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Logical properties of foundational relations in bio-ontologies

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Summary

Objective: We compare the advantages of specifying the semantics of foundational relations in bio-medical terminology systems using different types of formal deductive systems: first-order logic (FOL) and description logics (DLs).
Method: As our focus example, we use a terminology whose basic terms are supposed to designate proper parthood relations, subdivision relations, and surrounded-by relations. Each type of relation captures an important and distinct aspect of the spatial organization of anatomical structures: the general part-whole structure (proper parthood), the division of salient anatomical objects into discrete, tree-like structures (subdivision-of), and the nesting of anatomical objects into containers (surrounded-by). We show that all three types of relations are strict partial orderings (i.e., asymmetric and transitive). Ontologies whose purpose is to specify the semantics of terms referring to these types of relations must include axioms strong enough to formally distinguish among them. We compare the extent to which axioms characterizing proper parthood, subdivision, and surrounded-by relations can be represented in first-order logic and various description logics.
Conclusions: The development of bio-medical ontologies requires a rigorous formal analysis of foundational relations. Different kinds of formal tools may be used in this process. Ideally, an analysis in a highly expressive language, such as first-order logic, should be worked out in conjunction with analyses in less expressive but computationally tractable deductive systems such as description logics.

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1. Introduction

The growth of bioinformatics has led to an increasing number of evolving ontologies which must be correlated with the existing terminology systems developed for clinical medicine and bio-medical research. A critical requirement for such correlations is the alignment of specific relations in the ontologies with specific terms in the terminology systems [1,2]. If successful, such an alignment would have many advantageous consequences for biomedical research. Most importantly, this sort of alignment is necessary for the automatic processing of biomedical data structured by different terminology systems. At least one major obstacle to such integration is that many existing bio-medical terminology systems and ontologies handle foundational relations such as parthood ambiguously and inconsistently [3].

Necessary first steps in overcoming this problem are: (i) to identify the logical properties characterizing specific foundational relations and (ii) to develop a combined representation of different types of foundational relations in a single deductive system that is expressive enough to make critical distinctions in logical properties explicit.

In this paper, we rigorously distinguish three types of foundational relations by analyzing their logical properties: proper parthood relations, subdivision relations, and surrounded-by relations. Each type of relation captures an important and distinct aspect of anatomical structures. Intuitively, proper parthood relations determine the general part-whole structure of an anatomical object. Subdivisions relations organize salient parts of anatomical objects into discrete, tree-like structures. Surrounded-by relations link anatomical objects to other anatomical objects in whose cavities they are contained. For example, my heart is surrounded by my thorax and a bolus of food may be surrounded by my stomach.

We begin this paper by distinguishing important logical properties of binary relations and determining which of these properties characterize proper parthood relations, subdivision relations, and surrounded-by relations. We then use first-order predicate logic to develop an axiomatic theory that is powerful enough to make explicit the critical distinctions between the three kinds of relations and which supports valid (but not necessarily efficient) reasoning about the interactions among these types of relations. Finally, we show how weaker characterizations of these relations can be developed in a less expressive but computationally tractable description logic [4] that enables automated reasoning.

Logical properties of parthood relations have been the subject of extensive study in philosophy [5,6], linguistics [7], knowledge representation [8—12], and more recently in bio-informatics [13—16]. For applications in bio-informatics, it has been useful to introduce subtypes of the parthood relation such as functional parthood or componenthood [17,7,9].[1]

However, so far there has been little investigation of formal theories that combine different types of parthood relations or combine parthood relations with other types of spatial relations (for example, containment relations or surrounds relations). Recent work stressing the importance of such theories for anatomical and bio-medical ontologies includes [19,20].

Notice that in this paper, we focus on foundational relations between individuals (Joe Doe's head is part of Joe's body). Systems like the FMA or GALEN use instead relations between classes. However, the relations between classes are understood in terms of relations between the individuals in those classes as is shown in refs. [20,21]. Consequently, a clear analysis of relations between individuals is a necessary first step in an analysis of the corresponding relations between classes. To see how the work presented here extends to class-based relations see refs. [20,22].

2. Binary relations

In this section, we introduce binary relations as sets of ordered pairs of members of a specific underlying domain. We define important properties of binary relations and use these properties to distinguish different types of relations.

2.1. Binary relations as sets of ordered pairs

A *binary relation R with domain of discourse $\mathcal{D}(R)$* is a set of ordered pairs of members of $\mathcal{D}(R)$, i.e., $R \subseteq \mathcal{D}(R) \times \mathcal{D}(R)$. If R is a binary relation with domain $\mathcal{D}(R)$ then we will also say that R is a binary relation on $\mathcal{D}(R)$. We write $R(x, y)$ to say that R holds between $x, y \in \mathcal{D}(R)$, i.e., $R(x, y)$ if and only if $(x, y) \in R$.

For any binary relation R, we can define the following additional binary relations on $\mathcal{D}(R)$ in terms of R:

[1] This insight underlies the modeling of the parthood relation in GALEN [15] and in the Foundational Model of Anatomy (FMA) [18,1]. GALEN distinguishes, among others, the following subtypes of the general parthood relation: structural parthood, segment-of, layer-of, component-of, portion-of [15]. The FMA distinguishes, among others, constitutional parthood, regional parthood, and systemic parthood [16].