An Ontological Engineering Methodology for Part-Whole Reasoning in Medicine

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Abstract

Part-whole relationships are fundamental ontological categories for medical reasoning. Part-whole modeling, however, still provides no conclusive methodology for adequate representations. We propose a new representation construct for part-whole reasoning based on the formal framework of description logics, thereby overcoming problems that arise in the context of previous formal approaches to part-whole modeling, as well as widely spread comprehensive medical terminologies.

Introduction

In medical informatics research, knowledge representation issues have been emphasized in recent years. It is becoming obvious that efficient classification, processing of structured data and free texts, as well as a broad variety of sophisticated information retrieval services (e.g., fact retrieval, text passage retrieval) and knowledge-based decision support require a common conceptual framework to facilitate semantic interoperability (Evans *et al.*, 1994; Friedman *et al.*, 1995). Concept systems routinely used in medicine and healthcare are essentially classifications which have a fixed set of alphanumeric codes for statistical analysis and accounting (e.g., ICD (WHO, 1992)), or thesauri for bibliographies, indexing and retrieval (e.g., MeSH (NLM, 1997)). While ICD has become a worldwide standard, many coding systems used in clinical routine have their scope restricted to national health systems or clinical specialities. Semantic interoperability between these systems is generally not achieved, nor even aimed at.

Even more sophisticated composite conceptual systems, such as the SNOMED nomenclature (Cote, 1993), lack a clear semantics as hierarchical links remain untyped. A mixture of generalization and partitive relations, often at the same hierarchical level, is typically found. For instance, "blood" subsumes "blood plasma" (partitive) as well as "fetal blood" (generalization). Conceptually invalid combinations (e.g., "fracture of the blood") are not rejected, and often the same concept can be classified by various code combinations not linked with each other.

An attempt to unify 53 conceptual systems (with a total of 476,313 concepts) is constituted by the UMLS (Unified Medical Language System) project (NLM, 1998; McCray & Razi, 1995). The designers of UMLS are fully aware of the problems encountered in the existing terminologies, and, although they make considerable effort to add semantics to concepts and links, UMLS is still far from being a logically sound ontology of medicine. The inconsistencies inherited from the sources, also concerning part-whole relationships, are crucial and create continuous problems for UMLS.

The Common Reference Model for medical terminology, developed within the GALEN and GALEN-IN-USE projects (Rector *et al.*, 1995; Rector & Horrocks, 1997) marks, until now, the only major attempt to construct a large-scale medical ontology in a strict, i.e., 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). Interestingly enough, GRAIL, unlike most description logics, has a built-in mechanism for part-whole reasoning.

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 natural language text understanding system that processes pathology reports (Hahn, Schnattinger, & Romacker, 1996; Hahn & Romacker, 1997). Since MEDSYNDIKATE had been ported from an information technology (IT) report understanding application, the architecture was to be kept as stable as possible. The IT version of MEDSYNDIKATE makes use of a standard KL-ONE-style terminological representation language (LOOM), which is strong with respect to reasoning in generalization hierarchies but provides no special support for part-whole reasoning.

Hence, we faced the challenge of accounting for this relevant portion of medical knowledge in a systematic way, i.e., using the terminological classifier for part-whole reasoning in the same way as for taxonomic reasoning along IS-A hierarchies. The solution we arrived at is general in the sense that given a simple encoding schema for meronymic knowledge, versatile part-whole reasoning is made possible.

Part-whole (also called meronymic) reasoning has two as-

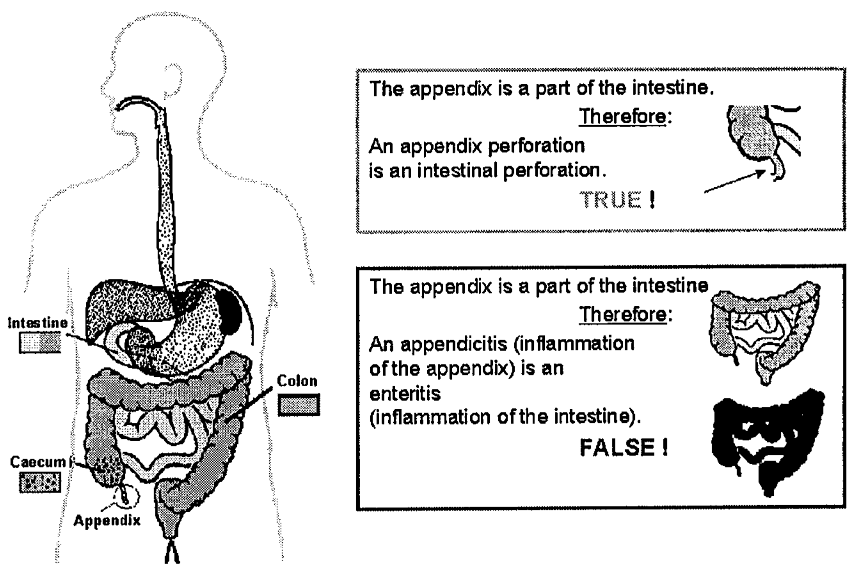


Figure 1: Digestive Tract and its Parts. Left: Position of the Appendix within a Part-Whole Hierarchy. Right: Disease Concepts related to *Appendix* and to *Intestine*, with and without Part-Whole Specialization along the Part-Whole Hierarchy

pects: *transitivity* and *specialization*.

Transitivity. The transitivity of part-whole 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[1] 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 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 relation is applied. Note, that the notion of *meronymic* relations refers to all specializations of the general PART-OF relation, such as MEMBER-OF, PHYSICAL-PART-OF etc. By *partitive* relations, however, only the specific subrelation PHYSICAL-PART-OF and all of its subrelations are refered to.

In the anatomy domain, part-whole relations are generally applied to 3-dimensional spaces or 2-dimensional surfaces. If an anatomical object is located within the physical boundaries of another one, which itself is included in a larger structure, the first is also a PART-OF this larger structure. For instance, the APPENDIX 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).[2] So, we assume that transitivity generally holds for the PART-OF relation, applied to anatomical objects. We are nevertheless aware that for certain subrelations of the anatomical PART-OF relation the transitivity assumption is questionable or may even be rejected.

Part-whole specialization Besides transitivity, *specialization* is the other important issue related to part-whole hierarchies. It is either known as "coordination of multiple taxonomies based on relations other than subsumption" (Horrocks, Rector, & Goble, 1996) or as "inheritance along part-whole taxonomies" (Artale *et al.*, 1996). For better understanding we will use the term *part-whole specialization* henceforth. Incorporating the definition of relations which are transitive along other domain-specific relations at the level of conceptual modeling constitutes a major desideratum for properly designed medical knowledge bases. As illustrated by Fig. 2, part-whole specialization means that one concept related to a "part" has to be subsumed by another concept related by the same conceptual relationship to the corresponding "whole". In a medical concept system, e.g., we want to infer that a concept such as FRACTURE-OF-THE-SHAFT-OF-THE-FEMUR is subsumed by a concept FRACTURE-OF-THE-FEMUR given that SHAFT-OF-THE-FEMUR is a PART-OF the FEMUR (Horrocks, Rector, & Goble, 1996).

Standard description logics as those implemented within the KL-ONE language family, do not support inheritance other than along generalization hierarchies. It is no wonder

[1]We use SMALLCAPS to denote concept and relation identifiers.

[2]As the following example refers to details of the human anatomy, we will introduce some basic concepts for ease of understanding: The *digestive tract* is a part of the human body and constitutes the following sequence of tubular organs: *oesophagus, stomach, intestine*. The *intestine* is divided into the *small intestine* and the *colon*. The *colon* is divided into the *caecum* (or *blindgut*), the *ascending colon* and some other segments. The *caecum* (localized in the right lower part of the abdomen), has a worm-shaped, dead-ending tubular part called *vermiform process* or *appendix*. An acute inflammation of this part is generally known as *appendicitis*. The radial division of the digestive tract exhibits an ordered sequence of discrete layers. The innermost layer is called the *digestive mucosa*.

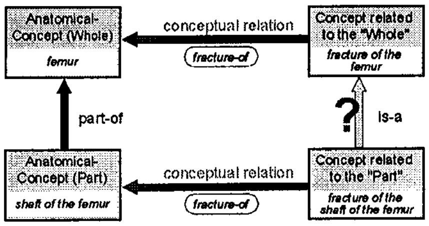


Figure 2: Inheritance along *Part-Of* Relations, also called Part-Whole Specialization

that due to the outstanding importance of part-of reasoning in medicine, special concept representation languages such as GRAIL (Rector *et al.*, 1997) have been developed. Here, part-whole specialization is modeled as a property of certain conceptual relations, e.g. HAS-LOCATION is specialized by PART-OF. This means that the HAS-LOCATION relation is *always* inherited along hierarchies based on PART-OF, independent of which concepts are involved.

As a result of our experience with the construction of a pathology knowledge base and based on shared medical expertise we may state that

1. Part-whole specialization does not generally hold: A PERFORATION-OF the APPENDIX can be classified as an INTESTINAL-PERFORATION, whereas APPENDICITIS (INFLAMMATION-OF the APPENDIX) is definitely not an ENTERITIS (INFLAMMATION-OF the INTESTINE), cf. Fig. 1, right side.
2. The *same* conceptual relation (here: INFLAMMATION-OF) may support part-whole specialization in one case though not in another one: Whereas we have stated that APPENDICITIS is not an ENTERITIS, the same relation INFLAMMATION-OF, applied to another organ, e.g. the KIDNEY, exhibits a different behavior: PYELONEPHRITIS, an INFLAMMATION-OF the PYELON (a part of the kidney) can be subsumed consistently by NEPHRITIS (INFLAMMATION-OF the KIDNEY).

We claim that in a medical ontology all phenomena typical of part-whole hierarchies should be adequately represented. Neither established large-scale terminologies, nor dedicated medical knowledge representation languages such as GRAIL achieve this goal. We look for a general solution within the framework of terminological knowledge representation.

In the following, we present a formal model that accounts for these phenomena. It incorporates both previous work on large-scale medical coding systems and description logics (Schulz, Price, & Brown, 1997; Schmolze & Marks, 1991).

An Ontological Engineering Methodology for Part-Whole Reasoning

Part-Whole Hierarchies and SEP Triplets. Standard knowledge representation languages based on description logics do not permit the definition of relations as being transitive. In the following we describe a methodology how this lack of expressiveness can be overcome using the generalization hierarchy to emulate useful inferences that are typical for transitive relations. We will, moreover, show how the same formalism allows conditioned part-whole specialization without adding special features to standard description logics capabilities.

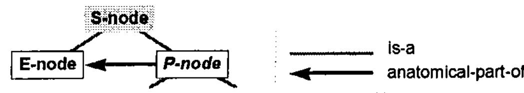


Figure 3: Basic Construct of Part-Whole Hierarchies

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 entities are modelled (cf. Fig. 3).

A triplet consists, first of all, of a composite "structure" concept, the so-called **S-node** (e.g. INTESTINE-STRUCTURE). The S-node subsumes pairs of concept siblings, namely the **E-nodes** and the **P-nodes**, that are conceptually related by the relation ANATOMICAL-PART-OF (cf. Fig. 3). The E-node denotes the *whole* organ to be modeled (e.g. INTESTINE), the P-node stands for any part of the corresponding E-node. As an example, Fig. 4 illustrates the model of a segment of the gastro-intestinal anatomy subdomain.

Let C and D be E-nodes (e.g., the organs CAECUM and APPENDIX), and $AStr$ be the top-level concept of a domain subgraph (e.g., ORGANISM-STRUCTURE). $CStr$ and $DStr$ are the S-nodes that subsume C and D, respectively, just as $CPart$ and $DPart$ 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:

$$D \sqsubseteq DStr \sqsubseteq CPart \sqsubseteq CStr \sqsubseteq .. \sqsubseteq APart \sqsubseteq AStr \quad (1)$$

$$C \sqsubseteq CStr \sqsubseteq .. \sqsubseteq APart \sqsubseteq AStr \quad (2)$$

The P-node is defined as follows:

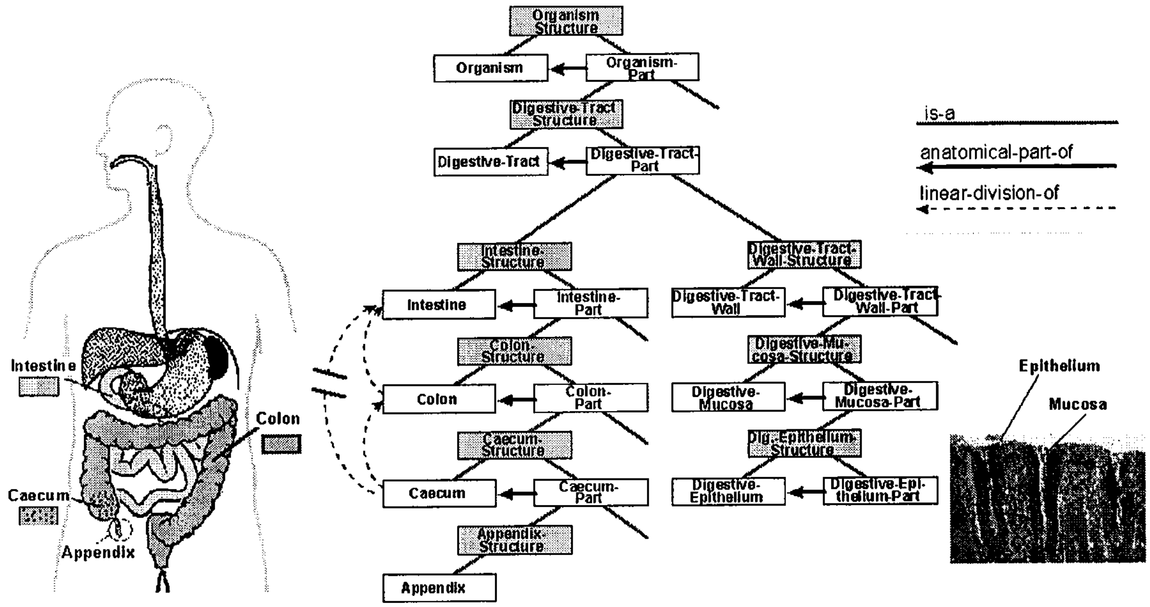
$$CPart \doteq CStr \sqcap \exists anatomical\text{-}part\text{-}of.C \quad (3)$$

Since D is subsumed by $CPart$ (1) we infer that D is an ANATOMICAL-PART-OF the organ C :

$$D \sqsubseteq \exists anatomical\text{-}part\text{-}of.C \quad (4)$$

It is obvious that this pattern holds at any level of the part-whole hierarchy. In our example (cf. Fig. 5), formula (1) 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 ascending order. In the same way, C is identified with CAECUM which is a subconcept of CAECUM-STRUCTURE, etc. (2). Between CAECUM-PART and CAECUM there exists an ANATOMICAL-PART-OF relation (3). Consequently, it can be concluded that a relation of the type ANATOMICAL-PART-OF holds between APPENDIX and CAECUM (4), but also between APPENDIX and COLON, APPENDIX and INTESTINE, COLON and INTESTINE, etc.

This *part-of inheritance* can be resumed as follows: For any concept A related to a concept B by the relation



Anatomy of the Digestive Tract: Longitudinal Division

Anatomy of the Digestive Tract: Division of the Wall

Figure 4: Part-Whole Taxonomy of the Gastrointestinal Tract, using Triplets

ANATOMICAL-PART-OF and a superconcept of B related to C by ANATOMICAL-PART-OF, too, a third relation of the same type, relating A to C, is produced by inheritance. The dotted arrows in Fig. 4 demonstrate how the construction principle can be modified in order to obviate *part-of inheritance*: This is relevant especially for subrelations of ANATOMICAL-PART-OF, such as LINEAR-DIVISION-OF: COLON is a LINEAR-DIVISION-OF INTESTINE , CAECUM is a LINEAR-DIVISION-OF COLON, but CAECUM is NOT a LINEAR-DIVISION-OF INTESTINE.

Thus, we provide an easily applicable ontology engineering methodology which incorporates part-whole reasoning by introducing a single "proto node" (*viz.* S-node) as a means to introduce reasoning about partonomies simply into IS-A taxonomies, producing similar results as an extension of the language that allows for the definition of relations as being transitive. Note, that different "dissection axes" can be modeled: both INTESTINE-STRUCTURE and DIGESTIVE-TRACT-WALL-STRUCTURE are subconcepts of DIGESTIVE-TRACT-PART (cf. Fig. 5), corresponding to the longitudinal and the radial dissection of the organ system. Morover, one S-node can be subsumed by more than one P-node.

Coordination of taxonomies (part-whole specialization). Part-whole specialization is a more generalized part-of inheritance that includes relations other than PART-OF, using the same triplet structure made of E-node, P-node and S-node. Whenever a disease concept is related to an anatomical concept, the knowledge engineer must explicitly determine whether it includes part-whole specialization or not (see the femur example from Fig. 2). Part-whole specialization is inferred when a disease concept is linked to an S-node. In order to prevent part-whole specialization it has to be connected to an E-node.

An example is shown in Fig. 5 (at the left). The concept INTESTINAL-PERFORATION - meant as the perforation of any part of the INTESTINE - is linked via the PERFORATION-OF relation to INTESTINE-STRUCTURE - an S-Node. This way, PERFORATION-OF-APPENDIX, PERFORATION-OF-CAECUM and PERFORATION-OF-COLON are classified as INTESTINAL-PERFORATION.

At the right side of Fig. 5, however, the concept ENTERITIS (an INFLAMMATION-OF the whole INTESTINE) is linked via the INFLAMMATION-OF relation to an E-node. Thus, an APPENDICITIS as an INFLAMMATION-OF the APPENDIX is not classified as being subsumed by ENTERITIS. This corresponds to the usage of these terms and, consequently, the meaning of the concepts in clinical practice.

We consider the same taxonomy as described in the terminological statements (1) to (4) Let S be a role that supports part-whole specialization, while R does not. W, X, Y, Z are concepts that stand for a PATHOLOGICAL-PHENOMENON. From

$$W \doteq \exists S.CStr \quad (5)$$

$$X \doteq \exists S.DStr \quad (6)$$

$$DStr \sqsubseteq CStr \quad (7)$$

we conclude that

$$X \sqsubseteq W \quad (8)$$

On the other hand, if we look at a role R that does not allow part-whole specialization applied to the concepts Y and Z:

$$Y \doteq \exists R.C \quad (9)$$

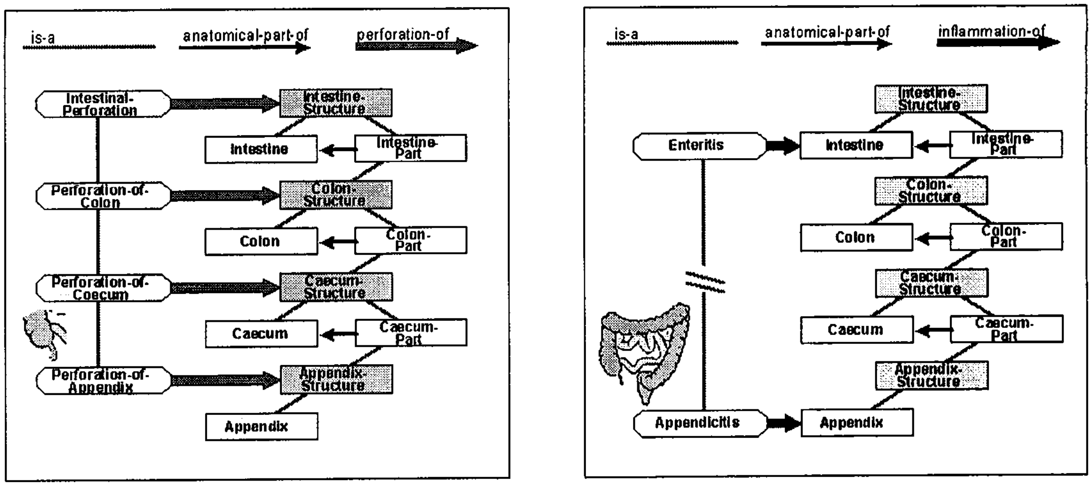


Figure 5: Conditioned Part-Whole Specialization in a Part-Whole Hierarchy. Left: Enabled Part-Whole Specialization, Right: Disabled Part-Whole Specialization

$$Z \doteq \exists R.D \qquad (10)$$

the conclusion that

$$Z \sqsubseteq Y \qquad (11)$$

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

In our example, (5) and (6) can be interpreted as follows: The INTESTINAL-PERFORATION is a PERFORATION-OF an INTESTINE-STRUCTURE and the PERFORATION-OF-APPENDIX is a PERFORATION-OF an APPENDIX-STRUCTURE. Since APPENDIX-STRUCTURE is subsumed by INTESTINE-STRUCTURE (7) it follows that a PERFORATION-OF-APPENDIX is an INTESTINAL-PERFORATION (8). ENTERITIS, however, is not linked to the S-Node INTESTINE-STRUCTURE by the role INFLAMMATION-OF, but to the E-node INTESTINE instead (9), just as APPENDICITIS is linked to the E-node APPENDIX (10). As INTESTINE does notsubsume APPENDIX no subsumption relation between APPENDICITIS ($= Z$) and ENTERITIS ($= Y$) can be inferred. (11).

Discussion

The implementation of the complete taxonomy of the tubular organs of the digestive tract (in a linear order as shown in the above example and in a radial order describing the layers of the wall of the digestive tract) follows a fixed ontology engineering scheme and is, therefore, straightforward and easy to realize.

The claim that the basic triplet pattern captures the regularities of the medical domain in an adequate manner is supported by a reasoning that allows for part-whole related inferences similar to those that transitive relations would produce. This model, however, implies trading off the number of "proto nodes" we require (S-nodes for structures) against other forms of increase in complexity, e.g., by additional part-whole-specific reasoning procedures. What's more, it seems to be a matter of taste as to whether the inclusion of these proto nodes constitutes an "artificial" effort, since one might claim that they are really needed for adequate reasoning. This is particularly true in the case where part-whole reasoning must be explicitly enabled and disabled.

Even if the transitivity assumption of the anatomical PART-OF relation, and therefore the necessity of part-of inheritance in our methodology seems to be realistic in most cases, we give the knowledge engineer enough freedom to exclude intentionally particular branches of the partonomy from part-whole reasoning when this is required.

The necessity ro relativate assertions about transitivity in meronymic relations, together with our observation that meronymic relations commonly being considered transitive (e.g., PART-OF applied to anatomical concepts) have subrelations that are explicitely not transitive [3] support our argument that, what is understood by the "transitivity or part-whole relation", is better expressed within the conceptual

[3]This defies the hypothesis that any part-whole relation can be considered "transitive as long as the meaning of 'a single sense of part' is kept" (Winston, Chaffin, & Herrmann, 1987). It can be shown using the relation LAYER-OF, a subrelation of ANATOMICAL-PART-OF, applicable to walls of organs, surfaces, or membranes. Let us consider the following assertions:

1. The wall of the intestine consists of five layers.
2. The intestinal mucosa is the innermost layer of the wall of the intestine.
3. The intestinal mucosa consists of three layers.
4. The epithelial layer is a layer of the intestinal mucosa.

Is the epithelial layer also a layer of the wall of the intestine? If the answer were true, transitivity for LAYER-OF would hold, but this is obviously not the case. There is, on the other hand, no doubt that the relation ANATOMICAL-PART-OF between intestinal mucosa and intestinal wall would be true. In examples like these, we have to face the cumbersome situation that the more general relation is transitive, while the more special one is not. If transitivity were an inheritable property of the relation this would impair the construction of a consistent relation hierarchy.

structure of the taxonomy itself than by meronymic relations with a problematic "transitivity" property.

When modeling part-whole specialization as proposed in our model, a serious limitation of the GALEN method is overcome, *viz.* that the part-whole specialization property is an invariable part of a relation definition. The solution we offer is the possibility of modifying the range of the respective relation. If the type of its range is an S-node, part-whole specialization is enabled, whereas if it is an E-node, part-whole specialization is "switched off". Likewise part-of inheritance, the part-whole specialization property is *not* contained in the relation definition, but coded in the structure of the ontology itself.

The reasons for the significant differences in the meaning of apparently similar concepts (cf. the different semantics of APPENDICITIS vs. PERFORATION-OF-APPENDIX and APPENDICITIS vs. PYELONPHRITIS) deserve a more thorough investigation. At a first glance, it seems perplexing that part-whole specialization holds in some cases, while in others it does not, even when the conceptual relations involved are the same. One explanation for this phenomenon claims an incongruence between the anatomist's formal descriptions of the organism and the clinician's view. The semantics given to concepts such as ENTERITIS may vary in comparing theoretical medicine with clinical routine. Formally, there is no doubt that the APPENDIX belongs to the DIGESTIVE-TRACT, but APPENDICITIS and ENTERITIS exhibit such an enormous difference in regard to their respective clinical manifestations and the therapies recommended. As a consequence, the part-whole relation between APPENDIX and INTESTINE becomes secondary for the clinician. Comparing, in contrast, PERFORATION-OF-APPENDIX with INTESTINAL-PERFORATION, the symptoms, signs, complications and adequate therapies are much more similar. Further analyses of the medical terminology are surely needed in order to detect those regularities where part-whole specialization is approporiate and those where it is not.

The described terminological problems concerning Part-Whole reasoning cannot be confined to the medical domain. Consider a simple commonsense scenario like the following. The car-body is clearly a part of the car. From the car-body's color we may infer the color of the car. The seats are part of a car, too. Would you, however, really want to infer the color of the car from that of the seats?

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