

Chapter 15

Guideline Representation Ontologies for Evidence- based Medicine Practice

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ABSTRACT

An ontology in the context of guideline representation is a specification of conceptualizations that constitutes evidence-based clinical practice guidelines. It represents the elements of a guideline by specifying its attributes and defining the relationships that hold among them. For example, a guideline representation ontology would define a set of medical decisions and actions (concepts), as well as a set of rules (relationships) that relate the evaluation of a decision criterion to further reasoning steps or to its associated actions. A rigorously defined computational ontology provides considerable promise of producing computable representations that can be visualized, edited, executed, and shared using computer-based systems. A widely acknowledged ontology, or standard representation schema, is the key to facilitating the dissemination of guidelines across computer systems and healthcare institutions. The first part of this chapter presents the evolution of ontology research in guideline representation. Several representative ontologies are reviewed and discussed, with in-depth analyses of two popular models: GLIF (Guideline Interchange Format) and PROforma. The second part of the chapter analyzes seven key elements constituting a guideline representation. It also discusses the criteria for evaluating competing ontologies and some known limitations in the existing models. At the end of this chapter, four key steps are outlined that converts a guideline into computerized representation, which can be then used in Clinical Decision Support Systems (CDSSs).

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INTRODUCTION

Evidence-based medicine is “the conscientious, explicit, and judicious use of current best evidence in making medical decisions about the care of individual patients” (Sackett, Rosenberg, Gray, Haynes, & Richardson, 1996). Evidence-based clinical practice guidelines, a condensed form of evidence-based medicine, are published and maintained by professional organizations. For example, the National Guideline Clearinghouse¹ currently hosts over 1,700 active guidelines, spanning a wide variety of disease areas and conditions. However, these guidelines are usually prepared and disseminated as unstructured, descriptive textual documents that are primarily intended for human readers, inhibiting their automated use within Clinical Decision Support Systems. In addition, the lack of a standard schema also impairs the interoperability of guideline representations, resulting in a waste of implementation time and resources, and potentially increased error rates. To alleviate the problem, the development of standard, structured guideline representation ontologies is of vital importance.

The contemporary usage of the term *ontology* is derived from its much older usage in philosophy, in which it studies existence, its component entities, and the relationships between them. A contemporary ontology can be defined as “a formal, specific conceptualization of a domain; by adopting an ontology, an agent makes an ontological commitment to use only the vocabulary the ontology provides, and to use it only to denote the concepts provided” (Gruber, 1993). This definition articulates the concepts existing in a domain as well as its taxonomy, rules, and the relationships existing between them. These concepts are usually abstract, simplified views of the knowledge existing in a domain, such as medicine or biology. They do not necessarily represent complete knowledge of that domain, but can be composed of a subset, required for a specific application².

The main purpose of introducing the concept of ontology into knowledge representation is to enable knowledge sharing and reuse. Rigorously defined ontologies---with computer-recognizable semantics and structure that support some level of computation---are called *computational ontologies*. Computational ontologies are pivotal component of computer systems designed for intelligent reasoning, for example, Clinical Decision Support Systems.

The concept of ontology has been widely adopted in many disciplines such as computer science, information science, medicine, and genomics. Well known examples are 1) the semantic web ontologies that provide a universally accessible platform for data to be shared and processed by automated tools over web³; and 2) the gene ontologies that provide controlled vocabularies to describe gene and gene product attributes in any organism⁴. Not surprisingly, every knowledge-base or knowledge-based expert system should implement an ontology, explicitly or implicitly (Gruber, 1993).

In the context of clinical practice guideline representation, an ontology is a specification of concepts and relationships that constitute an evidence-based clinical practice guideline. It conceptualizes the elements of a guideline, their properties, and defines the relationships that hold among them. For example all guideline representation ontologies have a set of medical decisions and relevant actions (*concepts*), and a set of temporal rules that relate decision evaluation results to associated actions (*relationships*). A well established and generally acknowledged guideline representation ontology ensures that the resulting representations can be easily understood by non-authoring human readers, therefore facilitates the dissemination of guidelines across institutions. Well defined computational ontologies also provide considerable promise of enabling automated guideline acquisition, visualization, execution, and sharing. Such characteristics are prerequisites for a computer-recognizable, interchangeable guide-

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