Towards Very Large Ontologies for Medical Language Processing

Udo Hahn  Stefan Schulz

Text Knowledge Engineering Lab
Freiburg University
Werthmannplatz 1
D-79098 Freiburg, Germany
http://www.coling.uni-freiburg.de/

Abstract
We describe an ontology engineering methodology by which conceptual knowledge is extracted from an informal medical thesaurus (UMLS) and automatically converted into a formal description logics system. Our approach consists of four steps: concept definitions are automatically generated from the UMLS source, integrity checking of taxonomic and partonomic hierarchies is performed by the terminological classifier, cycles and inconsistencies are eliminated, and incremental refinement of the evolving knowledge base is performed by a domain expert. We report on experiments with a knowledge base composed of 164,000 concepts and 76,000 relations.

1. Introduction

Medical language processing (for a survey, cf. Spyns (1996)) is an application area of natural language processing, which deals with the analysis of medical documents such as discharge, X-ray, admission, and finding reports. The sublanguage patterns these documents exhibit reveal that understanding their contents, whether by humans or machines, requires access to knowledge and the terminology of the medical domains being covered.

Unlike many other disciplines, medicine has a long standing tradition in assembling and structuring medical knowledge, e.g., disease taxonomies, medical procedures, anatomical terms, etc., in a wide variety of medical terminologies, thesauri and classification systems. These efforts are typically restricted to the provision of broader and narrower terms, related terms or (quasi-)synonymous terms. This is most evident in the UMLS, the Unified Medical Language System (McCray and Nelson, 1995; McCray, 1998), an umbrella system which covers more than 60 medical thesauri and classification systems. Its metathesaurus component contains more than 800,000 concepts which are structured in hierarchies and classified by 134 semantic types (provided by the UMLS Semantic Network). Their semantics is shallow and entirely intuitive, which is due to the fact that their usage was primarily intended for humans as a backbone for various forms of clinical knowledge management, e.g., cross-mapping between different terminologies, disease and procedure encoding, etc.

Given the size, the evolutionary diversity and inherent heterogeneity of the UMLS, there is no surprise that the lack of a formal semantic foundation leads to inconsistencies, circular definitions, etc. (Cimino, 1998). This may not cause utterly severe problems when humans are in the loop and its use is limited to disease encoding, accountancy or document retrieval tasks. However, anticipating its use for more knowledge-intensive applications such as natural language understanding of medical narratives (Hahn et al., 1999b), those shortcomings might lead to an impasse.

As a consequence, formal models for dealing with medical knowledge have been proposed, using representation mechanisms based on conceptual graphs, semantic nets, or description logics (Cimino et al., 1994; Mays et al., 1996; Rector et al., 1997; Volot et al., 1998; Gangemi et al., 1999). Not surprisingly, there is a price to be paid for more expressiveness and formal rigor, viz., increasing modeling efforts and, hence, increasing maintenance costs. Operational systems making full use of this rigid approach, especially those which employ high-end knowledge representation languages, are usually restricted to rather small subdomains. The most comprehensive of these sources we know of is the GRAIL-encoded GALEN knowledge base which covers up to 9,800 concepts (Rector et al., 1997). The limited coverage then hampers their routine usage, an issue which is always highly rewarded in the medical informatics community.

The knowledge bases developed within the framework of the above-mentioned terminological systems have almost all been designed from scratch – without making systematic use of the large body of knowledge contained in those medical terminologies. An intriguing approach would be to join the massive coverage offered by informal medical terminologies with the high level of expressiveness supported by rigid knowledge representation systems in order to develop formally solid medical knowledge bases on a larger scale. In the paper, we describe such a knowledge engineering methodology. The resulting medical ontology forms the domain knowledge backbone of MEDSYNDIKATE, a system for the automatic acquisition of factual and evaluative (subjective) knowledge from medical finding reports (Hahn et al., 1999b; Hahn et al., 1999a). Its size, more than 240,000 concepts and roles, is truly competitive and supplies the system with a fair amount of medical knowledge in the core areas of anatomy and pathology.

2. Reasoning Along Part-Whole Hierarchies

Medical ontologies are organized around taxonomic (IS-A relation) and partonomic (PART-OF relation) knowledge. Hence, medical knowledge representation efforts have to take account of both hierarchy types and the reasoning patterns they imply.1

1In an exploratory quantitative study of the particular text genre we are dealing with, viz., medical finding reports, we found
Partonomic knowledge has been an issue within diverse areas ranging from philosophy (mereology), data modeling for database systems and object-oriented programming, to knowledge representation proper in the field of artificial intelligence. Major strands of this work are discussed by Artale et al. (1996) under the heading of object-centered representation approaches. This also includes the description logic (DL) paradigm to which we subscribe in our work, too. From our application domain, the need arises to have formally solid inference mechanisms for taxonomic (generalization hierarchies), as well as partonomic reasoning (part-whole hierarchies) available within a uniform representation model. We also require an inference engine which performs this style of advanced reasoning on large data sets (\( \geq 10,000 \) items). Hence, we consider KL-ONE-type descriptions logics (Woods and Schmolze, 1992), at the formal representation level, and LOOM’s classification-based inference machine (MacGregor and Bates, 1987; MacGregor, 1994), at the system level, the most convenient match of our requirements and the current state of the art in terminological reasoning.

Unlike generalization-based reasoning in concept taxonomies, no fully conclusive mechanism exists up to now for reasoning along part-whole hierarchies. In the description logics community several language extensions have been proposed which provide special constructors for part-whole reasoning (Rector et al., 1997; Horrocks and Sattler, 1999). This seems a reasonable way to proceed, as long as the transitivity property of a relation can be assumed, in general. In the medical (Hahn et al., 1999c) as well as commonsense domains (Cruse, 1979), however, various exceptions exist such that the transitivity of part-of relations cannot be granted, in general. Hence, both the expression of regular transitive use, as well as exception handling for nontransitive part-of relations have to be taken into consideration. Motivated by informal approaches sketched by Schmolze and Mark (1991) and Schulz et al. (1997), we formalized a model of partonomic reasoning (Hahn et al., 1999c) that meets the above requirements and also does not exceed the expressiveness of the well-understood, parsimonious concept language \( ALC \) (Woods and Schmolze, 1992).

Our style is centered around a particular data structure for partonomic reasoning, so-called SEP triplets (cf. Figure 1). They define a characteristic pattern of IS-A hierarchies which support the emulation of inferences typical of transitive part-of relations. In this formalism, the relation ANATOMICAL-PART-OF describes the partitive relation between physical parts of an organism.

(Hahn et al., 1999a) that 45% of the (indirect) reference relations between sentences are due to bridging or functional anaphora, which refer heavily to partonomic knowledge. On the other hand, 34% of the (direct) reference relations are due to nominal anaphora, which bring taxonomic knowledge to bear.

\( ALC \) allows for the construction of hierarchies of concepts and relations, where \( \sqsubseteq \) denotes subsumption and \( \equiv \) definitional equivalence. Existential (\( \exists \)) and universal (\( \forall \)) quantification, negation (\( \neg \)), disjunction (\( \lor \)) and conjunction (\( \land \)) are supported. Role filler constraints (e.g., typing by \( C \)) are linked to the relation name \( R \) by a dot, \( \exists RR.C \).

A triplet consists, first of all, of a composite ‘structure’ concept, the so-called S-node (e.g., HAND-STRUCTURE). Each Structure concept subsumes directly both an anatomical Entity and each of the anatomical Parts of this entity, i.e., the corresponding E-node and P-node, e.g., HAND and HAND-PART, respectively. Unlike entities and their parts which refer to specific ontological objects, structures have no physical correlate in the real world — they constitute a representational artifact required for the formal reconstruction of systematic patterns of partonomic reasoning. The E-node denotes the whole anatomical entity to be modeled, whereas the P-node is the common subsumer of those concepts that have their role ANATOMICAL-PART-OF filled by the corresponding E-node concept, as an existential condition. Hence, for every P-node there exists a corresponding E-node for the role ANATOMICAL-PART-OF. A reconstruction of some basic anatomical relations in terms of SEP triplets is illustrated in Figure 2.

The reconstruction of the relation ANATOMICAL-PART-OF by taxonomic reasoning proceeds as follows. Let us assume that \( C_E \) and \( D_E \) denote E-nodes, \( C_S \) and \( D_S \) denote the S-nodes that subsume \( C_E \) and \( D_E \), respectively, and \( C_P \) and \( D_P \) denote the P-nodes related to \( C_E \) and \( D_E \), respectively, via the role ANATOMICAL-PART-OF (cf. Figure 1). These conventions can be captured by the following terminological expressions:

\[
C_E \sqsubseteq C_S \sqsubseteq D_P \sqsubseteq D_S \tag{1}
\]

\[
D_E \sqsubseteq D_S \tag{2}
\]

The P-node is defined as follows (note the disjointness between \( D_E \) and \( D_P \), i.e., no anatomical concept can be ANATOMICAL-PART-OF itself):

\[
D_P \equiv D_S \sqcap \neg D_E \sqcap \exists \text{anatomical-part-of} . D_E \tag{3}
\]
and, finally, (4) manual curation and refinement of the formal representation structures. These four steps are illustrated by the workflow diagram depicted in Figure 4.

Step 1: Automatic Generation of Terminological Expressions. Sources for concepts and relations are the UMLS semantic network and the mrrel, mrcon and mrsty tables of the 1999 release of the UMLS metathesaurus. The mrrel table which contains approximately 7.5 million records (for a fragment, cf. Figure 5) exhibits the semantic links between two UMLS CUIs (concept unique identifier): the mrcon table contains the concept names and mrsty keeps the semantic type(s) assigned to each CUI. These tables, available as ASCII files, were imported into a Microsoft Access relational database and manipulated using SQL embedded in the VBA programming language. For each CUI in the mrrel subset its alphanumerical code was substituted by the English preferred term found in mrcon.

![Diagram](image_url)

Figure 3: Enabling/Disabling Transitivity in a SEP-Encoded Partonomy

Since \( C_E \) is subsumed by \( D_F \) (according to (1)), we infer that the relation ANATOMICAL-PART-OF holds between \( C_E \) and \( D_E \), too:

\[
C_E \subseteq \exists \text{anatomical-part-of}.D_E
\]

The encoding of concept hierarchies in terms of SEP triplets allows the knowledge engineer to switch the transitivity property of part-whole relations off and on, dependent on whether the E-node or the S-node is addressed as the target concept for a conceptual relation. In the first case, the propagation of roles across part-whole hierarchies is disabled, in the second case it is enabled. As an example (cf. Figure 3), Enteritis is defined as HAS-LOCATION INTESTINE. The range of the relation HAS-LOCATION is restricted to the E-node of INTESTINE. This precludes, for instance, the classification of APPENDICITIS as ENTERITIS though the APPENDIX is related to the INTESTINE via an ANATOMICAL-PART-OF relation. GLOMERULONEPHRITIS (HAS-LOCATION GLOMERULUM \(_2\)), however, is classified as NEPHRITIS (HAS-LOCATION KIDNEY \(_2\)), since the GLOMERULUM is an ANATOMICAL-PART-OF the KIDNEY. In the same way, PERFORATION-OF-APPENDIX is classified as INTESTINAL-PERFORATION (cf. Hahn et al. (1999c) for an in-depth analysis of these phenomena).

3. Knowledge Import and Refinement

Our goal is to extract conceptual knowledge from two major subdomains of the UMLS, viz., anatomy and pathology, in order to construct a formally sound knowledge base using a terminological knowledge representation language. This task will be divided into four steps: (1) automatic generation of terminological expressions, (2) automatic consistency checking by a terminological classifier, (3) manual restititution of formal consistency in case of inconsistencies, and, finally, (4) manual curation and refinement of the formal representation structures. These four steps are illustrated by the workflow diagram depicted in Figure 4.

![Diagram](image_url)

Figure 5: Semantic Relations in the UMLS Metathesaurus

After a manual remodeling of the 135 top-level concepts and 247 relations of the UMLS semantic network, we extracted, from a total of 85,899 concepts, 38,059 anatomy and 50,087 pathology concepts from the metathesaurus. The criterion for the inclusion into one of these sets is the assignment to predefined semantic types. Also, 2,247 concepts were found to be included in both sets, anatomy and pathology. Since we wanted to keep the two subdomains strictly disjoint, we maintained these 2,247 concepts duplicated, and prefixed all concepts by ANA- or PAT- according to their respective subdomain. This can be justified by the observation that these hybrid concepts exhibit, indeed, multiple meanings. For instance, TUMOR has the meaning of a malignant disease on the one hand, and of an anatomical structure on the other hand.

As target structures for the anatomy domain we chose SEP triplets. These are expressed in the terminology language LOOM which we had previously extended by a special DEFTRIPLET macro (cf. Table 1 for an example). Only UMLS supplied part-of, has-part and is-a relation attributes were considered for the construction of taxonomic and partonomic hierarchies (cf. Figure 4). Hence, for each anatomy concept one SEP triplet was created. The result is a mixed IS-A and PART-WHOLE hierarchy a straightforward example of which is depicted in Figure 2.

As a convention in UMLS, any two CUIs must be connected by at least a shallow relation (in Figure 5, CHID relations in the column REL are assumed between CUIs). Shallow relations may be refined in the column RELA, if a thesaurus is available which contains more specific information. Some CUIs are linked either by part-of or is-a. In any case, the source thesaurus for the relations and the CUIs involved is specified in the columns X and Y (e.g., MeSH 1999, SNOMED International 1998).
| UMLS relation number of links | Automatic generation of Loom definitions, augmented by P-Loom language elements | Submission to Loom classifier, Validation for formal consistency by Loom | Manual restitution of formal consistency | Manual rectification and refinement of the resulting knowledge base |
|-----------------------------|---------------------------------------------------------------------------------|------------------------------------------------------------------|----------------------------------------|---------------------------------------------------------|
| sibling_of                  | 267.218 | add negations in order to express taxonomic or partitive disjointness          |                                                                     |                                         |
| child_of                    | 59.808 | include related concepts into :is-primitive or :part-of clause where plausible  |                                                                     |                                         |
| narrower_term               | 24.223 |                                                                     |                                                                     |                                         |
| isa                         | 9.755  | check for definitional cycles                                               | remove taxonomic parent concepts                                   | substitute of primitive links by non-primitive ones where possible |
| location_of                 | 4.803  |                                                                     |                                                                     |                                         |
| has_location                | 4.803  | include related concepts into :has-part clause where plausible               |                                                                     |                                         |
| has_part                    | 4.321  | check whether this part is mandatory (under "real-anatomy" assumption)       |                                                                     |                                         |
| has_conceptual_part         | 128    |                                                                     |                                                                     |                                         |
| part_of                     | 4.321  | 1. check for partonomic cycles                                              | 1. remove partonomic or taxonomic parent concepts                   | check for plausibility and completeness |
| conceptual_part_of          | 128    | 2. check for disjointness between E and P node                              | 2. redefine triplet as single concept                               |                                         |
| parent                      | 59.808 | include related concepts into :has-part clause where plausible               |                                                                     |                                         |
| broader_term                | 24.223 |                                                                     |                                                                     |                                         |
| inverse_isa                 | 9.755  |                                                                     |                                                                     |                                         |
| associated_with             | 14     |                                                                     |                                                                     |                                         |
| mapped_from                 | 2.643  |                                                                     |                                                                     |                                         |
| other_relation              | 10.908 |                                                                     |                                                                     |                                         |
| qualified_by                | 1.864  |                                                                     |                                                                     |                                         |
| allowed_qualified           | 1.864  |                                                                     |                                                                     |                                         |
| mapped_to                   | 2643   |                                                                     |                                                                     |                                         |
| <other named relations>     | 11.888 | check for inherited constraints                                             | remove constraints                                                  | remove or add constraints                       |

**Pathology Concepts Linked to Pathology Concepts**

| sibling_of                  | 457.342 | add negations in order to express taxonomic disjointness          |                                                                     |                                         |
| child_of                    | 72.426  | check for definitional cycles                                               | remove parent concepts                                   | substitute primitive links by non-primitive ones whenever possible |
| narrower_term               | 26.972  | :is-primitive                                                             |                                                                     |                                         |
| isa                         | 3.635   |                                                                     |                                                                     |                                         |
| inverse_isa                 | 3.635   |                                                                     |                                                                     |                                         |
| associated_with             | 13.902  |                                                                     |                                                                     |                                         |
| mapped_to                   | 15.024  | <do nothing>                                                             |                                                                     |                                         |
| mapped_from                 | 15.024  |                                                                     |                                                                     |                                         |
| part_of                     | 1       | <do nothing>                                                             |                                                                     |                                         |
| has_part                    | 72.426  |                                                                     |                                                                     |                                         |
| parent                      | 28.972  |                                                                     |                                                                     |                                         |
| other_relation              | 25.796  |                                                                     |                                                                     |                                         |
| qualified_by                | 6.255   |                                                                     |                                                                     |                                         |
| allowed_qualified           | 6.255   |                                                                     |                                                                     |                                         |
| <other named relations>     | 4.162   | <some x> check for inherited constraints                                  | remove constraints                                                  | remove or add constraints                       |

**Pathology Concepts Linked to Anatomy Concepts**

| CUIpat = CUIana              | 2.247   | plausibility check of concept "duplication" (assignment to both domains) |                                                                     |                                         |
| <missing>                   |         | <do nothing>                                                             |                                                                     |                                         |
| associated_with             | 2.314   | check for consistency                                                    | render links complete, link to E-node instead of S-node when role propagation has to be disabled |
| has_location                | 9.230   | <some x> has_location                                                    |                                                                     |                                         |

Figure 4: Workflow Diagram for the Construction of a Loom Knowledge Base from the UMLS
For the pathology domain, we treated \textit{CHD} (child) and \textit{RN} (narrower relation) from the UMLS as indicating taxonomic links. No part-whole relations were considered, since this category does not apply to the pathology domain. Furthermore, for all anatomy concepts contained in the definitions of pathology concepts the S-node is the default concept to which they are linked, thus enabling the propagation of roles across the part-whole hierarchy.

As a fundamental semantic assumption all roles generated in this process were considered as existentially quantified. This means that any relation \( r \) (\textit{PART-OF}, \textit{HAS-LLOCATION}, etc.) which holds between two concepts, \( A \) and \( B \), is mapped to a role \( R.B \) which is a necessary condition in the definition of the concept \( A \). All conceptual constraints for a concept definition are mapped to a conjunction of constraints.

In both subdomains, shallow relations such as the extremely frequent sibling relation (\textit{SIB}) were included as comments into the code to provide heuristic guidance for the subsequent manual refinement phase.

**Step 2: Automatic Consistency Checking by the LOOM Classifier.** The import of UMLS anatomy concepts resulted in 38,059 DEFTREPILET expressions for anatomical concepts and 50,087 DEFCONCEPT expressions for pathological concepts. Each DEFTREPILET was expanded into three DEFCONCEPT (\textit{S}-, \textit{E}-, and \textit{P}-nodes), and two DEFRELATION (ANATOMICAL-PART-OF-X, INVANATOMICAL-PART-OF-X) expressions, summing up to 114,177 concepts. This yielded (together with the concepts from the UMLS semantic network) a total of 240,764 definiency LOOM expressions.

From 38,059 anatomy triplets, 1,219 DEFTREPILET statements contained a \textit{HAS-PART} clause followed by a list of a variable number of triplets, with more than one argument in 823 cases (average cardinality: 3.3). 4,043 DEFTREPILET statements contained a \textit{PART-OF} clause, only in 332 cases followed by more than one argument (average cardinality: 1.1). The resulting knowledge base was then submitted to the terminological classifier and checked for terminological cycles and consistency. In the anatomy subdomain, one terminological cycle and 2,328 inconsistent concepts were found, in the pathology subdomain 355 terminological cycles though not a single inconsistent concept were determined (cf. Table 2).

**Step 3: Manual Restitution of Consistency.** The inconsistencies in the anatomy part of the knowledge base identified by the classifier could all be traced back to the simultaneous linkage of two triplets by both \textit{is-a} and \textit{part-of} links, an encoding that raises a conflict due to the disjointness required for corresponding \textit{P-} and \textit{E}-nodes. In most of these cases the affected parents belonged to a class of concepts that obviously cannot be appropriately modeled as SEP triplets, e.g., \textit{SUBDIVISION-OF-ASCENDING-AORTA} or \textit{ORGAN-PART}. The meaning of each of these concepts almost paraphrases that of a \textit{P}-node, so that the violation of the SEP-internal disjointness condition could be accounted for by substituting the involved triplets with simple LOOM concepts, by matching them with already existing \textit{P}-nodes or by disabling \textit{IS-A} or \textit{PART-OF} links.

In the pathology part of the knowledge base, we expected a large number of terminological cycles, as a consequence of interpreting the thesaurus-style \textit{narrower term} and \textit{child} relations in terms of taxonomic subsumption (\textit{IS-A}). Bearing in mind the size of the knowledge base, we consider 355 cycles a tolerable number. Those cycles were primarily due to very similar concepts, e.g., \textit{ARTERIOSCLEROSIS} vs. \textit{ATHEROSCLEROSIS}, \textit{AMAUROSIS} vs. \textit{BLINDNESS}, and residual categories ("other", "NOS" = \textit{not otherwise specified}). These were directly inherited from the source terminologies and are notoriously difficult to interpret out of their definitional context, e.g., \textit{OTHER-MALIGNANT-NEOPLASM-OF-SKIN} vs. \textit{MALIGNANT-NEOPLASM-OF-SKIN-NOS}.

The cycles were analyzed and a negative list which consisted of 630 concept pairs was manually derived. In a subsequent extraction cycle we incorporated this list in the automated construction of the LOOM concept definitions and, with these new constraints, a fully consistent knowledge base was generated.

**Step 4: Manual Rectification and Refinement of the Knowledge Base.** To set up this high-volume knowledge base including the aforementioned working steps required three months of work for a single person, in total. The fourth step – when performed for the whole knowledge base – is very time-consuming and requires broad and in-depth medical expertise. An analysis of random samples from both subdomains is currently being performed by the second author, a domain expert. The preliminary data we here supply refer to the analysis of two random samples of each 100 anatomy and 100 pathology concepts. This took one person about a single month. From the experience we gained in the anatomy and pathology subdomains so far, the following workflow can be derived:

- **Checking the correctness of the taxonomic and partenomic hierarchies.** Taxonomic and partenomic links are manually added or removed. Primitive subsumption is substituted by non-primitive one whenever possible. This is a crucial point, because the automatically generated hierarchies contain only information about the parent concepts and necessary conditions.

| Triplets | Anatomy | Pathology |
|----------|---------|-----------|
| 38,059   |         | 50,087    |
| 114,177  |         | 355       |
| 2,328    | 0       |           |

Table 2: Classification Results
As an example, the automatically generated definition of DERMATITIS includes the information that it is an INFLAMMATION, and that the role HAS-LOCATION must be filled by the concept SKIN. An INFLAMMATION that HAS-LOCATION SKIN, however, cannot automatically be classified as DERMATITIS.

Results: In the anatomy sample, only 76 concepts out of 100 could be unequivocally classified as belonging to ‘canonical’ anatomy. (The remainder, e.g., ANA-PHALANX-OF-SUPERNUMERARY-DIGIT-OF-HAND, referring to pathological anatomy was immediately excluded from analysis.) Besides the assignment to the UMLS semantic types, only 27 (direct) taxonomic links were found. 83 UMLS relations (mostly “child” or “narrower” relations) were manually upgraded to taxonomic links. 12 (direct) part-of and 19 has-part relations were found. Four part-of relations and one has-part relation had to be removed, since we considered them as implausible. 51 UMLS relations (mostly “child” or “narrower” relations) were manually upgraded to part-of relations, and 94 UMLS relations (mostly “parent” or “broader” relations) were upgraded to has-part relations. After this workup and upgrade of shallow UMLS relations to semantically more specific relations, the sample was checked for completeness again. As a result, 14 is-a and 37 part-of relations were still considered missing.

In the pathology sample, the assignment to the pathology subdomain was considered plausible for 99 of 100 concepts. A total of 15 false is-a relations were identified in 12 concept definitions. 24 is-a relations were found to be missing.

- Check of the :has-part arguments assuming ‘real anatomy’. In the UMLS sources part-of and has-part relations are considered as symmetric. According to our transformation rules, the attachment of a role HAS-ANATOMICAL-PART to an E-node BE with its range restricted to A_E implies the existence of a concept A for the definiion of a concept B. On the other hand, the classification of A_E as being subsumed by the P-node BP, the latter being defined via the role ANATOMICAL-PART-OF restricted to BE, implies the existence of B_E given the existence of A_E. These constraints do not always conform to ‘real’ anatomy, i.e., anatomical concepts that may exhibit pathological modification. Figure 6 (left) sketches a concept A that is necessarily ANATOMICAL-PART-OF a concept B, but whose existence is not required for the definiion of B. This is typical of the results of surgical interventions, e.g., a large intestine without an appendix, or an oral cavity without teeth, etc.

Results: All 112 has-part relations obtained by the automatic import and the manual workup of our sample were checked. The analysis revealed that more than half of them (62) should be eliminated in order not to obviate a coherent classification of pathologically modified anatomical objects. As an example, most instances of ILEUM do not contain a MECKEL’S DIVERTICULUM, whereas all instances of MECKEL’S DIVERTICULUM are necessarily ANATOMICAL-PART-OF ILEUM. Many surgical interventions that remove anatomical structures (appendix, gallbladder, etc.), produce similar patterns. In our formalism, this corresponds to a single taxonomic link between an S-node and a P-node (cf. Figure 6, left part). The contrary is also possible (cf. Figure 6, right part): the definiion of A_E does not imply that the role ANATOMICAL-PART-OF be filled by B_E, but B_E does imply that the inverse role be filled by A_E. As an example, a LYMPH-NODE necessarily contains LYMPH-FOLLICLES, but there exist LYMPH-FOLLICLES that are not part of a LYMPH-NODE. This pattern is typical of the meroelogical relation between macroscopic (countable) objects, such as organs, and multiple uniform microscopic objects.

- Analysis of the sibling relations and defining concepts as being disjoint. In UMLS, the SIB relation links concepts that share the same parent in a taxonomic or partonomic hierarchy. Pairs of sibling concepts may have common descendants or not. If not, they constitute the root of two disjoint subtrees. In a taxonomic hierarchy, this means that one concept implies the negation of the other (e.g., a benign tumor cannot be a malignant one, et vice versa). In a partitive hierarchy, this can be interpreted as spatial disjointness, viz. one concept does not spatially overlap with another one. As an example, ESOPHAGUS and DUODENUM are spatially disjoint, whereas STOMACH and DUODENUM are not (they share a common transition structure, called PYLORUS), such as all neighbor structures that have a surface or region in common. Spatial disjointness can be modeled so that the definiion of the S-node of the concept A implies the negation of the S-node of the concept B (Schulz et al., 2000).

Results: We found on the average 6.8 siblings per concept in the anatomy domain, 8.8 in the pathology domain. So far, the analysis of sibling relations has been performed only for the anatomy domain. From a total of 521 sibling relations, 9 were identified as is-a, 14 as part-of, and 17 as has-part, whereas 404 referred to topologically disconnected concepts.

4In Table 1 the concepts marked by italics, viz. AORTIC-VALVE and PULMONARY-VALVE should be eliminated from the :HAS-PART list, because they may be missing in certain cases as a result of congenital malformations, inflammatory processes or surgical interventions.
Completion and modification of anatomy–pathology relations. Surprisingly, only very few pathology concepts contained an explicit reference to a corresponding anatomy concept. These relations must, therefore, be added by a domain expert. In each case, the decision must be made whether the E-node or the S-node has to be addressed as the target concept for modification such that the propagation of roles across part–whole hierarchies is disabled or enabled.

Results: In the sample we found 522 anatomy–pathology relations, from which 358 (69%) were judged incorrect by the domain experts. In 36 cases an adequate anatomy–pathology relation was missing. All 164 HAS-LOCATION roles were analyzed as to whether they were to be filled by an S-node or an E-node of an anatomical triplet. In 153 cases, the S-node (which allows propagation across the part–whole hierarchy) was considered to be adequate, in 11 cases the E-node was preferred. The analysis of the 100 pathology concepts revealed that only 17 were to be linked with an anatomy concept. In 15 cases, the default linkage to the S-node was considered to be correct, in one case the linkage to the E-node was preferred, in another case the linkage was considered to be false.

The high number of implausible constraints points to the lightweight semantics of HAS-LOCATION links in the UMLS sources. While we interpreted them in terms of a conjunction for the import routine, a disjunctive meaning seems to prevail implicitly in many definiions of top-level concepts such as TUBERCULOSIS. In this example, we find all anatomical concepts that can be affected by this disease linked by HAS-LOCATION. All these constraints (e.g., HAS-LOCATION URINARY-TRACT) are inherited to subconcepts such as TUBERCULOSIS-OF-BRONCHUS. A thorough analysis of the top–level pathology concepts is necessary, and conjunctions of constraints will have to be substituted by disjunctions where necessary.

4. Discussion and Conclusions

In medical language processing, knowledge of the underlying domain has to be supplied on a larger scale. Instead of developing sophisticated medical knowledge bases from scratch, we here propose a ‘conservative’ approach — reuse existing large-scale resources, but refill the data from these resources so that advanced representational requirements imposed by more expressive knowledge representation languages are met. The resulting knowledge bases can then be used for sophisticated applications requiring formally sound medical reasoning such as text understanding.

The benefits and problems of converting conceptual knowledge from semantically weak species cations to a rigorous knowledge representation formalism have been described by Pisanelli et al. (1998). They extracted knowledge from the UMLS semantic network, as well as from parts of the metathesaurus and converted it into a description logics system. Spackman and Campbell (1998) describe how the SNOMED nomenclature evolves from a multi-axial coding system into a formally founded ontology. Their general goal is to avoid ambiguous or semantically invalid representations of composite concepts. However, both approaches do not provide a special reasoning mechanism for partonomic relations.

Within the formal framework of GALEN, a fragment of the Read Thesaurus was translated into GRAIL, a knowledge representation system also based on description logics (Rogers et al., 1998). In a cross-validation study it was checked, on the one hand, whether the definitions contained in the Read Thesaurus were logically consistent and, on the other hand, whether the GRAIL domain model was rich enough to encode them. Although GRAIL comes with a special-purpose reasoning mechanism dedicated to partonomies, the adaptation was limited to simple generic hierarchies as only these structure the Read Thesaurus.

The developers of VOXEL-MAN (Schubert and Höhne, 1998), a multimedia tutoring systems for anatomy, and of the Digital Anatomist (UWDA), an anatomical semantic network (Rosse et al., 1998), have both emphasized partitive hierarchies though at an informal level. Whereas in VOXEL-MAN a fine-grained ontology of partonomic relations is sketched that accounts for various part–whole relations found in the anatomy domain, the UWDA developers restrict themselves to a small set of relations leading to a precise separation between partonomic and taxonomic hierarchies. They excel with a high granularity of description and a broad coverage.

Our approach tries to combine the broad coverage and fine-grained concept descriptions of the UWDA with the formal rigor of description logics. Additionally, we enhance the imported knowledge with part–whole specific reasoning capabilities indispensable in the medical domain, though this has already been described as a hard problem for terminological languages (Haimowitz et al., 1988).

Several alternatives have been brought forward to solve this problem within the DL framework. The first approach extends a basic language definiions (usually, ACC) by generally allowing role hierarchies and transitivity of roles (Horrocks and Sattler, 1999), or by supplying dedicated transitive partonomy operators such as for the GRAIL language (Rector et al., 1997). This way, the transitivity property is hard-wired to role defniions and assumed to hold, in general. This contradicts empirical data from anatomical ontologies as discussed in Hahn et al. (1999c), which reveal that the same relation may be transitive and nontransitive depending on the anatomical context.

Alternatively, one may want to preserve standard language definiions for reasons of simplicity and parsimony. Accordingly, Schmolze and Mark (1991) propose a ‘data structure’ 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, however, that these additional concepts are necessary from an ontological point of view, as the distinct mechanisms for conditioned specialization modeling reveal (cf. Figure 3).

It remains to be seen whether conservative structural extensions of a stable language platform are able to carry over to the many varieties of partonomic reasoning and differ-
ent part-whole relations, 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. We have evidence that the triplet mechanism we here propose can be straightforwardly extended to cover mereotopological and (limited) spatial reasoning, as well (Schulz et al., 2000; Schulz and Hahn, 2001).

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