Creating and Using Ontologies: What Informatics is All About

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In search of common denominators

The field of medical informatics continues to have an identity crisis. Our most articulate spokespeople seem to be unsure whether our discipline is called “medical informatics,” or “biomedical informatics,” or “health informatics,” or some other term. When asked what is the subject matter of our field, we typically respond first by saying, “it’s interdisciplinary” or “it brings together computers and health care.” We tend to define what we do in terms of what other academic communities do, without highlighting our unique intellectual contribution.

For medical informatics to flourish as an academic discipline, we must clarify the central themes of our research and demonstrate how those themes resonate throughout the work that we do. Nearly two decades ago, Blois [1] argued that the essence of medical informatics lies in understanding the unique structure of medical knowledge—in particular, the hierarchical relationships among concepts that cleave medical knowledge into multiple planes of abstraction. Blois clearly was on to something, but that it was not until the explosion of interest in formal ontologies in the 1990s [2] that workers in medical informatics could view Blois’ work in its proper perspective. We now recognize ontologies as explicit descriptions of the concepts (and relationships among concepts) that characterize an application area. Although Blois’ depiction of the structure of medical knowledge was purely philosophical, we now recognize the value of representing ontologies as computable representations that make explicit to our software systems (and to their developers and to their users) the concepts in an application area and how those concepts can be used computationally.

In the world of e-commerce and the Semantic Web [3], the development and use of ontologies are beginning to emerge as the key elements of the technology that drives the “new economy.” What is ironic, however, is that formal ontologies are at the core of work in medical informatics—and have had that role for more than one hundred years.

The “O” Word:
It’s not just for e-commerce anymore

The term ontology seems like just another arcane buzzword. Yet the notion of ontology is central to the controlled medical terminologies that have formed the centerpiece of work in medical informatics since the nineteenth century. Ontologies form the basis by which clinicians communicate with electronic patient record systems and enter case descriptions into decision-support systems. Ontologies make it possible to build large, maintainable knowledge bases that can codify what we know about specific areas of clinical practice in precise, unambiguous terms [4]. Ontologies define how nurses record observations regarding their patients. Ontologies define the terms with which health-care consumers interact with online information resources.

The development, use, and adaptation of ontologies are foundational to all work in informatics. In this talk, I will trace the use of ontologies throughout a variety of research programs in clinical informatics, stressing the common themes. The goal will be to show that understanding the role of ontologies in medical informatics (and in health informatics, and in biomedical informatics) makes it possible to concentrate more clearly on our fundamental scientific contributions. The emphasis on ontologies allows us both to elucidate the ultimate questions of informatics and to chart a unified research agenda that can transcend particular clinical application areas.

References