Chapter 14
Using Ontologies in eHealth and Biomedicine

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ABSTRACT

The main goal of the chapter is to emphasize (by the enumeration of some important results, activities, systems, groups, institutions, theories) the benefits the ontologies bring to the eHealth and biomedicine domains and the effort already given in this respect. The second goal of the chapter is to describe (by means of a case study) the representation and integration of the semantic and modeling layers of a system based on ontologies for the prevention of occupational risks. This system is an application of ontologies in the occupational health domain. The case study enumerates basic concepts and relationships in the proposed reference ontology and model, as well as the basic reasoning for them.

INTRODUCTION

Ontologies play an important role in medical informatics, contributing to the interoperability between systems (possibly distributed on web), to the access to heterogeneous information sources, to natural language processing, to the reuse of voluminous and complex information involved in health care activities. The ontologies provide a common language for a domain, a vocabulary used for data description and analysis.

After initially representing a technology proposed by researchers for solving problems of heterogeneous data sources, the ontologies have become a conceptual tool used by specialists in biomedicine for the consistent annotation of genotypic

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and phenotypic data. Thus, the ontologies' role in bioinformatics has changed from a collateral activity to a main one. In medicine, artifacts called ontologies are used to build controlled domain lexicons.

Unlike physics and chemistry, in biology, it is difficult to translate laws and models into mathematical formula. In general, biologists use knowledge about certain entities for obtaining new knowledge (e.g. facts about unknown entities). For example, when they are looking for similarities between sequences, they use algorithms that interpret ontology-based annotations.

eHealth and biomedicine are both scientific domains (biomedical research) and industrial ones (clinical practice, pharmaceutics). The research community has already recognized the need for ontologies in biomedical knowledge and data management.

The objectives of this chapter are:

- to present some introductory concepts about ontologies and terminological systems in eHealth and biomedicine;
- to present some major ontologies, repositories, and terminologies in eHealth and biomedicine;
- to enumerate some basic formalisms and instruments used in bio-ontologies;
- to present a case study on the semantic-based modeling and eTraining for the prevention of occupational risks.

BACKGROUND

This section provides basic definitions and a brief overview on *ontologies* that semantically support eHealth and biomedical applications. Specific terms of conceptualization, sharing, formal specification, and explicit specification are briefly explained.

ONTOLOGY

Ontology in philosophy is a field that studies what exists in the world or human being. In computer science (especially in artificial intelligence), in information science, in bioinformatics and biomedical informatics, it is a sharable, reusable, machine-readable data structure, emphasizing the practical usage (Gruber, 1993, p. 908).

Gruninger and Lee (2002) provide a well-known definition of the ontology: "a formal, explicit specification of a shared conceptualization for a domain of interest".

The interpretation of these terms is as follows:

- *conceptualization* refers to a model (usually a classification) composed of concepts and the specialization-generalization relationships between them, constraints upon concepts and relationships and axioms for the interpretation of and reasoning on concepts and relationships;
- *shared* implies that the community in a certain domain can reach a consensus on the conceptualization of the domain;
- *formal specification* means that the specification must be machine-readable and machine-understandable;
- *explicit specification* indicates that the concepts (meanings) and the relationships between them are explicitly defined (Yoo, 2008, p. 110-111), i.e. they are not encoded.

Artificial intelligence and Web researchers have co-opted the term for their own jargon and they consider the ontology a document or file that formally defines the relations among terms. The most typical kind of ontology for the Web relies on a *taxonomy* and a set of inference rules. (Berners-Lee, Hendler & Lassila, 2001, p. 2).

Ontologies provide essential knowledge to drive data integration, information retrieval, natural language processing, and decision support. Many application areas currently take advantage