Biomedical ontology in action

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Biomedical ontology

Biomedical ontology is now an important element of biomedical research [1], healthcare [2] and public health [3], especially when it comes to integrating datasets in support of translational research [4]. Broadly defined, biomedical ontologies include the thesauri, terminologies, classifications and other coding systems used in biomedicine, in addition to those resources concerned with the identification of categories and their interrelations in various subdomains of biomedicine. As an academic discipline, biomedical ontology has investigated the organization of biomedical ontologies (e.g., [5]), has defined and contrasted the relations used in these ontologies [6], and, more generally, has studied the characteristics of biomedical ontologies (e.g., [7]). Some groups have developed principles and languages for representing and creating ontologies (e.g., [8]), as well as algorithms and tools for editing, aligning and visualizing ontologies (e.g., [9]), and assessing their consistency (e.g., [10, 11]). A significant proportion of the literature about biomedical ontologies emphasizes their design and structural characteristics, mentioning their use only in passing [11-17].

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In contrast, fewer reviews provide a functional perspective on biomedical ontologies [18, 19]. In general, a limited number of articles have presented the role played by several biomedical ontologies in specific applications, such as clinical decision support [20] and discovery applications [21], or in a specific domain, such as bioinformatics [22]. The papers of this special issue grew out of contributions to the *Second International Workshop on Formal Biomedical Knowledge Representation (KR-MED 2006)*, held in Baltimore, Maryland on November 8, 2006 [23]. The focus of this workshop was “Biomedical ontology in action”, emphasizing how current research can be brought to bear on the practical problems associated with the development of applications supported by biomedical ontologies. Out of the fourteen papers published in the online proceedings of this workshop [24], five were selected by the program committee due to their scientific excellence. We want to thank the reviewers for their iterative work to review the submissions to the workshop and, finally, the selected contributions to this special issue.

Contributions to this special issue

This special issue presents three applications of biomedical ontology to the extraction, interpretation and visualization of statements extracted from the biomedical literature. The remaining papers discuss the role of biomedical ontologies in electronic health records, and formal anatomical reasoning, respectively.

The article by Ai Kawazoe titled *The development of a schema for semantic annotation: gain brought by a formal ontological method* [25] illustrates the benefit of using formal ontological analysis (with OntoClean) in the development of a schema for semantic annotation used in *BioCaster*, a text minig system for the extraction of disease outbreaks. The authors compare the performance of *BioCaster* against a gold standard established manually. They demonstrate improved performance using an annotation schema informed by OntoClean requirements compared to the original schema.

In their article *On the ambiguity of ordinary statements in biomedical literature* [26], Stefan Schulz and Ludger Jansen analyze the possible meanings of what looks like a simple, typical statement from the biomedical literature about the interaction between two biochemical entities. They demonstrate that different interpretations are possible depending on whether such statements are understood as referring to interaction events or to the disposition of biochemical entities to interact with one another. They also show that different interpretations are possible for individuals vs. collectives (e.g., of molecules).

In an article titled *Using ontology visualization to facilitate access to knowledge about human disease genes* [27], Mary Dolan and Judith Blake explore solutions for visualizing human genes and their relations to molecular functions, biological processes, anatomical locations and diseases. Ontologies such as the Gene Ontology, used for the annotation of gene products in several model organisms, are exploited to inform the visualization and guide the exploration of integrated knowledge. The authors suggest that graphical visualization based on ontologies facilitates the understanding of complex knowledge about human disease genes.

Alan Rector, Rahil Qamar and Thomas Marley in their article *Binding ontologies and coding systems to electronic health records and messages* [28] propose a model for formally associating sets of codes from a biomedical terminology (e.g., SNOMED CT) to elements of an information model for electronic health records (e.g., HL7) through an ontology. The authors propose an implementation based on OWL DL for the code binding interface, in which the model of meaning (i.e., the ontology), the model of codes (i.e., the terminology) and the information model are distinct components.

In addition, the work on the logical properties of foundational mereogeometrical relations in bio-ontologies presented by Thomas Bittner at the KR-MED workshop will appear in an article in an upcoming issue of *Applied Ontology*.

This collection of articles illustrates how biomedical ontology effectively benefits a variety of biomedical applications, including text mining, knowledge visualization, electronic health records and anatomical reasoning. In other words, it shows biomedical ontology *in action*.

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