International Journal of Medical Informatics 64 (2001) 207–221

International Journal of Medical Informatics

www.elsevier.com/locate/ijmedinf

Medical knowledge reengineering—converting major portions of the UMLS into a terminological knowledge base

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Abstract

We describe a semi-automatic knowledge engineering approach for converting the human anatomy and pathology portion of the UMLS metathesaurus into a terminological knowledge base. Particular attention is paid to the proper representation of part-whole hierarchies, which complement taxonomic ones as a major hierarchy-forming principle for anatomical knowledge. Our approach consists of four steps. First, concept definitions are automatically generated from the metathesaurus, with LOOM as the target language. Second, integrity checking of the emerging taxonomic and partonomic hierarchies is automatically carried out by the terminological classifier. Third, terminological cycles and inconsistencies are manually eliminated and, in the last step, the knowledge base built this way is incrementally refined by a medical expert. Our experiments were run on a terminological knowledge base which is composed of 164 000 concepts and 76 000 relations. Empirical evidence for the lack of logical consistency, adequacy and improper granularity of the UMLS knowledge source is given, and finally, assessments of what kind of efforts are needed to render the formal target representation structures complete and empirically adequate. © 2001 Elsevier Science Ireland Ltd. All rights reserved.

Keywords: UMLS; Description logics; Anatomy; Pathology

1. Introduction

The health care domain and the biomedical sciences are somewhat unique compared with other scientific areas, since large portions of their terminological knowledge are already structured in terms of controlled terminologies, classification systems and thesauri. According to the different tasks they have been designed for, such as statistics, clinical communication, accounting or document indexing, they exhibit considerable variability both in terms of coverage and granularity. Also the way knowledge is organized differs between heterogeneous types of medical terminologies [1,2]. *Classifications* aim at providing exhaustive sets of mutually exclusive categories (or classes) such as the *International Classification of Diseases* (ICD) [3]. More complex systems such as *nomenclatures* (e.g.

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PII: S1386-5056(01)00201-5

SNOMED [4] or NHS clinical terms [5]) and *thesauri* (e.g. MeSH [6]) provide additional descriptive flexibility by way of compositionality of concepts, polyhierarchies and semantic links—often, however, at the price of increasing ambiguity and semantic vagueness. Although various kinds of medical terminologies are well adapted to different needs, the demand for homogeneous multi-purpose terminology servers has been increasingly expressed [7–11].

The '*Unified Medical Language System*' (UMLS) [12] can be considered as a direct response to this request. It contains about 800 000 concepts from more than 60 different classifications, nomenclatures and thesauri, all of which have been merged into the UMLS Metathesaurus. Additional semantic structure can be imposed on concepts by using 134 semantic types, provided by the UMLS Semantic Network, together with 54 semantic relations.[1]http://umlsinfo.nlm.nih.gov/ Given its size, evolutionary diversity and inherent heterogeneity, there is no surprise at all that the lack of a solid formal foundation leads to a bunch of inconsistencies, circular definitions, etc. [13,14]. This may not cause utterly severe problems when humans are in the loop and its use is limited to tasks such as those mentioned above. However, anticipating its use for more knowledge-intensive applications, such as natural language understanding of medical narratives [15] or medical decision support systems [16], 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 networks or description logics [17–19]. Not surprisingly, there is also a price to be paid for more expressiveness and formal rigor in terms of increasing modeling efforts and, hence, increasing maintenance costs. Therefore, concrete medical knowledge bases making full use of this rigid approach, especially those which employ high-end, KL-ONE-style knowledge representation languages (for a survey, cf. [20]), are usually restricted to rather small subdomains. Those systems developed within the framework of the abovementioned formal approaches have all been designed from scratch—without making systematic use of the large body of knowledge contained in informal medical terminologies.

An intriguing approach would be to combine the massive *coverage* offered by informal medical terminologies with the high level of *expressiveness* supported by formally solid knowledge representation systems in order to develop sophisticated medical knowledge bases on a larger scale. This idea has already been fostered by Pisanelli et al. [10], who extracted knowledge from the UMLS semantic network as well as from parts of the metathesaurus and merged it with generic ontologies from other sources. In a similar way, Spackman and Campbell [21] describe how SNOMED [4] can be transformed from a multi-axial coding system into a formally founded ontology. Unfortunately, efforts up to now are entirely focused on taxonomic reasoning along generalization hierarchies (expressed by *is-a* relations) and lack a reasonable coverage of part-whole (i.e. *part-of* or *has-part*) relationships, a second major conceptual construct needed for reasoning in the anatomy domain, in particular.

This article is organized as follows. In Section 2, we argue for the relevance of part-whole reasoning for the medical domain and introduce a representation model which is rooted in a description logics framework [20]. In particular, we propose a tripartite data structure for encoding anatomical concepts in

[1] UMLS is accessible via http://umlsinfo.nlm.nih.gov/.

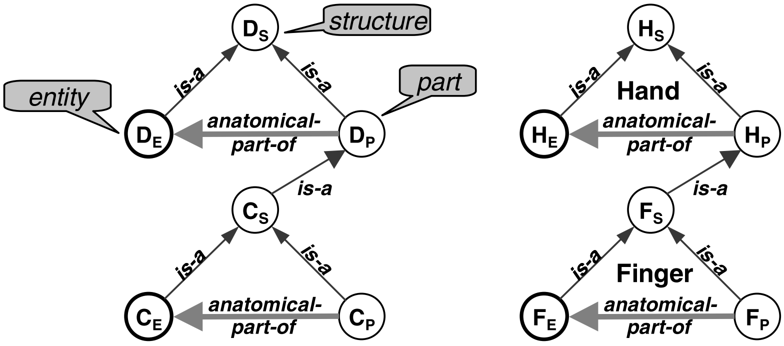


Fig. 1. SEP triplets: partitive relations within taxonomies.

order to emulate partonomic reasoning by taxonomic reasoning. Section 3 contains an in-depth description of a four-step knowledge engineering procedure for semi-automatically converting UMLS specifications into a terminological knowledge base. Throughout this procedure our emphasis is on maintaining the consistency of the emerging knowledge base. We conclude in Section 4 by discussing some implications of our approach and prospects of future work.

2. Part-whole reasoning

As far as medical knowledge is concerned, two main hierarchy-building relationships can be identified, namely taxonomic (is-a) and partonomic (part-whole) ones. Unlike taxonomic reasoning in concept hierarchies, no fully conclusive mechanism exists up to now for reasoning along partonomic hierarchies in description logic systems. As anatomical knowledge, a crucial portion of medical knowledge, is principally organized along part-whole hierarchies, any proper medical knowledge representation has to take account of both hierarchy types [22].

The outstanding importance of part-whole hierarchies for anatomy and, consequently, for clinical medicine has recently motivated the development of semantic networks of anatomical concepts [23,24]. Although they provide ontologically precise descriptions of partonomies, their granularity level is usually rather high. Also, these terminological resources do not provide a formally founded methodology for part-whole reasoning that underlies various object-centered representation approaches as discussed by Artale et al. [25]. In one of these branches, the description logics community, several language extensions for knowledge representation systems have been proposed which provide special constructors for part-whole reasoning [19,26].

Motivated by proposals from Schmolze and Mark [27], as well as by design principles underlying the *Read Codes Version 3* [28], we advocate an alternative solution for part-whole reasoning, one that does not exceed the expressiveness of the well-understood, parsimonious concept language $\mathcal{ALC}$ [29]. Unlike the constructor-based approaches mentioned before, our approach can easily cope with many of the exceptions to the transitivity of the part-of relation, which one encounters not only in medicine [30,31] but also in commonsense domains [32,33].

Instead of defining new operators with a built-in transitivity property, our proposal is centered around a particular data structure, so-called *SEP triplets*, especially designed for empirically adequate part-whole reasoning (cf. the structural description in Fig. 1). They define a characteristic pattern of *is-a* hierarchies, which support the emulation of inferences typical of transitive *part-of* relations, as well as exceptions to it. In this formalism, the relation *anatomical-part-of* describes the partitive relation between physical parts of an organism.

Each basic anatomical concept node is expanded to an *SEP triplet*. Such a triplet consists, first of all, of the anatomical concept itself, the so-called E-node (*entity node*). As an example, in Fig. 1, H_E stands for the concept of the entire *Hand*. The second node of the triplet construct, the P-node (*part node*) is defined as the common subsumer of all concepts which have the role *anatomical-part-of* filled by the corresponding E-node. P-nodes can therefore be considered as a kind of reification of the relation *anatomical-part-of*. In Fig. 1, the P-node H_P subsumes every concept which has H_E (*Hand*) as a filler of the role *anatomical-part-of*, e.g. F_E (*Finger*). Finally, both, the P- and E-node, have a common direct subsumer, the so-called S-node (*structure node*), H_S (*Hand-Structure*) in Fig. 1. By definition, E-nodes and P-nodes are mutually disjoint, thus restricting *anatomical-part-of* to proper parthood, i.e. no anatomical concept can be *anatomical-part-of* itself (e.g. no object in the world can be considered a *Hand* and a *Part of a Hand* simultaneously. This constraint might be relaxed under certain circumstances [34].) The SEP triplet construct can then be used to emulate transitive *part-of* hierarchies by linking S-nodes to P-nodes (cf. the *is-a* link between C_S and D_P in Fig. 1), and to exclude transitivity by linking nontransitive properties to the corresponding E-node of a SEP triplet [30,31]. The solution we propose is computationally neutral insofar as we extend the number of concept nodes by a constant factor (viz. two additional nodes per concept at most).

3. Semi-automatic transformation of an informal knowledge repository into a formal terminological knowledge base

Our goal is to extract conceptual knowledge from two highly relevant subdomains of the UMLS, anatomy and pathology, and to map it (semi-)automatically into a formally sound medical knowledge base. We use LOOM [35,36], a KL-ONE-style terminological knowledge representation language, as our implementation platform (for alternatives, cf. [37]), though our approach does in no way depend on particular features of that language.[2] The knowledge transformation task is divided into four steps: (1) the automatic generation of terminological assertions, (2) their submission to a terminological classifier[3] for consistency checking, (3) the manual restitution of formal consistency in case of inconsistencies, and, finally, (4) the manual rectification and refinement of the resulting knowledge base. These four steps are illustrated by the workflow diagram depicted in Fig. 2.

3.1. Step 1: automatic generation of terminological assertions

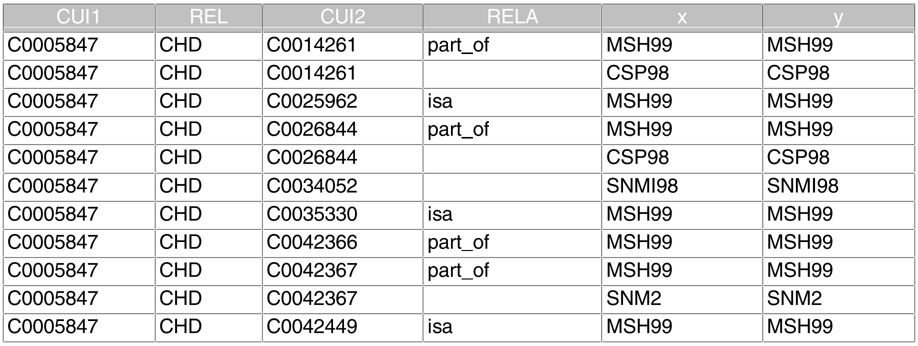
Sources for concepts and relations were the UMLS semantic network and the *mrrel*, *mrcon* and *mrsty* tables of the 1999 release of

[2] Cf. also the work of Carenini and Moore [38] who have already suggested a graphical interactive tool for mapping UMLS concepts semi-automatically into a LOOM knowledge base environment.

[3] The description classifier of a terminological knowledge representation system [36] is the inference engine that computes subsumption relations between concepts, i.e. the generalization hierarchies that can be derived from *is-a* relations.

		Step 1	Step 2	Step 3	Step 4
UMLS relation	number of links	Automatic generation of Loom definitions, augmented by P-Loom language elements *;;; = comment line*	Submission to Loom classifier. Validation for formal consistency by Loom	Manual restitution of formal consistency	Manual rectification and refinement of the resulting knowledge base
Anatomy Concepts Linked to Anatomy Concepts					
sibling_of	267.218	*;;; SIB*			add negations in order to express taxonomic or partitive disjointness
child_of	59.808	*;;; CHDRN*			include related concepts into :is-primitive or :part-of clause where plausible
narrower_term	24.223	*;;; CHDRN*			
isa	9.755	:is-primitive	check for definitional cycles	remove taxonomic parent concepts	substitute of primitive links by non-primitive ones where possible
location_of	4.803	*;;; LOCATION_OF*			include related concepts into :has-part clause where plausible
has_location	4.803	*;;; HAS_LOCATION*			include related concepts into :part-of clause, where plausible
has_part	4.321	:has-part			check whether this part is mandatory (under "real-anatomy" assumption)
has_conceptual_part	126				
part_of	4.321	:part-of	1. check for partonomic cycles 2. check for disjointness between E and P node	1. remove partonomic or taxonomic parent concepts 2. redefine triplet as single concept	check for plausibility and completeness
conceptual_part_of	126				
parent	59.808	*;;; PARRB*			include related concepts into :has-part clause where plausible
broader_term	24.223	*;;; PARRB*			
inverse_isa	9,755	<do nothing>			
associated_with	14				
mapped_from	2.643				
other_relation	10.908				
qualified_by	1.864				
allowed_ qualifier	1.864				
mapped_to	2643				
<other named relations>	11.886	(:some x)	check for inherited constraints	remove constraints	remove or add constraints
Pathology Concepts Linked to Pathology Concepts					
sibling_of	457.542	;;; SIB			add negations in order to express taxonomic disjointness
child_of	72.426	:is-primitive	check for definitional cycles	remove parent concepts	substitute primitive links by non-primitive ones whenever possible
narrower_term	26.972				
isa	3.635				
inverse_isa	3.635	<do nothing>			
associated_with	13.902				
mapped_to	15.024				
mapped_from	15.024				
part_of	1				
has_part	1				
parent	72.426				
broader_term	28.972				
other_relation	25.796				
qualified_by	6.255				
allowed_qualifier	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	(:some has_anatomic_correlate)			plausibility check of concept "duplication" (assignment to both domains)
<missing>		<do nothing>			add pathology-anatomy links
associated_with	2.314	(:some associated_with <anatomy_concept>_S)		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 has_location <anatomy_concept>_S)			
<other>		<do nothing>			

Fig. 2. Workflow diagram for the construction of a terminological knowledge base from the UMLS.



CUI1	REL	CUI2	RELA	x	y
C0005847	CHD	C0014261	part_of	MSH99	MSH99
C0005847	CHD	C0014261		CSP98	CSP98
C0005847	CHD	C0025962	isa	MSH99	MSH99
C0005847	CHD	C0026844	part_of	MSH99	MSH99
C0005847	CHD	C0026844		CSP98	CSP98
C0005847	CHD	C0034052		SNMI98	SNMI98
C0005847	CHD	C0035330	isa	MSH99	MSH99
C0005847	CHD	C0042366	part_of	MSH99	MSH99
C0005847	CHD	C0042367	part_of	MSH99	MSH99
C0005847	CHD	C0042367		SNM2	SNM2
C0005847	CHD	C0042449	isa	MSH99	MSH99

Fig. 3. Semantic relations in the UMLS metathesaurus.

the UMLS metathesaurus. The *mrrel* table contains roughly 7.5 million records and exhibits the semantic links between two concept unique identifiers (CUIs)[4], the *mrcon* table contains the concept names and *mrsty* keeps the semantic type(s) assigned to each CUI. These tables (cf. Fig. 3 for a fragment), 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 alphanumeric code was substituted by the English preferred term given in *mrcon*.

From a total of 85 899 concepts, we extracted 38 059 anatomy and 50 087 pathology concepts from the metathesaurus. Each concept was included in this set, which belonged to a set of predefined anatomy[5] and pathology[6] types given in the UMLS semantic network. 2247 concepts were included in both sets, anatomy and pathology. This finding can easily be justified by the observation that these hybrid concepts exhibit, indeed, multiple meanings.[7] As we wanted to keep

Table 1
A triplet in extended LOOM format

(deftriplet HEART
:is-primitive HOLLOW-VISCUS
:has-part (:p-and
ANATOMICAL-FEATURE-OF-HEART
FIBROUS-SKELETON-OF-HEART
WALL-OF-HEART
CAVITY-OF-HEART
CARDIAC-CHAMBER-NOS
LEFT-CORONARY-SULCUS
RIGHT-CORONARY-SULCUS
SURFACE-OF-HEART-NOS
LEFT-SIDE-OF-HEART
RIGHT-SIDE-OF-HEART
AORTIC-VALVE
TRICUSPID-VALVE
PULMONARY-VALVE
MITRAL-VALVE
HEART-VALVES-100))

[4] As a coding convention in UMLS, any two CUIs must be connected by at least a shallow relation (in Fig. 3, CHilD relations in the column REL are assumed between CUIs). These 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 (MSH99), SNOMED International 1998 (SNMI98).

[5] *Anatomical Structure*, *Embryonic Structure*, *Congenital Abnormality*, *Acquired Abnormality*, *Fully Formed Anatomical Structure*, *Body System*, *Body Part Organ* or *Organ Component*, *Tissue*, *Cell*, *Cell Component*, *Gene* or *Genome*, *Body Location* or *Region*, *Body Space* or *Junction*, *Anatomical Abnormality*.

[6] *Pathologic Function*, *Disease* or *Syndrome*, *Mental* or *Behavioral Dysfunction*, *Cellular* or *Molecular Dysfunction*, *Experimental Model of Disease*, *Neoplastic Process*.

[7] For instance, *Tumor* has the meaning of a malignant disease on the one hand, and of an anatomical structure on the other hand. The same applies to congenital and acquired malformations, e.g. *Claw Foot*.

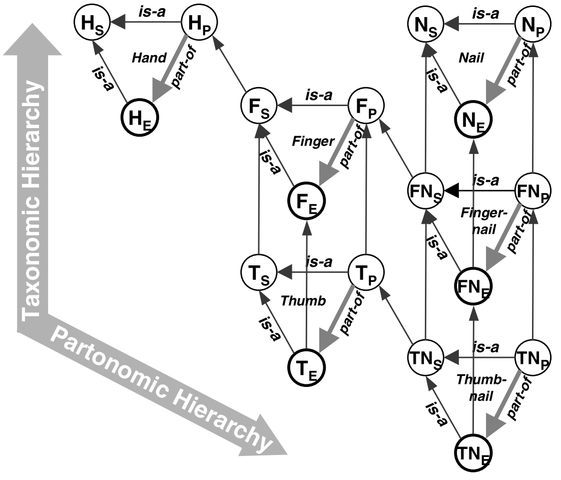


Fig. 4. A mixed *is-a* and *part-of* hierarchy.

the two subdomains strictly disjoint, we duplicated these hybrids and prefixed them with '*ana*-' or '*pat*-' according to their respective subdomain. The a priori assignment to the above-mentioned semantic types in the UMLS is the only selection criterion; we refrained from any manual interference at this processing stage.

Anatomy and pathology concepts received a different formal treatment, however. As target structures for the anatomy domain we chose SEP triplets. These were expressed in the terminological language LOOM, which we had previously extended by a special DEFTRIPLET macro (cf. Table 1 for an example).[8] Only *part-of*, *has-part* and *is-a* relation attributes (RELA fields in the *mrrel* table) from the UMLS were considered for the construction of taxonomic and partonomic hierarchies (cf. Fig. 2). Hence, for each anatomy concept, one SEP triplet is created. The result is a mixed *is-a*/*part-whole* hierarchy (cf. Fig. 4).

For the pathology domain, we assumed the values *CHD* (child), *ISA* and *RN* (narrower relation) from the *mrrel* REL field as indicators of taxonomic links. For all anatomy concepts referred to in the definitional statements 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 (see below).

In both subdomains, shallow relations, such as the extremely frequent *SIB* (sibling) relation, were included as comments into the code to give some heuristic guidance for the manual refinement phase (cf. Fig. 2).

3.2. Step 2: consistency checking by the description classifier

The import of UMLS anatomy concepts resulted in 38 059 DEFTRIPLET expressions for anatomical concepts and 50 087 DEFCONCEPT expressions for pathological concepts. Each DEFTRIPLET was expanded into three DEFCONCEPT (S-, E-, and P-nodes), and two DEFRELATION (*anatomical-part-of-x*, *inv-anatomical-part-of-x*) expressions, summing up to 114 177 concepts and 76 118 relations. Thus we obtained (together with 382 concepts from the semantic network) a total of 240 764 definitory LOOM expressions.

From 38 059 anatomy triplets, 1219 DEFTRIPLET statements exhibited a *:has-part* clause followed by a list of a variable number of triplets, containing more than one argument in 823 cases (average cardinality: 3.3). 4043 DEFTRIPLET statements contained a *:part-of* clause, only in 332 cases followed by more than one argument (average cardinality:

[8] The UMLS anatomy concepts are mapped an intermediate language, P-LOOM, the reason being that the manual refinement of automatically generated LOOM triplets is time-consuming and too error-prone due to their complex internal structure. P-LOOM provides the full expressiveness of LOOM, enriched by special constructors for the encoding of the part-whole relations, as well as for direct manipulation of the triplet elements, whenever necessary. The main feature of P-LOOM, the macro DEFTRIPLET, shares the syntax of the concept-forming LOOM constructor DEFCONCEPT, augmented by the keywords *:part-of* and *:has-part* (both are followed by a list of SEP triplets).

1.1). The obtained knowledge base was then submitted to the terminological classifier and automatically checked for terminological cycles and consistency. A terminological cycle is given when *A* subsumes *B* and *A* is subsumed by *B*, as well. Inconsistencies occur when constraints (e.g. role restrictions) are violated. In the anatomy subdomain, one terminological cycle and 2328 inconsistent concept definitions were identified; in the pathology subdomain 355 terminological cycles were determined though no inconsistent concept definition at all was found (cf. Table 2).

3.3. Step 3: manual restitution of consistency

The inconsistencies of the anatomy part of the knowledge base identified by the classifier could be traced back to the simultaneous linkage of two triplets by both *is-a* and *part-of* links, an encoding that raises a conflict due to the disjointness required for corresponding P- and E-nodes. In most of these cases the affected parents belong to a class of concepts that obviously cannot be appropriately modeled as SEP triplets, e.g. *Subdivision-Of-Ascending-Aorta*, *Organ-Part*. The meaning of these concepts almost paraphrases that of a P-node, so that in these cases 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 P-nodes, by relaxing the disjointness constraint, or by disabling *is-a* or *part-of* links.

Table 2
Classification results for anatomy and pathology concepts

	Anatomy	Pathology
Triplets	38 059	–
DEFCONCEPT statements	114 177	50 087
Cycles	1	355
Inconsistencies	2328	0

In the pathology part of the knowledge base, we expected a large number of terminological cycles to arise as a consequence of interpreting the notoriously weak, thesaurus-style *RN* (narrower) and *CHilD* relations through taxonomic subsumption (*is-a*). Bearing in mind the size of the knowledge base, we consider 355 cycles a tolerable amount of noise. Those cycles were primarily due to very similar concepts, e.g. *Arteriosclerosis vs. Atherosclerosis*, *Amaurosis vs. Blindness*, and residual categories (‘other’, ‘NOS’ = *not otherwise specified*). These were directly inherited from the source terminologies and are always difficult to interpret out of their definitional context, e.g. *Other-Malignant-Neoplasm-of-Skin vs. 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. By adding these new constraints a fully consistent knowledge base was generated.

3.4. Step 4: manual rectification and refinement of the knowledge base

Adding value to a consistent though possibly underspecified or even misspecified knowledge base is an extremely time-consuming job and requires broad and in-depth medical expertise. In order to roughly assess the potential workload for future knowledge base finishing, we extracted two random samples ($n = 100$ each) from both the anatomy and pathology part of the knowledge base; the samples were then analyzed by a medical student and a physician. From the experience we gained in both subdomains so far, the following workflow can be derived:

3.4.1. Checking the correctness and completeness of both the taxonomic and partitive hierarchies

Taxonomic and partitive links are manually added or removed in order to eliminate inadequate concept descriptions and to increase the completeness and to deepen the granularity of concept descriptions. Primitive subsumption (where necessary conditions for a specialization relation between concepts are specified only) is substituted by a nonprimitive one (where necessary and sufficient conditions for a specialization relation between concepts are specified) whenever possible. This is a crucial point, because the automatically generated hierarchies contain only information about the parent concepts and necessary conditions. 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 be classified automatically as *Dermatitis*.

3.4.2. 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. Another 83 UMLS relations (mostly *CHilD* or *RN* (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 *RN* (narrower) relations) were manually upgraded to *part-of* relations, and 94 UMLS relations (mostly *PARRB*, i.e. parent and 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, while 24 *is-a* relations were considered to be missing.

3.4.3. Checking :has-part arguments assuming 'real anatomy'

In the UMLS sources *part-of* and *has-part* are considered symmetric. According to our transformation rules, the attachment of a role *has-anatomical-part* to an E-node B_E, with its range restricted to A_E implies the existence of a concept A_E for the definition of concept B_B. On the other hand, the classification of A_E as being subsumed by the P-node B_P, the latter being defined via the role *anatomical-part-of* restricted to B_E, implies the existence of B_E given the existence of A_E (cf. Fig. 5, left). This assumption does not always match 'real' anatomy, i.e. anatomical concepts that may exhibit pathological modifications. Fig. 5 (left part) sketches a concept $\mathrm{A_E}$ that is necessarily *anatomical-part-of* a concept $\mathrm{B_E}$, but whose existence is not required for the definition of $\mathrm{B_E}$. This is typical of the results of surgical interventions, e.g. a large intestine without an appendix, or an oral cavity without teeth, etc.

3.4.4. 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

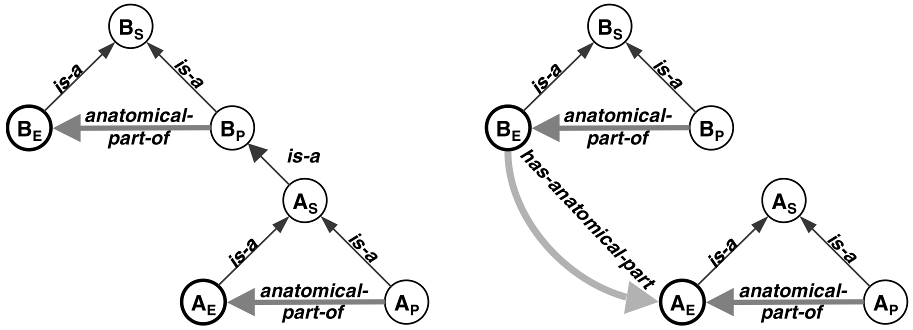


Fig. 5. Patterns for partonomic reasoning using SEP triplets: *anatomical-part-of* without *has-anatomical-part* (left), *has-anatomical-part* without *anatomical-part-of* (right).

a coherent classification of pathologically modified anatomical objects.[9] 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 a P-node and a S-node (cf. Fig. 5, left part). The non-linkage situation is also possible (cf. Fig. 5, right part). The definition of A_E does not imply the role *anatomical-part-of* to 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 characteristic of mereological relations between macroscopic (countable) objects, such as organs, and multiple uniform microscopic objects [34].

3.4.5. Analysis of the sibling relations and defining concepts as being disjoint

In the UMLS *mrrel* table, the *SIB(ling)* relation targets at concepts which share the same parent in a taxonomic or partonomic hierarchy. Pairs of sibling concepts may either 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 benignant tumor cannot be a malignant one, et vice versa). In a partitive hierarchy, this corresponds to two topologically disconnected objects, C_E and D_E, with the following interpretation: There are no common parts shared by any instance of C_E with any instance of D_E. In our triplet formalism this can be expressed as follows (for a formalization and further discussion of topological aspects in anatomical ontologies, cf. [39,40]): topological disconnectedness refers to a pair of concepts, C_E and D_E, whose S-nodes, C_E and D_E, belong to two disjoint subgraphs (i.e. C_S implies the negation of D_S). As a consequence there is no instance that is both *anatomical-part-of* an instance of C_E and *anatomical-part-of* of an instance of D_E, (cf. Fig. 6). As an example, the concepts *Right Hand* and *Left Hand* are topologically disconnected, whereas *Right Hand* and *Right Forearm* are not (there are instances which share a common boundary structure).

[9] In Table 1, the concepts marked by *italics*, viz. *Aortic-valve*, *Tricuspid-valve*, *Pulmonary-valve* and *Mitral-valve* should all 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.

3.4.6. Results

We found, on average, 6.8 siblings per concept in the anatomy domain, and 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.

3.4.7. Completion and modification of anatomy–pathology relations

For each pathology concept (such as determined by the LOOM system after classification) it has to be checked whether the anatomy–pathology links are correct and complete. Incorrect constraints have to be removed from a concept definition itself or from one of the subsuming concepts. For each correct anatomy–pathology relation the decision must be taken whether the E-node or the S-node has to be addressed as the target concept for modification. In the first case, the propagation of roles across part-whole hierarchies is disabled. As an example (cf. Fig. 7), *Enteritis* implies *has-location Intestine*. The range of the relation *has-location* is restricted to the E-node of *Intestine*, I_E. This precludes, for instance, the computation of an *is-a* relation between *Appendicitis* and *Enteritis*, though *Appendix* is related to *Intestine* via an *anatomical-part-of* relation. In the second case, the target is the S-node of the anatomical triplet, and, thus, the propagation of roles is enabled. *Glomerulonephritis* (*has-location Glomerulum*) is therefore subsumed by *Nephritis* (*has-location Kidney*), since *Glomerulum* is defined as an *anatomical-part-of Kidney*. In the same way, *Perforation-of-Appendix* is generalized as *Intestinal-Perforation* (cf. [30,31] for a comprehensive analysis and formal specification of these phenomena).

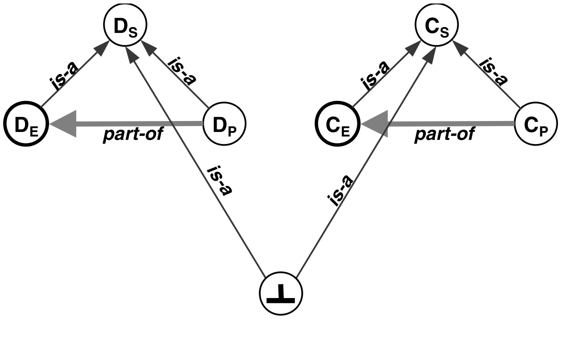


Fig. 6. Triplet representation for topologically disconnected concepts.

3.4.8. Results

In our random sample we found 522 anatomy–pathology relations, from which 358 (i.e. 69%!) were judged as 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 random sample of 100 pathology concepts revealed that only 17 of them 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 a given 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 definitions 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

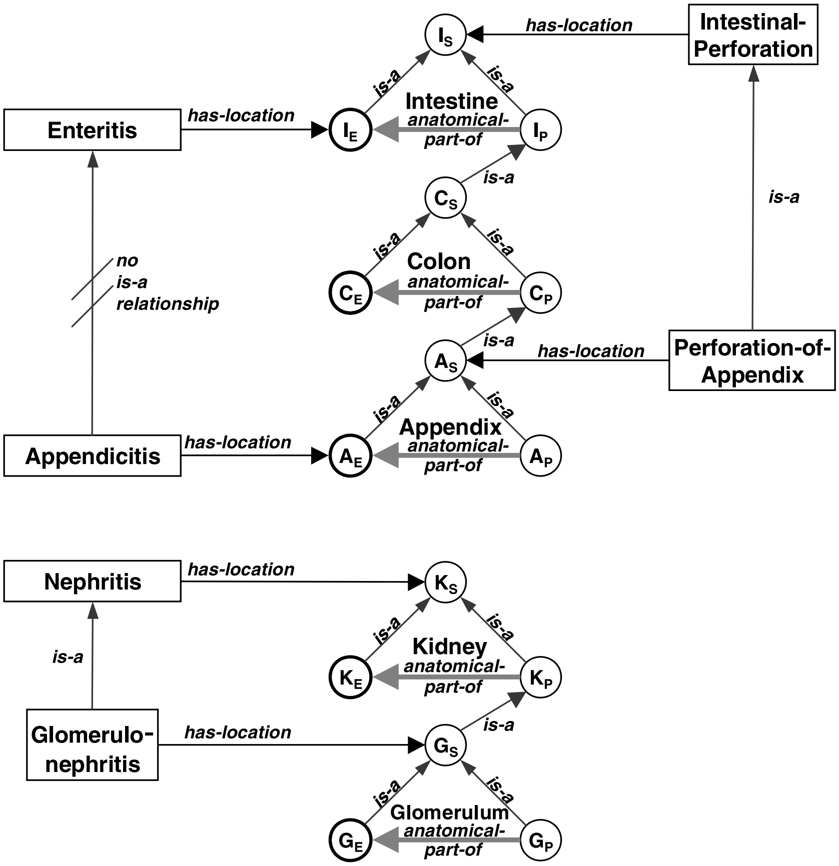


Fig. 7. Alternative linkages of a pathology concept: either to the S-node or to the E-node of an anatomical triplet in order to enable or preclude computation of *is-a* relations, respectively.

constraints (e.g. *has-location Urinary-Tract*) are inherited to subconcepts such as *Tuberculosis-of-Bronchus*. Hence, 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. Conclusions

There is a growing demand for high-quality terminology services and their embedding in functionally advanced health information systems. Among the desiderata that have to be fulfilled is the need to make consistent, conceptually rich knowledge bases available so that their inference engines can derive valid results. While there is a long tradition of developing medical knowledge bases from scratch, we here propose a conservative approach—reuse existing large-scale repositories, but refine the data from these resources so that advanced requirements imposed by more expressive knowledge representation languages are met. Consistency checking comes almost for free, once the informal knowledge sources are embedded in a formal reasoning framework (cf., e.g. the work of Mejino and Rosse who recognized inconsistencies in the UMLS based on formal representation structures in the Digital Anatomist model [41]). The resulting knowledge bases can then be used for sophisticated applications requiring sound medical reasoning.

The knowledge engineering approach we have proposed in this paper does exactly this. It provides a formally solid description logics framework with a modeling extension which supports not only taxonomic reasoning, but also incorporates partonomic reasoning adapted to the requirements of anatomy as the foundation of medical terminology. In spite of their evident weaknesses, the subsets of the UMLS we analyzed proved to be useful as a source of terminological knowledge on a large scale. Whereas the restitution of logical consistency could be achieved in a straightforward way, the cleansing of the resulting knowledge base from inadequate concept definitions and specification gaps implies a high degree of manual involvement, which requires enormous efforts when it has to be performed on the knowledge base as a whole. A realistic setting would be to eliminate inadequacies once and for all, but to remedy specification gaps only when required by concrete applications.

For anatomy and pathology, the domains under analysis, this study sheds light on the conditioned usability of the conceptual ‘raw material’ the UMLS metathesaurus provides for knowledge engineering. For macroscopic anatomy the existing resources proved fruitful due to the inclusion of the UWDA (University of Washington Digital Anatomist, cf. [42]) knowledge base which delivers semantically precise relationships. Severe weaknesses and underspecification arise in the pathology portion where the necessary linkage to anatomy proved to be entirely insufficient.

While plain automatic conversion from semi-formal to formal environments causes problems of adequacy of the emerging representation structures, the step-wise refinement methodology we propose already inherits its power from the terminological reasoning framework. In our concrete work, we found the implications of using the terminological classifier, the inference engine which computes subsumption relations, of utmost importance and of outstanding heuristic value. Hence, the knowledge refinement cycles are truly semi-automatic, fed by medical expertise on the side of the human knowledge engineer, but also driven by the reasoning system which makes explicit the consequences of (im)proper concept definitions.

Acknowledgements

Stefan Schulz was supported by a grant from DFG (Ha 2097/5-2).

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