Partonomic Reasoning as Taxonomic Reasoning in Medicine

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Abstract

Taxonomic anatomical knowledge, a major portion of medical ontologies, is fundamentally characterized by is-a and part-whole relations between concepts. While taxonomic reasoning in generalization hierarchies is well-understood, no fully conclusive mechanism as yet exists for partonomic reasoning. We here propose a new representation construct for part-whole relations, based on the formal framework of description logics, that allows us to fully reduce partonomic reasoning to classification-based taxonomic reasoning.

Introduction

In the fields of health sciences and health care, broad-coverage terminologies have evolved over the years. A prime terminology source is the Unified Medical Language System (UMLS) metathesaurus (NLM 1998). It combines 53 heterogeneous conceptual systems, composed of a hierarchy totaling 476,313 concepts (updated on a yearly basis). From a knowledge representation perspective, UMLS can be viewed as a huge semantic network. Unfortunately, it shares all the drawbacks pointed out in the seminal paper by Brachman (1979). Hence, given its size, evolutionary diversity and long-lasting maintenance history, the apparent lack of a formal semantic foundation leads to inconsistencies, circular definitions, etc. (Cimino 1998).

Taxonomic anatomical knowledge, a major portion of these ontologies, is fundamentally characterized by is-a and part-whole relationships between concepts. As a matter of fact then a frequent mixture of generalization (IS-A) and partitive (PART-OF) relations occur at the same hierarchical level. For instance, "blood" subsumes "blood plasma" (partitive), as well as "fetal blood" (generalization).

The Common Reference Model for medical terminology, developed within the GALEN and GALEN-IN-USE projects (Rector *et al.* 1995) marks, for the time being, one of the few attempts to construct a large-scale medical ontology in a formally founded way. In this context, GRAIL, a KL-ONE-like knowledge representation language, has been developed and, by design, specifically adapted to the requirements of the medical domain (Rector *et al.* 1997). GRAIL, unlike most description logics, has a built-in mechanism that explicitly targets at part-whole reasoning, an extension that reflects the outstanding importance of this reasoning pattern in the medical domain.

In our research, the necessity to account for medical knowledge in a principled way arose from the need to make deductive reasoning capabilities available to MEDSYNDIKATE, a text understanding system that processes pathology reports (Hahn, Schulz, & Romacker 1999). To supply MEDSYNDIKATE with the enormous amount of medical knowledge already specified in the UMLS metathesaurus, we transfer UMLS specifications to the more rigorous framework of description logics.

Hence, generalization hierarchies (via IS-A and INSTANCE-OF relations), as well as PART-OF relations have to be accounted for in a systematic way. In the course of many ontology engineering cycles we recognized some problems that challenged conventional wisdom in medical knowledge representation. In particular, we encountered many exceptions to the rule of transitivity of PART-OF and the way it effects specialization of associated concepts.

We here abandon the notion of "flat" concept nodes and rather replace them by a tripartite concept encoding that fully incorporates part-whole knowledge. Since we embed our approach into the framework of KL-ONE-style description logics (Woods & Schmolze 1992), we subsequently rely upon the standard terminological classifier for partonomic reasoning along PART-OF relations, basically, in the same way as for taxonomic reasoning along generalization (IS-A) hierarchies.

Part-Whole Reasoning Problems

Two aspects of reasoning on part-whole relations have received special attention — whether transitivity can be considered a general property and how partonomic reasoning relates to taxonomic reasoning, i.e., whether specialization relations can be inferred from part-whole relations in related parts of a knowledge base.

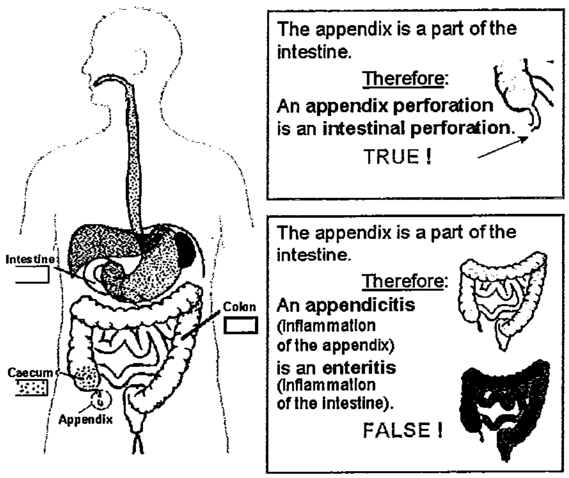

Figure 1: Digestive Tract and its Parts
Left: Position of the Appendix within the Digestive Tract. Right: Disease Concepts Related to *Appendix* and to *Intestine*, with and without Concept Specialization

Transitivity. The importance of transitivity of the PART-OF relation for adequate reasoning has largely been discussed in the literature (cf. the overview in Artale *et al.* (1996)). Winston, Chaffin, & Herrmann (1987) argue that part-whole relations can be considered transitive as long as "a single sense of part" is kept. This means that the general PART-OF relation is not transitive, whereas *each* distinct subrelation of PART-OF is transitive. As soon as more than one single-sense PART-OF subrelation is involved in a relation chain, transitivity no longer holds, in general. For instance, a FINGER is a PHYSICAL-PART-OF an ARM which is a PHYSICAL-PART-OF a MUSICIAN; a MUSICIAN is a MEMBER-OF an ORCHESTRA. Because FINGER and MUSICIAN are related by the same PART-OF subrelation (viz. PHYSICAL-PART-OF) we conclude that a FINGER is a PHYSICAL-PART-OF a MUSICIAN, whereas it is not a PART-OF an ORCHESTRA, since a second kind of a PART-OF (viz. MEMBER-OF) relation comes into play.

The transitivity property is widely acknowledged in the domain of medical anatomy, too. If an anatomical object is PART-OF another one, which itself is included in a larger structure, the first one is also a PART-OF the larger structure. For instance, the APPENDIX is a PART-OF the CAECUM, the CAECUM is a PART-OF the COLON, and the COLON is a PART-OF the INTESTINE. Hence, the APPENDIX is also a PART-OF the INTESTINE (cf. Fig. 1, left side). Since we have encountered many instances of subrelations of the anatomical PART-OF relation, for which the transitivity assumption is questionable or may even be rejected (cf. our discussion of the phenomena illustrated in Fig. 5), we consider it as a decision at the level of ontology engineering — for each and every PART-OF relation — whether transitivity must be granted or not. In particular, it turns out that this problem cannot be solved at the level of the axiomatic definition of knowledge representation languages and the operators they supply.

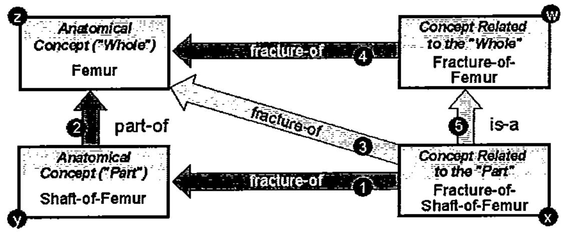

Figure 2: Taxonomic Reasoning in Partonomies

Taxonomic reasoning in partonomies. Rector *et al.* (1997) discuss two taxonomic reasoning patterns that crucially depend on part-whole relations. The first one accounts for **role propagation** in partonomies, i.e., the portion of a knowledge base that is linked via PART-OF relations. Consider, e.g., Fig. 2, where a concept x (FRACTURE-OF-SHAFT-OF-FEMUR) is related to a "part" concept y (SHAFT-OF-FEMUR) via some relation **R** (FRACTURE-OF ❶). The "part" concept y is an anatomical PART-OF (❷) a "whole" z (FEMUR). Given that a concept from the range of the relation FRACTURE-OF (y) is in the domain of a PART-OF relation whose range concept is z, the relation **R** (FRACTURE-OF) can also be propagated to z (❸). More generally, when two relations, **R** and **S**, are given, **S** being a subrelation of PART-OF, the following implication holds:

$$x\mathbf{R}y \wedge y\mathbf{S}z \Rightarrow x\mathbf{R}z \qquad (1)$$

Second, the above framework also allows for **concept specialization** in partonomies. As an example (cf. Fig. 2), we assume the relation FRACTURE-OF to link x (FRACTURE-OF-SHAFT-OF-FEMUR) and y (SHAFT-OF-FEMUR) (❶), as well as w (FRACTURE-OF-FEMUR) and z (FEMUR) (❹). Given the PART-OF relation between y and z (❷), we conclude that x (FRACTURE-OF-SHAFT-OF-FEMUR) specializes w (FRACTURE-OF-FEMUR) (❺), hence x IS-A w. The general reasoning pattern can be phrased as follows for two relations, **R** and **S**, **S** being a subrelation of PART-OF:

$$x\mathbf{R}y \wedge w\mathbf{R}z \wedge y\mathbf{S}z \Rightarrow x\mathbf{ISA}w \qquad (2)$$

Obviously, the reasoning pattern (2) is a special form of (1). Along the lines of these two schemes, dedicated knowledge representation languages, such as GRAIL (Rector *et al.* 1997), have been developed. In this framework, taxonomic reasoning in partonomies can be defined as a property of a conceptual relation by an axiom in the form **R** *specializedBy* **S**, iff $\mathbf{S} \sqsubseteq$ PART-OF. This implies that the relation **R** is *always* propagated along hierarchies based on **S**, i.e., the inheritance mechanism is invariably associated with the relation **S**, and that concept specialization is deduced on the basis of PART-OF relations (hence, *"part-whole" specialization*).

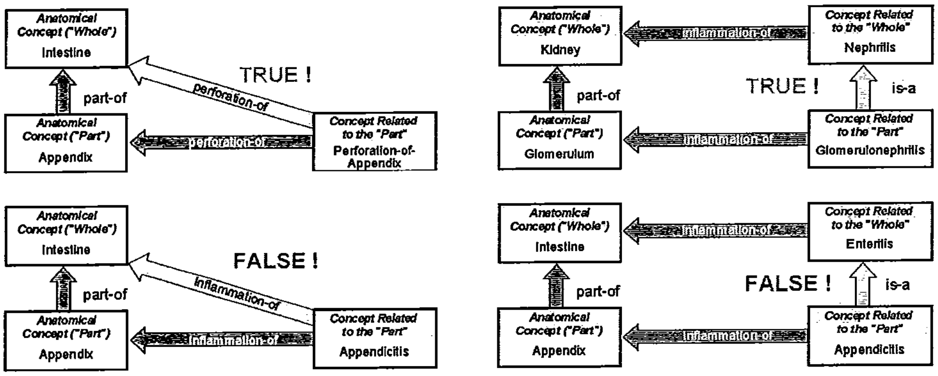

Figure 3: Regular and Irregular Reasoning Patterns (Upper vs. Lower Part) for Role Propagation and Concept Specialization (Left vs. Right Part)

This way, partonomic reasoning is dealt with at the axiomatic language definition level. We have, however, collected empirical evidence at the ontology engineering level that such an axiomatic approach might be fundamentally inadequate. We make the following claims:

1. Role propagation in partonomies does *not generally* hold. Consider Fig. 3 (left side), where a PERFORATION-OF the APPENDIX (PERFORATION-OF-APPENDIX) implies a PERFORATION-OF the INTESTINE, whereas an INFLAMMATION-OF the APPENDIX (APPENDICITIS) does not imply an INFLAMMATION-OF the INTESTINE, given that APPENDIX is an ANATOMICAL-PART-OF the INTESTINE.
2. Also concept specialization in partonomies does *not generally* hold for certain concepts related to a partonomy by the *same* relation. For instance, given that GLOMERULUM and KIDNEY are related by an ANATOMICAL-PART-OF relation just like APPENDIX and INTESTINE, we observe another clash of inference results (cf. Fig. 3 right side). For example, in contradistinction to the fact that a GLOMERULONEPHRITIS (an INFLAMMATION-OF the GLOMERULUM) specializes a NEPHRITIS (an INFLAMMATION-OF the KIDNEY), an APPENDICITIS (INFLAMMATION-OF the APPENDIX) does not specialize the concept ENTERITIS (INFLAMMATION-OF the INTESTINE).

   Both reasoning patterns interact. Concept specialization requires the role propagation pattern to be true. Vice versa, if the role propagation pattern is false, consequently also concept specialization cannot hold (cf. Fig. 3, left and right side, lower example).

Currently, neither established large-scale terminologies nor dedicated medical knowledge representation languages are able to properly account for the above-mentioned, regular as well as irregular, phenomena typical of part-whole hierarchies. The solution we propose rests on the assumption that the generality of the reasoning patterns (1) and (2) have to be restricted. Instead of giving them the status of generally valid axioms or devise (costly) language built-ins such as transitive closure or part-of operators, we reduce partonomic reasoning entirely into standard classification-based taxonomic reasoning. In order to circumvent many of the contradictions we have pointed out we introduce tripartite concept descriptions that already incorporate part-whole relations. This allows us to assign the decision as to whether transitivity or specialization actually hold down to the ontology engineering level where medical expertise becomes decisive.

Partonomic Reasoning Goes Taxonomic

We now turn to the special properties of partonomic reasoning by reducing it to taxonomic reasoning. The crucial point about the feasibility of this reduction lies in the provision of a tripartite concept encoding, so-called SEP triplets, to which we turn first. Following on that, we exploit the generalization hierarchy to enable useful inferences that are typical of transitive relations and show, moreover, how the same formalism allows conditioned taxonomic reasoning on partonomies.

SEP-Triplets. In our domain model, the relation ANATOMICAL-PART-OF describes the partitive relation between physical parts of an organism and is embedded in a specific triplet structure by which anatomical concepts are modeled (cf. Fig. 4). The restriction to a single tree of subrelations of PART-OF is sufficient for the logical deductions we encounter in the medical domain. A triplet consists, first of all, of a composite "structure" concept, the so-called **S-node** (e.g. INTESTINE-STRUCTURE). Each "structure" concept subsumes both an anatomical *entity* and each of the anatomical *parts* of this entity. Unlike entities and their parts, "structures"

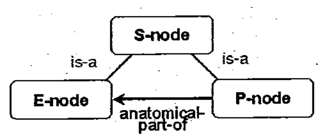

Figure 4: Structure of SEP-Triplets

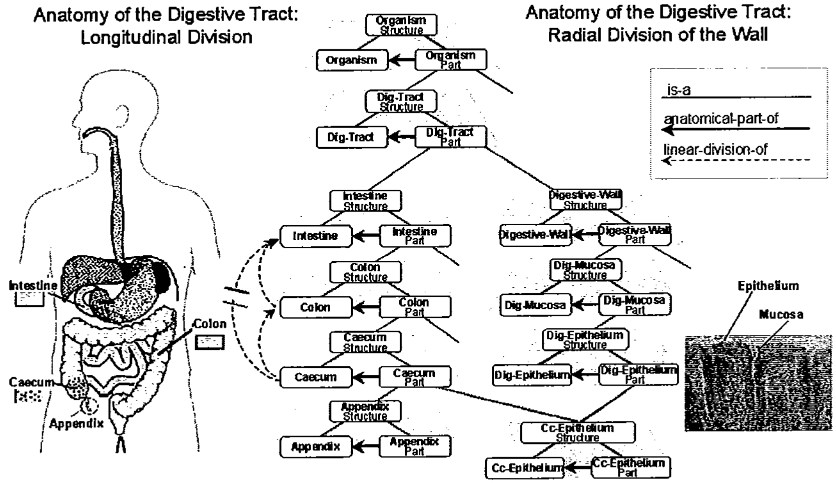

Figure 5: Segment of the Part-Whole Taxonomy of the Gastrointestinal Tract in SEP-Triplet Encoding

have no correlate in the real world — they constitute a representational artifact required for the formal reconstruction of the patterns of part-whole reasoning we have already discussed.

The two direct subsumees of an S-node are called **E-node** ("entity") and **P-node** ("part"), e.g., INTESTINE and INTESTINE-PART, respectively. Unlike an **S-node**, these nodes refer to specific ontological objects. The E-node denotes the *whole* anatomical entity to be modeled, whereas the P-node is the common subsumer of any of the parts of the E-node. Hence, for every P-node there exists a corresponding E-node for the role ANATOMICAL-PART-OF. Fig. 5 illustrates the model of a segment of the gastro-intestinal anatomy subdomain. Note that the formalism supports the definition of concepts as conjunctions of more than one P-node concept, as illustrated by the concept CC-EPITHELIUM.

Transitivity via Inheritance of SEP-Triplets. Let C and D be E-nodes (e.g., the organs CAECUM and APPENDIX), and $AStr$ be the top-level structure concept of a domain subgraph (e.g., ORGANISM-STRUCTURE). $CStr$ and $DStr$ (e.g., CAECUM-STRUCTURE and APPENDIX-STRUCTURE), are then the S-nodes that subsume C and D, respectively, just as $CPart$ and $DPart$, e.g., CAECUM-PART and APPENDIX-PART, are the P-nodes related to C and D, respectively, via the role ANATOMICAL-PART-OF. All these concepts are embedded in a generalization hierarchy such that

$$D \sqsubseteq DStr \sqsubseteq CPart \sqsubseteq CStr \sqsubseteq .. \sqsubseteq APart \sqsubseteq AStr \quad (3)$$

$$C \sqsubseteq CStr \sqsubseteq .. \sqsubseteq APart \sqsubseteq AStr \quad (4)$$

The P-node for $CPart$ is defined as follows:

$$CPart \doteq CStr \sqcap \exists \text{ANATOMICAL-PART-OF}.C \quad (5)$$

Since D is subsumed by $CPart$ (3), we infer that D is an ANATOMICAL-PART-OF the organ C :

$$D \sqsubseteq \exists \text{ANATOMICAL-PART-OF}.C \quad (6)$$

Clearly, this pattern of ***part-of inheritance*** holds at every level of the part-whole hierarchy. In our example (cf. Fig. 5), the subsumption relation expressed in (3) may be illustrated by identifying the concept D with APPENDIX that is a subconcept of APPENDIX-STRUCTURE, CAECUM-PART, CAECUM-STRUCTURE etc. up to ORGANISM-PART and ORGANISM-STRUCTURE. In the same way, C is identified with CAECUM which is a subconcept of CAECUM-STRUCTURE, etc. (4). Between CAECUM-PART and CAECUM, there exists an ANATOMICAL-PART-OF relation (5). We conclude that a relation ANATOMICAL-PART-OF also holds between APPENDIX and CAECUM (6), but also between APPENDIX and COLON, APPENDIX and INTESTINE, COLON and INTESTINE, etc.

Analyzing the ontological structure of the medical domain reveals an interesting observation. Various specializations of ANATOMICAL-PART-OF are not transitive, although transitivity seems to hold for the general ANATOMICAL-PART-OF relation. The PART-OF inheritance mechanism is able to cope with this exception phenomenon. This feature is illustrated by the dotted arrows in Fig. 5 (left side). PART-OF inheritance can be selectively obviated in case of certain subrelations of ANATOMICAL-PART-OF, such as LINEAR-DIVISION-OF. This is achieved by linking the LINEAR-DIVISION-OF relation to the entity nodes rather than to the structure nodes of the concepts involved. We then describe COLON as a LINEAR-DIVISION-OF INTESTINE, CAECUM as a LINEAR-DIVISION-OF COLON, but CAECUM cannot be described as a LINEAR-DIVISION-OF INTESTINE.

Thus, SEP-triplets provide a flexible and powerful ontology engineering methodology which embeds reasoning about partonomies simply into IS-A taxonomies. Their characteristic properties, viz. ***transitivity*** and ***antisymmetry***, by which acyclicity is guaranteed, apply directly to the way we model partonomic relations.

Concept specialization on partonomies is based on the transitivity of the PART-OF relation and the spezialization axiom (3). Provided our triplet structures consisting of E-nodes, P-nodes and S-nodes, we can flexibly enable or suppress concept specialization on partonomies, i.e., the inference of a subsumption relation between concepts that are related to partonomies. The decision whether the switch is set to "on" or "off" has to be made by the medical expert. Whenever, e.g., a disease concept is related to an anatomical concept, the knowledge engineer must explicitly determine whether it effects concept specialization or not (see the ENTERITIS/NEPHRITIS example from Fig. 3, right part). Just as with transitivity, concept specialization on partonomies is enabled when a disease concept is attached to an S-node, while it is disabled when the concept is linked to an E-node. Why this is the case can be shown by looking at the same taxonomy as described in the terminological statements (3) to (6). Let R and S be relations that link the disease concepts W, X, Y, Z to the anatomical hierarchy. From

$$W \doteq \exists S.CStr \tag{7}$$

$$X \doteq \exists S.DStr \tag{8}$$

$$DStr \sqsubseteq CStr \tag{9}$$

we conclude that

$$X \sqsubseteq W \tag{10}$$

While the "S-node pattern", (7) to (10), allows concept specialization in partonomies, the following "E-node pattern" does not:

$$Y \doteq \exists R.C \tag{11}$$

$$Z \doteq \exists R.D \tag{12}$$

The conclusion

$$Z \sqsubseteq Y \tag{13}$$

cannot be drawn, since the extension of D is not a subset of the extension of C.

In our example (cf. Fig. 6: top, right side), (7) and (8) can be interpreted as follows: INTESTINAL-PERFORATION is a PERFORATION-OF an INTESTINE-STRUCTURE and PERFORATION-OF-APPENDIX is a PERFORATION-OF an APPENDIX-STRUCTURE. Since APPENDIX-STRUCTURE is subsumed by INTESTINE-STRUCTURE (9), it follows by the **S-node** pattern that a PERFORATION-OF-APPENDIX specializes INTESTINAL-PERFORATION (10).

Considering an alternative encoding in Fig. 6 (top, left side), the concept ENTERITIS is not linked to the S-node INTESTINE-STRUCTURE by the role INFLAMMATION-OF, but to the **E-node** INTESTINE instead (11), just as APPENDICITIS is linked to the E-node APPENDIX (12). As INTESTINE does not subsume APPENDIX, according to the **E-node** pattern *no* specialization relation (13) between APPENDICITIS (= Z) and ENTERITIS (= Y) can be inferred.

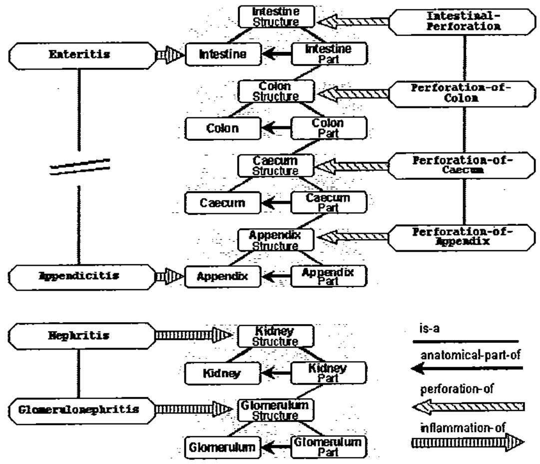

Figure 6: Conditioned Concept Specialization

It is therefore only the difference in the concept linkage patterns (linkage to S-nodes vs. linkage to E-nodes) that liberates or obviates concept specialization on partonomies. If $R = S$, the same relation is used for concept specialization in one case, though not in the other. Therefore, concept specialization on partonomies is not a property of the relation itself, but derives from the parametrization of SEP-triplets.

We may illustrate this case with the relation INFLAMMATION-OF, by comparing its use in two subgraphs. In the lower part of Fig. 6, the S-node pattern (expressions (7) to (10)) is applied to the KIDNEY subgraph in order to define the concepts NEPHRITIS and GLOMERULONEPHRITIS, whereas in the upper part the definition of ENTERITIS and APPENDICITIS obeys the E-node pattern in the INTESTINE subgraph. This example shows clearly how the same relation (INFLAMMATION-OF) supports concept specialization on partonomies in one case (KIDNEY), while in the other (INTESTINE) it does not. Thus, our methodology allows for conditioned enabling or disabling of concept specialization for concepts related to partonomies.

With these examples we may challenge the validity of the inference rule (2) in two ways. First, we have determined subrelations of S (e.g., LINEAR-DIVISION-OF), for which transitivity does not hold. Second, we may even claim that, depending on the choice of the domain/range concepts, a particular relation S allows transitivity, while in other cases it prohibits transitivity.

Role propagation on partonomies follows when specialization is given between the concepts related to a partonomy by the same relation. In Fig. 6 (right side), the deduction that a PERFORATION-OF an APPENDIX-STRUCTURE is also a PERFORATION-OF an INTESTINE-STRUCTURE clearly results from the fact that APPENDIX-STRUCTURE is subsumed by INTESTINE-STRUCTURE. The mapping of partonomies to generalization (IS-A) hierarchies provides the representational mechanisms for appropriate reasoning.

Related Work

For the medical domain, Haimowitz, Patil, & Szolovits (1988) first requested a representation formalism for *part-whole* relations and corresponding reasoning capabilities as an extension to terminological logics. As a response, three basic approaches can be distinguished.

In the first, part-whole reasoning is dealt with by extending a knowledge representation language by new operators dedicated to partonomic reasoning. Such a proposal, a transitive closure operator for roles, has been elaborated by Baader (1991), who also discusses the computational costs implied, viz. intractability of the resulting terminological system. In a similar vein, the GRAIL language constitutes an extension of terminological logics adapted to the part-whole reasoning patterns in the medical domain (Rector *et al.* 1997). However, role propagation and concept specialization are hard-wired to role definitions and, therefore, fail to match empirical data from anatomical ontologies.

In the second approach, reasoning patterns are adapted to particular (sub)relations (Cohen & Loiselle 1988). Since the concept nodes to which these relations are linked cannot be constrained, this approach fails when the same relation allows and prohibits, e.g., transitivity. The same counterargument hits proposals in which subrelations of PART-OF are declared to be transitive, in general (Hahn, Markert, & Strube 1996).

The third approach tries to preserve standard language definitions for reasons of simplicity and parsimony. Along this line, Schmolze & Marks (1991) proposed a solution similar to ours using subsumption to obtain inferences resembling those of transitive roles or transitive closure of roles. Artale *et al.* (1996) criticize this proposal for the "proliferation of (artificial) concepts" involved. We argue, on the contrary, that these additional concepts are necessary from an ontological point of view, as the distinct mechanisms for conditioned specialization modeling reveal (cf. Fig. 6).

It remains to be seen, however, whether conservative structural extensions of a stable language platform are able to carry over to the many varieties of partonomic reasoning and different part-whole relations (discussed in a survey by Sattler (1995)), or whether newly designed operators or other fundamental language extensions are needed. In the medical domain, at least, where the restriction to one subrelation of PART-OF, viz. ANATOMICAL-PART-OF, is sufficient, a relatively simple "data structure" extension like the SEP triplets yields already adequate results, without the necessity to resort to profound extensions of the terminological language.

Conclusion

In this paper, we have argued against two commonly shared opinions about partonomic reasoning. First, that part-whole relations are transitive and transitivity can be considered an inherent property of the relation itself; second, that subsumption relations invariably hold within partonomies.

Our alternative focuses on a tripartite encoding schema for concepts that incorporates part-whole specifications. Embedding the corresponding SEP-triplets into an inheritance hierarchy allows us to use standard terminological classifiers of description logics systems for partonomic reasoning in the same way they are used for taxonomic reasoning. The SEP-triplets provide the flexibility required for an ontology engineer to decide whether transitivity should hold or not.

This approach might generalize to other domains as well. Consider the following commonsense scenario. The car-body is clearly a part of the car. From the car-body's color we may infer the color of the car. So are the seats part of a car. The color of the car, however, would not be inferred from that of the seats.

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