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Mini Review

Ontologies as Knowledge Representation Strategy in Biomedicine

Fernanda Farinelli[1] and Mauricio B Almeida[2]*

[1]*Faculty of Languages Literature, Federal University of Minas Gerais, Brazil*

[2]*Information Science, Federal University of Minas Gerais, Brazil.*

***Corresponding author:** Mauricio B Almeida, Federal University of Minas Gerais, Av. Belo Horizonte, Minas Gerais, Brazil.

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Abstract

Ontologies are artifacts for knowledge representations vital to address the vast amounts of data produced within the Biomedicine field. Biomedical ontologies organize and represent experts' knowledge, contributing to standardization, integrating vocabularies, and improving computational processing.

Introduction

Ontologies are used in various applications and have become an essential resource in biomedical research. Examples are applications that employed them as computable knowledge sources, like natural language processing and decision support systems. Ontologies are also fundamental for knowledge discovery within biomedical research [1]. Biomedical ontologies are used to organize and represent knowledge in the biomedical field, contribute to the standardization and integration of vocabularies. They also allow one to analyze, curate, and interpret the massive volume of data and information produced in experiments and professional practice [1].

Information Silos

As new ontologies in the biomedical domain emerge, the lack of st andardization becomes apparent, and therefore problems of ambiguities and inconsistencies between the terms [1-2]. The proliferation of biomedical ontologies can be seen in ontology repositories such as Bioportal, which catalogs about 950 ontologies, and Ontobee, with about 240 ontologies based on Basic Formal Ontology (BFO) [2]. BFO is a transcategorial top-level ontology described in the international standard ISO/IEC: 21838-2. Therefore, searching for precise terms and terminological definitions in the space of biomedical ontologies is unsettled: one can find overlapping, closely related, and even equivalent terms in one or several ontologies. Search engines sometimes suggest an extensive list of results for a given entry term, demanding the challenging task of selecting the most appropriate ontological resource. Biomedical ontology development initiatives focused on their specific needs, resulting in new "information silos". The phenomenon of information silos occurs because ontologies are developed in different projects by people with varying views on specialized knowledge, causing incompatibilities regarding definitions. Consequently, the low interoperability between the ontologies makes it challenging to share the data linked to them [2-3]. Furthermore, building a new domain ontology takes time, and the lack of reusing existing ontologies causes redundancy in the definitions and terms [4].

Harmonization search

The lack of standardization and alignment between ontologies represents an obstacle to the integration and use of these ontologies. Ontology's experts agree that domain ontologies developed under the same top-level ontology favor ontology interoperability once the top-level ontology specifies the semantics for very general terms common to almost all domains [3,5]. Another practice in favor of interoperability is ontology reuse. Reuse minimizes redundancies and overlapping definitions. However, for ontologies to be reused, they must be developed following standards recognized by the community [6]. Several initiatives emerged to promote ontology building standardization, mainly in biomedical ontologies. The Good OD guideline (Good Practice Ontology Design Principles) [7] guides ontology building. It presents some principles about the inclusion of a class in an ontology in such a way that ontologist should avoid creating classes that:

a. Disagree with reality: multiple classes referring to the same type of entity in reality; classes that do not have entity in reality; a single class that refers to unrelated types.

b. Closed predicates: when a class refer to a predicate corresponding to a finite number of individuals by some inherent limitation.

c. Impure predicates: when a class refer to one or more individual entity of reality.

Additionally, within biomedical ontologies, one can find the Open Biological and Biomedical Ontologies (OBO) Foundry. The goal of OBO is to create and maintain a set of interoperable, well-formed ontologies that represent biomedical scientific knowledge, becoming a reference in the knowledge representation. The initiative of OBO began in the 2000s, producing a set of principles considered good practices in the construction of ontologies. However, initially, these principles were not validated precisely. The interpretation of them was subjective and not always correctly followed. Recently, an automated validation mechanism was established to assess the conformity of any ontology with each principle, bringing an action for governance and curation of the developed ontologies [8].

Conclusion

The function of ontologies in the biomedical field is to standardize data storage to make them interoperable between information systems. This standardization is essential to facilitate maintenance, sharing, and interoperability, in addition to the heterogeneous integration of data from different data sources [1]. However, quality issues and lack of a standard for biomedical ontologies have hampered their applicability and subsequent adoption in real-world applications. Researchers of the ontology community also recognize the need for ontologies to follow the FAIR principles (Findable, Accessible, Interoperable, and Reusable) [9]. These principles guide those who want to improve data reuse. There are already ongoing initiatives discussing how to apply the FAIR principles to ontologies. Note that the first initiative towards standardization of biomedical ontologies, OBO Foundry, date from before the establishment of FAIR principles. Due to the recent actions of the OBO Foundry community to curate OBO Foundry ontologies, we can refer to these ontologies as a good choice to reuse. Finally, efforts in biomedical ontology development have to address the need for harmonization and integration of ontologies in order to maximize their utility.

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