypes - are increasingly available within numerous distributed databanks, generally easily accessible also through Web interfaces. However, some issues hamper their effective and comprehensive use

for the simultaneous analysis of the several hundreds of relevant genes and proteins identified in each biomolecular test. Such difficulties include: spreading of the required information among many heterogeneous databanks, the way most databanks provide these information (i.e. within unstructured HTML pages, one page for each gene or protein entry with all the information in the databank about the entry), and still lack of usage of common terminologies and bio-ontologies to describe biomolecular structural and functional characteristics of genes and their protein products and their phenotypic manifestations.

THE NEED FOR ONTOLOGIES

Bioinformatics research is heavily based on distributed resources spread across the Web. These resources can either be data sources such as genomic or proteomic databanks, or algorithmic sources. This last are collectively known as *Web Services* and usually perform some complex or resource demanding algorithm on uploaded data. A very typical bioinformatics Web service is a *sequence similarity analyzer* that receives a nucleic or amino acidic sequence as input and tries to find all the similar sequences among those within the selected database/s. All of these Web services are invaluable resources for the researcher, but when one finds himself using more and more Web resources, he/she can find quite awkward to switch back and forth from one to another to make multiple step articulated analyses. Moreover, these resources often differ in the data format for input or output. Thus, one eventually finds himself to desire better automatized tools that can take care of converting from one data format to another or automatically picking up data from several databanks, using them with distinct online resources and filtering only the relevant results. To make it short, users would really enjoy more automatized tools. The main problem for their implementation is that the Web

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