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Ontology driven semantic profiling and retrieval in medical information systems

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ABSTRACT

We propose the application of a novel sub-ontology extraction methodology for achieving interoperability and improving the semantic validity of information retrieval in the medical information systems (MIS) domain. The system offers advanced profiling of a user's field of specialization by exploiting the concept of sub-ontology extraction, i.e., each sub-ontology may subsequently represent a particular user profile. Semantic profiling of a user's field of specialization or interest is necessary functionality in any medical domain information retrieval system; this is because the (structural and semantic) extent of information sources is massive and individual users are only likely to be interested in specific parts of the overall knowledge documents on the basis of their area of specialization. The prototypical system, OntoMOVE, has been specifically designed for application in the medical information systems domain. OntoMOVE utilizes semantic web standards like RDF(S) and OWL in addition to medical domain standards and vocabularies encompassed by the UMLS knowledge sources.

1. Introduction

The new era of e-health information systems has introduced a number of research and development issues whereby the efficient *i* ntegration of various health information domains is needed and timely retrieval and access to best practice information resources is vital. In general, underlying these issues are the closely related themes of *interoperability* of medical information sources and the efficient *retrieval* of semantically correct data from them [1,27]. In the last decade or so, the application of ontologies in information systems as a shared platform for information integration and for establishing a consensus on meaning has been promoted [19]. Grounded in the fundamental problems of integration and retrieval, applications of ontologies in the medical information systems (MIS) domain have been aplenty [10,14]. A comprehensive review of ontological applications in MIS is beyond the scope of this paper. As specific references, see [2] for a work on constructing medical domain terminological systems and [38] for a review of biomedical domain specific ontologies.

1.1. Ontologies on the MOVE

In this paper, we present our system OntoMOVE –'Ontologies on the MOVE' – that combines the use of ontology driven annotations and the application of a novel sub-ontology extraction methodology (called MOVE) for achieving interoperability and improving the effectiveness of information retrieval for the specific MIS domain. The main difference between our work and existing work in ontology-based information retrieval is the fact that our approach begins with the process of extracting a sub-ontology that meets the user requirements, which is then followed by the Contextualization of the sub-ontology through annotating existing documents or resources to the specified sub-ontology. One of the most significant objectives of our approach is to reduce the search space of information retrieval by establishing a semantic scope through the sub-ontology Contextualization and profiling. Our ontology is based on the Unified Medical Language System (UMLS) knowledge sources, namely the UMLS Semantic Network (UMLS-SN) and UMLS Metathesaurus®. As such, the approach is compatible with controlled vocabularies and classifications used in patient records, administrative health data, bibliographic and full-text databases that are based on the UMLS initiative [11,23,48]. Although we restrict ourselves to a specific type of resource in this paper, namely information documents contained in the Medical Therapeutic Guidelines (TG) [45], it should be noted that the precise nature of the resources being retrieved is of no relevance to the

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system (see Section 5.1). For instance, resources could even consist of patient records or other forms of health or operational data generated over a period of time. As long as annotational requirements (Section 5.3) of the framework are met, any type of resources repository can be integrated within the system presented in this paper.

1.2. Ontology, semantics and MOVE

The use of an ontology is intended to produce semantically correct results whilst retrieving information from knowledge sources consisting of large repositories of medical resources. The retrieval phase of the OntoMOVE system is based on the specialization of a general Materialised Ontology View Extraction (MOVE) framework for the medical domain exemplar presented in this paper. MOVE is our system for deriving semantically correct and independent sub-ontologies from potentially large base ontologies [6,8,9,50]. We exploit the general capability of the MOVE framework for deriving sub-ontologies toward operationalizing the concept of '*semantic profiling*' (Sections 4 and 6) of user's requirements based on the field of specialization and/or interest. The concept of semantic profiling is based on the premise that different users/customers/stakeholders of medical data have different informational requirements. This is especially important, keeping in mind the structural and semantic extent of most realistic medical domain information sources, such as the medical TG. Indeed, an individual user is not likely to be interested in every conceivable category of medical data that is present in the information sources or repositories. For instance, keeping in line with the medical domain application scenario discussed in this paper, consider the case of retrieving information from the medical therapeutic guidelines. Here, potential users include researchers, medical practitioners and even patients. In this scenario, a pharmacist may be more interested in drug and treatment related information whereas a biomedical scientist may be more interested in information about the latest findings related to certain cell malfunctions or new gene products. To serve user-specific (e.g., pharmacist, medical practitioner, biomedical scientist and so forth) information requirements, semantic profiling of their requirements is therefore a necessary functionality in any medical information retrieval system.

1.3. Organization

The rest of the paper is organised as follows: Section 2 consists of a review of work related to the use of ontologies in medical information systems. Section 3 presents a brief overview on the use of ontological formalism in the medical domain. The use of the OWL as a medium for information representation for UMLSKS content is introduced and the representational capability of OWL is highlighted. In Section 4, we illustrate the concept of sub-ontology extraction using the MOVE framework and also explain the intuition that underlies the use of sub-ontologies as semantic requirement profiles. In Section 5, all aspects of the OntoMOVE framework are presented in detailed and the framework is experimentally evaluated in Section 8. Finally, we conclude in Section 9 with a brief discussion of our approach and pointers to the future directions of this work.

2. Related work

A recent study conducted by Gartner, Inc. [17] indicates that an integrated 'Semantic Web' will be one of the highest impact emerging technologies in the next five to ten years and that many public domain industries have started to engage in this technology. Ontologies are utilized as a foundation to enable interoperability within the Semantic Web, and as a result the number of domain ontologies have grown significantly in the last few years. In the medical domain, researchers and health care standard bodies have also started to introduce semantic standard encoding for their clinical data specification [10]. It is crucial that medical information systems are built with an underpinning technology that supports global information interactions and management, and the various works in ontology have played an important role in this area.

The past few years have witnessed a range of applications and studies in the area of ontology management and processing. From the viewpoint of this paper, we categorise existing research and/or systems into the following groups: (a) foundational work encompassing topics such as ontology evolution, ontology editing and alignment, ontology merging, etc. (b) applications of ontologies in diverse domains, (c) ontological tools that provide general support within arbitrary domains of interest, and (d) specialized information retrieval systems in the medical/biomedical domain that utilize ontologies in some way. Here, we selectively describe some of the existing works from these categories and outline how our approach differs from that of existing systems.

2.1. Ontology evolution

Research in this area encompasses foundational work that focus on the ontology manipulation and tailoring techniques with an ultimate goal of ontology reuse and integration. The work by Maedche et al. [29] covers the area of ontology reuse and evolution in the context of ontology management within a distributed system environment. Their proposed method allows the creation of a new ontology by reusing an existing ontology, whilst taking into consideration ontology evolution and integration given the fact that the created ontologies are distributed on many different sites. Similar work by the same authors in [30] also describes the notion of 'ontology registration' in order to provide means to locate existing ontologies for reuse. Whilst this work has addressed many important issues in a distributed ontology environment, there are a few areas that have not been fully addressed. Firstly, the proposed method lacks a proper technique to optimize the created (reused) ontologies—although an algorithm to check the validity of the ontology is proposed, the method does not include a mechanism to derive the most optimum ontology. Note that optimality involves several criteria revolving around the size of the resulting ontology, e.g., semantic simplicity and/or the minimization of redundant content in the resulting ontology. Secondly, the proposed technique only focuses on the extraction of a new ontology from an existing ontology, without consideration for retrieving other artefacts or resources that might be linked or annotated against the existing ontology—any refinement of the structure or extent of the ontology bears a direct relation to its semantic scope within whatever resources have been described using that ontology.

2.2. Ontological applications

The second category of existing works in the ontology area include a variety of ontology-based domain specific applications. In particular, there have been numerous research and developments in the area of medical ontologies as mentioned in Section 1. Most of these works can be categorised into: utilizing medical ontologies to build a knowledge repository such as [39,46], consolidating and merging biomedical ontologies such as [4,26,28], integration of medical terminologies [16], and ontology based collaborative work in medical domain such as [39]. All these existing applications are focussing on utilizing a domain ontology (in this case a medical ontology) to support collaboration and communication between resources, domain experts, etc., in the area. As opposed to utilizing the whole large ontology or engineering a new medical ontology [12], our work focusses on extracting and optimizing a sub-ontology

from a given large domain ontology that fulfills a user requirement profile.

2.3. Ontology engineering tools

It is important to differentiate the system proposed here with existing tools for ontology editing and alignment such as Protégé [33,40], or OntoEDIT [34]. Whilst the above tools provide efficient techniques to create, view, visualize, edit and align ontologies, they do not particularly address the issues of (i) user-driven automatic extraction of valid sub-ontologies, and (ii) semantic and structural optimization of the resulting ontologies. In addition, these tools do not address application level annotation and semantic mapping.

2.4. Specialized information retrieval systems

There are many different information retrieval and browsing systems from a specialized medical/bio-medical viewpoint. Principal among them include Textpresso, GoPubMed and XplorMed. Textpresso is an information retrieval and extraction system that processes the full-text of biological papers [31]. The system tokenizes text into several categories with respect to an ontology. GoPubMed is a information retrieval engine that presents PubMed results in an ontology-based hierarchical form [15]. The focus here is on presenting a dynamic taxonomical view for user's queries based on ontologies. Similarly, XplorMed is an exploratory tool that is aimed at overcoming the limitations of keyword based search [37]. The contributions of our research are foundational and are, in principle, aimed at leveraging upon the utility provided these or other specialized information retrieval algorithms and tools: we focus on the semantic profiling of a user's requirements prior to performing a query so that an underlying information retrieval system has a better approximation on the scope of the user's interest within a resource or document repository.

All of the above-mentioned works highlight the increase of interest to utilize ontology as a means to standardize processes or tasks. However, despite these recent efforts, there has been no real focus on tailoring the ontologies to meet user-specific needs as well as to integrate them with the extraction of ontology-annotated data sets or resources. Our proposed techniques will play an important role in applying the notion of reuse in ontology engineering.

3. Ontological representation for the medical domain

Ontologies play a pivotal role by providing a source of shared and precisely defined terms that can be used as meta-data, e.g., annotation of information-sources and other resources in order to make them accessible to automated agents. Although there are inherent distinctions between a taxonomy and an ontology, ontologies as typically used on the semantic web and software engineering applications consist of a hierarchical description of important concepts in a domain, along with descriptions of the properties of each concept. The degree of formality employed in capturing these descriptions can be quite variable [32], ranging from natural language to logical formalisms, but increased formality and regularity clearly facilitates machine understanding [21,44].

3.1. OWL—a formal knowledge representation structure

The Web Ontology Language (OWL) is a knowledge representing scheme designed specifically for use on the semantic web; it exploits existing web standards (XML and RDF), adding the familiar ontological primitives of object and frame based systems, and the formal rigor of a very expressive description logic (DL) that emerges from research in the field of Artificial Intelligence [22]. As exemplified in Tables 1 and 2, OWL consists a rich set of knowledge representation constructs that can be used to formally specify medical-domain knowledge, which in turn can be exploited by description logic reasoners for purposes of inferencing, i.e., deductively inferring new facts from knowledge that is explicitly available. The knowledge base (KB) of a typical DL based system comprises of two components, the TBOX and the ABOX. The TBox introduces the terminology, i.e., the vocabulary of an application domain (e.g., '*Neoplastic Process is a Biological Function*'), whilst the ABox contains assertions about named individuals in terms of this vocabulary ('*Cancer is an instance of a Neoplastic Process*'). The logical basis of the language means that reasoning services can be provided in order to make OWL described resources more accessible to automated processes thereby allowing one to infer implicitly represented knowledge from the knowledge that is explicitly contained in the knowledge base. From a formal point of view, OWL can be seen to be equivalent to a very expressive DL, with a OWL ontology corresponding to a DL terminology (TBox) whereas instance data pertaining to the ontology making up the assertions (ABox).

Table 1
OWL axioms.

Axiom	DL syntax	Example
Sub class	$C_1 \sqsubseteq C_2$	Alga $\sqsubseteq$ Plant $\sqsubseteq$ Organism
Equivalent class	$C_1 \equiv C_2$	Cancer $\equiv$ Neoplastic Process
Disjoint with	$C_1 \sqsubseteq \neg C_2$	Vertebrate $\sqsubseteq \neg$Invertebrate
Same individual	$x_1 \equiv x_2$	Blue_Shark $\equiv$ Prionace_Glauca
Different from	$x_1 \sqsubseteq \neg x_2$	Sea Horse $\sqsubseteq \neg$Horse
Sub property	$P_1 \sqsubseteq P_2$	has_mother $\sqsubseteq$ has_parent
Equivalent property	$P_1 \equiv P_2$	treated_by $\equiv$ cured_by
Inverse	$P_1 \equiv P_2^-$	location_of $\equiv$ has_location$^-$
Transitive property	$P^+ \sqsubseteq P$	part_of$^+ \sqsubseteq$ part_of
Functional property	$\top \sqsubseteq \leq 1P$	$\top \sqsubseteq \leq$ 1has_tributary
Inverse functional property	$\top \sqsubseteq \leq 1P^-$	$\top \sqsubseteq \leq$ 1has_scientific_name$^-$

Table 2
OWL class constructors.

Constructor	DL syntax	Example
Intersection	$C_1 \sqcap \ldots \sqcap C_n$	Anatomical_Abnormality $\sqcap$ Pathological_Function
Union	$C_1 \sqcup \ldots \sqcup C_n$	Body_Substance $\sqcup$ Organic_Chemical
Complement	$\neg C$	$\neg$Invertebrate
One of	$x_1 \sqcup \ldots \sqcup x_n$	Oestrogen $\sqcup$ Progesterone
All values from	$\forall P.C$	$\forall$co_occurs_with.Plant
Some values	$\exists P.C$	$\exists$co_occurs_with.Animal
Max cardinality	$\leq nP$	$\leq$ 1has_ingredient
Min cardinality	$\geq nP$	$\geq$ 2has_ingredient

Our use of the OWL language to represent the medical ontology is driven by the fact that OWL is industry standard and is recommended by the W3C [49] for the representation of ontologies. Furthermore, numerous semantic web tools, for example, Protégé [18,40] and its associated OWL Plugin [41], OntoMat [35], etc., supporting OWL have been already developed in the open-source community. In addition, tool builders have developed powerful reasoning systems that support reasoning with ontologies represented in the OWL language, the best example here being RACER [42]. As a part of further extensions to the work presented in this research (Section 9), we envisage to apply formal description logic based reasoning functionality supported by such tools in the medical domain.

3.2. UMLS Knowledge Source Server

The Unified Medical Language System Knowledge Source Server (UMLSKS) has been used in this work to gain access to the vast amount of knowledge contained in the UMLS by way of its two main components, viz—the UMLS Metathesaurus® and the UMLS

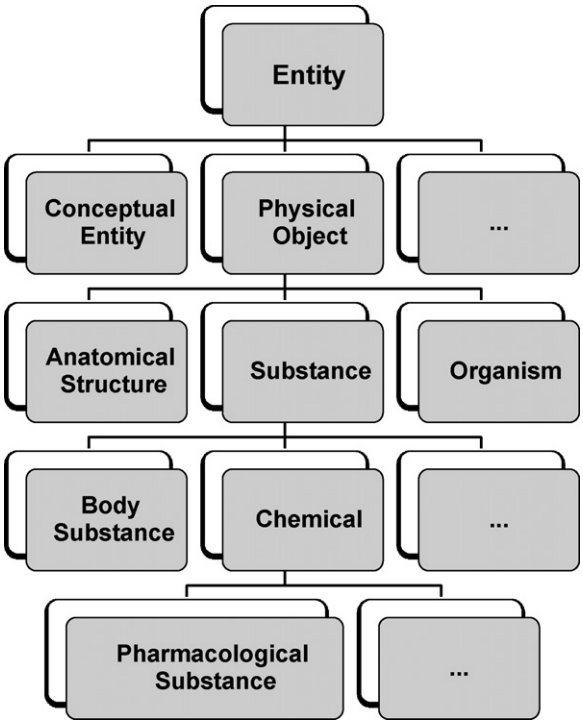


Fig. 1. UMLS semantic type—entity.

Semantic Network. Fig. 1 is a rather narrow view of the semantic network and is indicative of the sort of semantic types present in it. The information provided by the UMLSKS is accessed through a *UMLS Plugin* [47] that can be integrated with the Protégé ontology engineering environment [40]. This approach is useful and time-saving for our project since the plugin facilitates easy browsing of the UMLS Semantic Network and Metathesaurus in addition to supporting a mechanism whereby the knowledge contained therein may be easily integrated into our ontology. The UMLSKS allows a user/application to request information about particular Metathesaurus concepts, including attributes such as the concept's definition, its semantic types, concepts that are related to it, hierarchical context details, etc., all of which can be restricted to source specific details.

4. Sub-ontology view extraction for semantic profiling

As information on the web increases significantly in size, Web Ontologies also tend to grow bigger to such an extent that they become too large to be used in their entirety by any single application (e.g., the UMLS base ontology [48]). Also, because of the size of the original ontology, the process of repeatedly iterating the enormous number of nodes and relationships to derive a sub-ontology becomes very computationally extensive, thereby necessitating the use of parallel and/or distributed techniques for the extraction of the same from a massively sized base ontology. These problems have stimulated our work in the area of sub-ontology extraction in a general context and its optimization using a distributed architecture.

4.1. Materialized ontology view extraction

The extraction process, referred to as Materialised Ontology View Extraction (MOVE) [7,8,50], is capable of deriving sub-ontologies, also referred to as materialised views, from a (typically large) base ontology. More important in the context of this paper, it should be noted that the resulting ontology is semantically complete [6] and a valid ontology independent of the base ontology. This is achieved in MOVE through the enforcement of relevant constraints that take the form of various 'optimization schemes' such as requirements consistency, semantic completeness, well-formedness, and total simplicity, which ensure the correctness and the optimality of the resulting sub-ontology. Broadly, the following four categories of optimization schemes (OS1–OS4) are applied for deriving a independent sub-ontology from a base ontology:

OS1. Requirements consistency: before a sub-ontology is derived, a user or application must provide an approximate indication of its interests in the form of a '*labelling*' (Section 6) of conceptual categories in the base ontology. Requirements consistency checks for the consistency of these user specified labelling/requirements for a specific sub-ontology derivation process. Henceforth, we refer to this labelling as the *r* equirements specification.
OS2. Semantic completeness: semantic completeness considers the completeness of the concepts, e.g., if one concept is defined in terms of an another concept, the latter cannot be omitted from the sub-ontology without loss of semantic meaning of the former concept.
OS3. Well formedness: it might be possible that the user requirements (labelling) is consistent, but there might be statements that inevitably lead to a solution that is not a valid ontology. Well-formedness contains the proper rules to prevent this from happening.
OS4. Total simplicity of solution: finally, applying total-simplicity to an existing solution (along with its requirements specification) will result in the smallest possible solution that is still a valid ontology. Total-simplicity achieves this by working on not only the solution, but also its requirements specification.

An elaboration of these optimization schemes in (OS1–OS4) and the overall extraction process of Fig. 2 is described in our earlier work [7,8,50]. However, what is relevant in the present context is the manner in which the specialization and application of MOVE for deriving sub-ontologies may be utilized as a foundational basis of user-specific semantic requirement profiling. An overview is presented in (P1–P2):
P1. The extraction process: Fig. 2 shows a schematic of the sequential extraction process. The process begins with the import of the ontology represented in the OWL language. The actual extraction process/execution of optimization schemes is initiated by way of *requirements specification* by a user or another application and the execution of the other optimization schemes. The externally provided *requirements specification* is used as the basis of the derivation/extraction process. The derived sub-ontologies are valid independent ontologies, known as Materialized Ontologies, that are specifically extracted to meet certain user/application needs. The result of the extraction process is not just simply an extracted sub-ontology, but rather an extracted *materialized ontology view*. In the extraction process, no new information is introduced (e.g. adding a new concept). However, it is possible that existing seman-

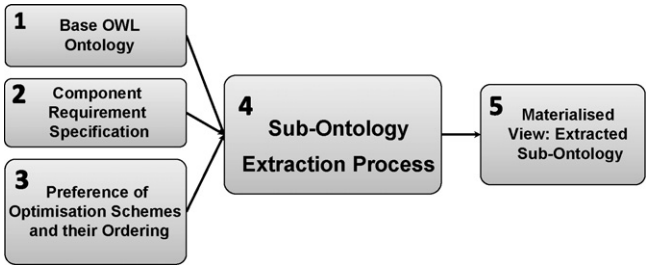


Fig. 2. The sequential sub-ontology extraction process.

tics are represented in a slightly different manner (i.e., a different *view* is established). Intuitively, the definition states that—starting from a base ontology, elements may be left out and/or combined, as long as the result is a valid ontology, i.e., should be a valid ontology even if the base ontology is taken away. In the process, no new elements should be introduced (unless the new element is a combination of a number of original elements, i.e., the *compression* of other elements).

P2. Sub-ontologies as semantic requirement profiles: Given the foundational (domain-independent) basis of the MOVE framework as afore-discussed, consider a scenario where the base ontology consists of a medical domain vocabulary encompassing one or more sources such as Gene Ontology [3], GALEN [43] and the UMLS [48]. Using the approach proposed herein, the area of interest for each medical practitioner (or any arbitrary user) is modelled using the notion of a 'semantic requirement profile', or simply a 'user profile', that semantically represents a user's interests or requirements. The '*user profile*' is essentially modelled as a sub-ontology, which is derived from a universal base ontology (e.g., the UMLS ontology) on the basis of user specified preferences/requirements. Precisely, the profile consists of user specific requirements in terms of the ontological constructs, concepts, relationships and associated constraints as definable using the OWL language. The concepts and relationships themselves are representative of the knowledge contained in the original base ontology from which the sub-ontology or the profile is derived by the application MOVE. In the context of this paper, the base ontology takes the form of the knowledge contained in the UMLS ontology. Further details pertaining to the ontology and the application of semantic profiles is presented in Sections 5.2 and 6 respectively.

5. OntoMOVE: a semantic requirement profiling and retrieval framework

Our earlier work in [9] describes the general framework of OntoMOVE for the purpose of ontology reuse in software development processes in general. In this paper, we highlight the idea of optimizing the processes of ontology-based information retrieval through the utilization of sub-ontology annotation and extraction. We also demonstrate the effectiveness of our proposal through a prototypical implementation in a medical domain with large size document resources in the form of the Medical Therapeutic Guidelines (TG) [45] and an empirical analysis of the results from the viewpoint of Contextualization.

5.1. Overview of the framework

OntoMOVE stands for 'Ontologies on the *MOVE*'. The term 'MOVE' itself is an acronym for 'Materialised Ontology View Extraction' (see Section 4), which is foundational to the work reported herein. A brief overview of the main aspects of the OntoMOVE framework illustrated in Fig. 3 follows in (F1–F6):

F1. Component requirement specification: We use the general term 'component' to refer to an application or user that can supply a set of requirements/preferences that are expressed as a partial labelling with respect to a base ontology (e.g., the UMLS-SN). This labelling is indicative of the semantic types (concepts and relationships) that reflect the component's field of specialization or interest. Indeed, for practically deployable scenarios, we do not expect users to manually provide such a labelling. However, its provision is achieved as such in our prototypical implementation. Henceforth, this will be referred to as a 'component requirement specification' or simply a *requirement specification*. Also, note the term 'requirements specification', which is utilized in several disciplines such as software and process modelling, where, in general,

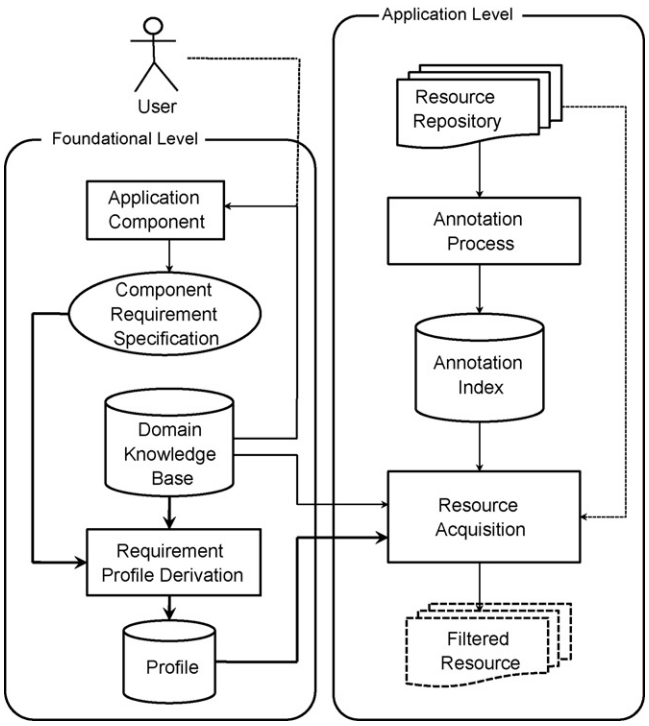


Fig. 3. OntoMOVE Framework.

it refers to the description of a system or process being modelled [24]. The usage of this term in this work must be disambiguated – we categorically state that here it refers to the approximately or incompletely specified knowledge by an user or an application program in the form of a partial labelling of a base ontology.[1]

F2. Requirement profile: On the basis of the initial requirement specification, a profile of the requesting component is derived (see 'requirement profile derivation' in Fig. 3) based on the concept of a sub-ontology (Section 4). A component requirement profile is a *complete* [2] requirement specification that is derived using the partial (initial) requirement specification that is provided by the component. Between the initial requirements specification for a component and the derivation of its complete *semantic requirement profile* lies the derivation of sub-ontologies (i.e., MOVE), as explained in Section 4. Finally, it is instructive to disambiguate a component's 'requirement specification' from a component's 'semantic requirement profile'. Whereas the former is a partial specification of interest from a base ontology, the latter is a complete, independent sub-ontology derived from the base ontology on the basis of the former. The derivation itself is achieved by the application of the extraction process in (P2), i.e., the optimisation schemes in (OS1–OS4).

F3. Resource: We subscribe to a general notion of a *resource* since the actual type of a resource is irrelevant to the resource acquisition framework, i.e., OntoMOVE. However, in so far as this paper is concerned, we focus on resources as being unstructured or semi-structured data sets (i.e., medical therapeutic guidelines) that are of interest in medical information retrieval domain that is being exemplified in this paper. However, in general, the assumption that is applicable in this context is that irrespective of the precise type of a resource, the resource under consider-

[1] We disambiguate, and use this term to conform with its usage as defined in previous works [7,8,50].

[2] The concept of *completeness* is non-trivial and involves qualitative benchmarks along several dimensions. Details are presented in [50].

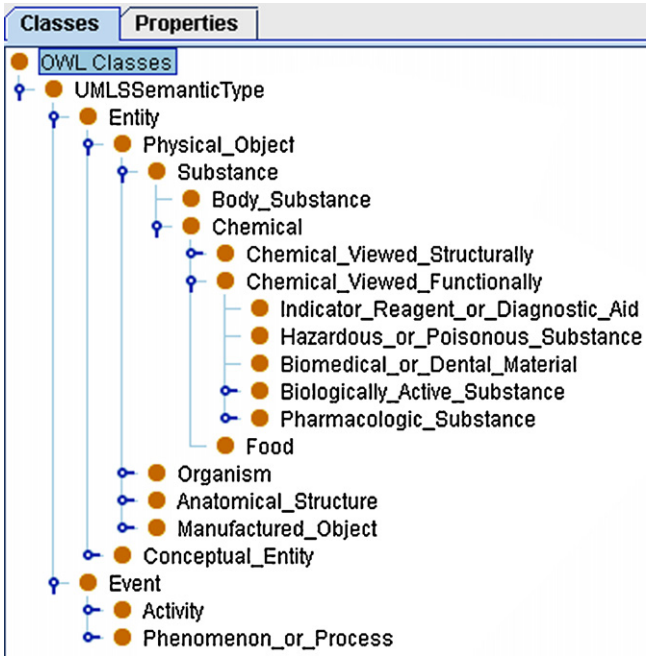


Fig. 4. OntoMOVE–UMLS-SN ontology class view.

ation should be (semantically) categorised using a well-defined ontology that is comprehensively representative of the resource domain.

F4. Resource annotation: Resource annotation refers to the categorization or classification of resources based on a some domain specific criteria such as functionality, its semantic type and similarity to others resources. Although shown as a part of the framework, 'resource annotation' is presently semi-automatically performed using a external tool (see Section 5.3.1).

F5. Resource acquisition: Resource acquisition refers to the process of selectively acquiring (i.e., filtering) resources from a resource repository.

F6. Component requirement manager: By a 'component requirement manager', we refer to that part of the framework which facilitates the creation and maintenance of component requirement specifications, associated requirement profiles, resource repositories and the mapping between a requirement profile and the corresponding resource repository.

In the sub-sections to follows, we elaborate every aspect of the OntoMOVE framework depicted in Fig. 3 for the specific case of requirement profiling, document Contextualization and resource retrieval in medical information systems domain.

5.2. UMLS semantic network ontology—the domain knowledge

The UMLS semantic network (UMLS-SN) represents the meta-level of the medical ontology that we develop for purposes of annotation. This is because the UMLS Metathesaurus uses UMLS-SN as its meta level to define medical domain concepts from various medical vocabularies. We represent selective parts of UMLS-SN taxonomy in the form of a subsumption hierarchy in the OWL language, i.e., as an OWL ontology (referred to as the UMLS-SN ontology henceforth). Fig. 4 illustrates a limited part of the entire UMLS-SN ontology; although all semantic types have been mapped, we have not mapped every property or relationship that exists between the semantic types since the set of relationships between the semantic types is too massive to be used in its entirety and also because it is not our objective to develop a comprehensive mapping of the entire UMLS-SN in the OWL language.[3] Instead, we intend to incrementally enrich and extend the SN relationships only in so much as what is required for us to encompass a given collection of medical TG documents. We use *Protégé* [40] and its associated OWL plugin [41] for the ontology development tasks. As mentioned previously, the Protégé *UMLS tab* (i.e., a plugin) [47] supports browsing of the UMLSKS and can be used to query the UMLS knowledge sources for medical terms and retrieve the results in a terminological format. The results can be exported into our own medical ontology. Obviously, the ontology that results is basically a very small view of the base UMLSKS—one that concerns our use of UMLSKS. The resulting ontology is imported in *OntoMat* [35] in synchrony with the medical information sources, namely the Medical Therapeutic Guidelines (TG) [45]. A subset of the guidelines can then be annotated using the ontology that we imported into OntoMat following which the modified (annotated) documents are serialized as a different version for purposes of testing.

5.3. Semantic categorization of information sources

We are using OntoMat [35], which is a publicly available annotation tool, for purposes of annotating the collection of Medical Therapeutic Guidelines (TG) used in this project. OntoMat annotizer supports the task of creating and maintaining ontology-based OWL mark-ups, i.e., creation of OWL-instances, attributes and relationships. It includes an ontology browser for the exploration of the ontology and instances and an HTML browser that displays the annotated parts of the text. OntoMat serves our purposes since it allows easy import of externally develop OWL ontologies and HTML mark-up, which in our case are the UMLS-SN ontology and the Medical TG respectively.

5.3.1. The annotation process

An annotation refers to the instantiation of an UMLS-SN ontology class in order to relate a chosen 'term' from a TG document with a semantic type from the UMLS-SN ontology. To preserve consistency with industry based medical vocabularies, we constrain our selection only to those terms that are identifiable with the concepts that are present in the UMLS Metathesaurus. The chosen term's placement in the ontological hierarchy is selected on the basis of its registered UMLS Semantic Network type in the thesaurus. Presently, the selected terms are essentially keywords present in the static HTML pages of the Medical Therapeutic Guidelines (TG) with the annotations being embedded in the TG documents using a pre-defined RDF vocabulary. For example, in Listing 1, the RDF-code representing one annotation object has been represented—the annotation object consists of the following information:

(1) The semantic type from the UMLS-SN ontology of the UMLS Metathesaurus *term* being annotated.
(2) The exact location of the *term* within the Medical Therapeutic Guidelines using XPointers.
(3) The precise *term* being annotated and a well-formed label associated with it meant for internal/system use.

Although the annotation objects are instances of semantic types in the UMLS-SN ontology (i.e., the TBox), the RDF based instantiations are different from the actual instances (i.e., the ABOX) that corresponds to the UMLS-SN terminology. Note that the example in Listing is only one form of annotation; additionally, we also distinguish between other forms of annotations, namely those obtained

[3] The ontological or knowledge engineering perspective to be applied whilst mapping all parts of the UMLS semantic network to the OWL language is problematic [25].

by establishing relationships between the UMLS-SN semantic types by way of datatype and object properties. Whilst such properties are very important from an information retrieval viewpoint since they establish links between disparate but conceptually related information sources, we regard such links to be only orthogonal to the main annotation task (by a domain expert) that establishes the semantic types of medical terms contained within the therapeutic guidelines. Once the main annotation task is achieved, the secondary annotation task of providing fillers or values for the *domain* and *range* of properties can be handled even by a non-specialist on the basis of the UMLS-SN semantic types and their instances that are already obtained via *term* annotation by the domain expert.

Listing 1: Annotation object.

```
<UMLS-SN:MEDICAL_DEVICE
    rdf:about="http://ontoserver/tgc/rsg/1078.htm#LARGE_VOLUME_NEBULIZER_DEVICE">
    <rdfs:label>Large volume nebulizer device</rdfs:label>
</UMLS-SN:MEDICAL_DEVICE>
<ontomat:ReificationDataIndividual>
    <ontomat:CreationSource>
        http://ontoserver/tgc/rsg/1078.htm
            #xpointer(//point()[523]/range-to(//point()[537]))
    </ontomat:CreationSource>
    <rdfs:label>
        about:http://ontoserver/tgc/rsg/1078.htm#LARGE_VOLUME_NEBULIZER_DEVICE
    </rdfs:label>
    <ontomat:AboutIndividual>
        http://ontoserver/complete/tgc/rsg/1078.htm#LARGE_VOLUME_NEBULIZER_DEVICE
    </ontomat:AboutIndividual>mat:AboutIndividual>
</ontomat:ReificationDataIndividual>
```

5.3.2. Scope of annotations

For our present demonstrative purposes, 1156 UMLS Metathesaurus *t* erms from a collection of 170 TG documents were selected for annotation on the basis of their UMLS-SN semantic type. Toward this task, 98 semantic types from a total of 135 have been utilized; with the difference representing those semantic types that did not have any instances in the selected document collection. It must be pointed out that these metrics (and others available within our system) are useful in determining the quantity and quality of the annotations being performed and can be dynamically obtained from within the system. For flexibility, the Annotation-Indexer (Section 5.3.3) and its associated statistics generation capabilities (for examples, see Sections 7.1 and 7.2 and the evaluation in Section 8) have been implemented to be usable either with or independent of the main *OntoMOVE* application.

5.3.3. Dynamic annotation and indexing

Dynamic annotation and indexing, in so far as this work is concerned, refers to the capability to dynamically integrate new annotations and build respective index entries thereby supporting integration of incremental updates and extensions to the underlying information sources (e.g., medical therapeutic guidelines). The new annotations could either belong to an existing index or they could be entirely new set of annotations for a different domain altogether. This functionality is important and essential for primarily two reasons, as stated in the following:

(1) Incremental annotations and information update: Annotation is not a one time task and is indeed often performed in an incremental manner. This is primarily because the task is inherently qualitative (or human expertise driven) in nature and as such, requires considerable refinements before a complete and consensual semantic categorization of data is obtained. Furthermore, existing information sources may possibly be extended so as to include completely new information thereby necessitating new annotations.
(2) Applicability in multiple-domains: We presently focus on semantic information retrieval the medical domain. However, the OntoMOVE framework is applicable in domains other than the one considered herein. As such, in order to preserve the generality and re-usability in other domains, it is necessary that the system remain functional when annotations for information sources from other domains are being utilized. This concept if re-usability is illustrated in Fig. 5: the annotation database consists of multiple indexes, with each possibly corresponding to a different set of information sources from which data needs to be retrieved

A built-in *Annotation-Indexer* maintains and builds the relationships, namely *index entries*, between the existing semantic types and their instances in the medical TG. For purposes of efficiency, the index generated by the Annotation-Indexer is serializable-deserialisable; as such, generation of the index is a one-time task and a user may either choose to reload an existing index or regenerate one if there has been some change in the information sources.

6. An application of OntoMOVE in the bio-medical domain

We incorporate the idea of a sub-ontology based user profile that contextualizes the respective user's area of specialization on the basis of the semantic types (and relationships) that are present in the corresponding sub-ontology. The concept of utilizing sub-ontologies has an interesting application: when only partial views or sub-ontologies of an existing ontology are used, the semantic range of the sub-ontology in the overall annotated dataset is significantly narrowed (see Fig. 6); something that leads to the idea of a requirement specification based semantic profile derivation.

This scenario, intuitively depicted in Fig. 6, illustrates a base ontology comprising of a collection of semantic types (solid circles) and relationships among them.[4] As evident from Fig. 6, the resulting sub-ontology (from the application of the process of sub-ontology extraction) essentially corresponds to a sub-set of the overall medical information sources. This is denoted in Fig. 6 by a collection of primitive annotation types[5] (solid rectangles). An *annotation index* connects the semantic types to the terms occurring in the medical document source, which in our case is the Medical Therapeutic Guidelines (TG) [45]. The annotation index (Section 5.3) is a mapping between individual terms in the TG and their semantic types in the ontology. Using this setup, a user's interest or

[4] Relationships are not explicitly represented in Fig. 6 since that is not relevant in the present context and would unnecessarily take up space. Indeed, it may be presumed that the conceptual entities, denoted as solid circles in Fig. 6, also have relationships among them.

[5] Depending on the granularity, this could be one document, or a medical vocabulary defined term occurring in it. In this paper, the latter interpretation applies.

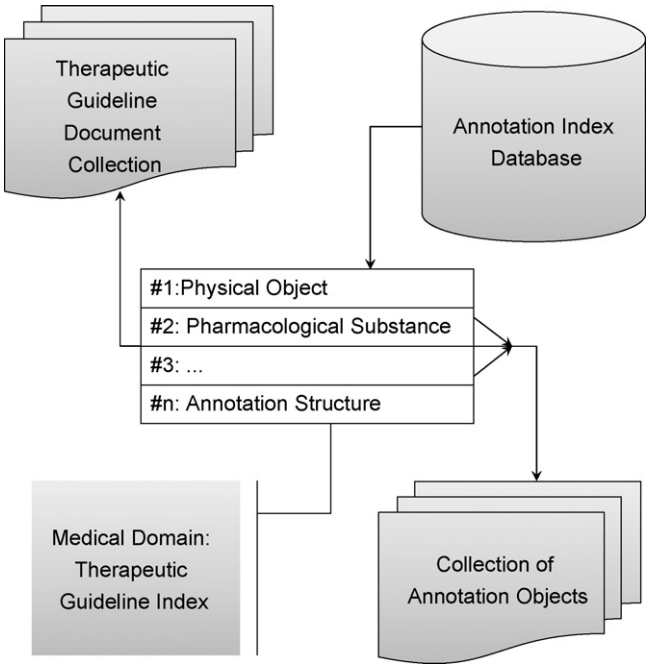


Fig. 5. Dynamic annotation and indexing.

field of specialization from the overall collection of TG is defined to be the mapping produced by a independent sub-ontology of the base ontology that reflects the semantic types related to the user's interest. Obviously, different sub-ontologies will map to different sets of documents/resources in the overall TG set, with the mapping information coming from a (pre-computed or dynamically generated) annotation index. This is denoted in Fig. 6 by the dashed-arrow connecting the two dashed-circles.[6] As discussed in Section 4, the semantic requirement profile itself is derived by applying the sub-ontology extraction algorithm on an incompletely specified '*requirements specification*' supplied by a end-user or application.

In Fig. 7, we apply the general idea of requirement driven resource retrieval (in Figs. 6 and 3) for the bio-medical domain. A brief description of the entire process, illustrated with an application-centric viewpoint in Fig. 7, follows:

Stage 1. Domain knowledge base: The domain ontology which represents the entire knowledge within the specified domain. In this example, we utilize and extract from UMLS (Unified Medical Language System) and GO (Gene Ontology) as the two base ontologies to contextualize the bio-medical area.

Requirements specification: a set of concepts and properties from both the UMLS-SN and GO base ontology are extracted on the basis of the area of interest or specialization of the end-user. Note that this specification is rather crude and does not constitute a valid sub-ontology. Basically, this specification takes the form of a 'labelling' of concepts, attributes and relationships that are of user's interest. For example, labelling for concepts related to 'Cancer'

Stage 2. Conceptualizing the requirements specification: Our system will extract a valid sub-ontology based on the requirement specification labelling as mentioned in Stage 1 above. The sub-ontology is indicative of the user's preferences as well as some implicit relevant concepts considered essential for inclusion by our system to make it a valid sub-ontology. For example, the valid sub-ontology for 'Cancer Treatment' is created from the UMLS, and a sub-ontology for 'Cancer Genome' is created from the Gene Ontology.

Semantic context based information retrieval: once a context has been established, all search queries that are performed within the context will narrow the range of the information documents to include only those documents (or their parts thereof) that are relevant to the context. Note that this functionality is encapsulated within the application of the sub-ontology extraction algorithm. In our example, we will retrieve all documents related to 'Cancer Palliative Treatment' from within the palliative care document repository (e.g., TG) and documents related to 'Cancer DNA Mutation' from the biosciences PubMed document repository.

Stage 3. Finally, collaborating end-users with disjoint and even overlapping specializations in the base ontology may obtain context specific sub-ontologies, and consequently, specialized search results whilst looking-up annotated resources in an annotated resource database. For example, the *Centre for Cancer Genome* would focus on the specific information from the Gene Ontology context, whereas a general *Cancer Research Centre* may need to create a context that combines concepts from the Medical ontology ('Cancer Treatment') as well as the Gene Ontology ('Cancer Genome' information).

In essence, there are 3 types of changes that have to be taken into consideration in the aforementioned OntoMOVE stages:

- Document collection updates: in this case, the Contextualization process in Stage 2 will need to be incrementally updated each time a new set of documents are required to be included or removed.
- User requirement changes: a new sub-ontology based on the new set of requirements will need to be created. The new sub-ontology can be created simply as a new 'version' of the previously built sub-ontology.
- Domain ontology evolution: whilst ontology is expected to be mostly stable, there are cases where the main ontology evolves. In this situation, the changes will have to be broadcasted to all relevant sub-ontologies. Whilst this is an important issue to address, the main focus of this paper is on the novelty of our proposed extraction process.

7. OntoMOVE: an implementation overview

We briefly present the accessibility information relevant to retrieving information from the medical sources via the *OntoMove Search Interface* (see Fig. 8(a)). The user performs a search query consisting of N keywords. In addition, the user may also specify the means of combining the keyword, viz - either conjunctively or disjunctively. By default, the '*AND*' operator will be used to combine the search keywords in case where $N > 1$. In addition, the user may optionally choose to apply a *profile* (Fig. 9(a)) in the context of which the search is to be performed. Note that it is also possible to apply more than one profile at the same time, for example, applying a *patient* and *pharmacist* profiles in conjunction. As discussed previously, the effect of applying a profile (or a combination thereof) is to establish context based on semantic information about the users area of interest that is contained in the profile. As can be seen in Fig. 8(a), the retrieved search results are sequentially listed in the lower part of the interface—the precise order of the results is based on the conceptual similarity/distance between the identifiable semantic type of the search keywords with the anno-

[6] In Fig. 6, solid lines denote information flows whereas dashed lines refer to conceptual links between related components.

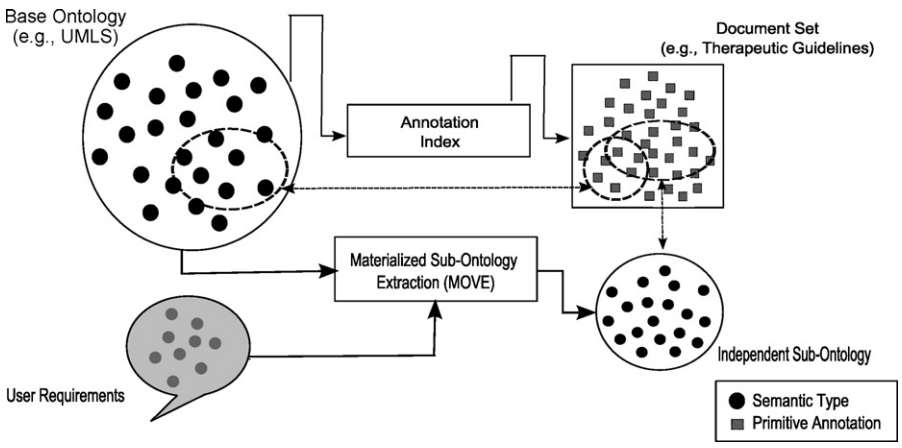


Fig. 6. Ontology based profiling and document contextualization.

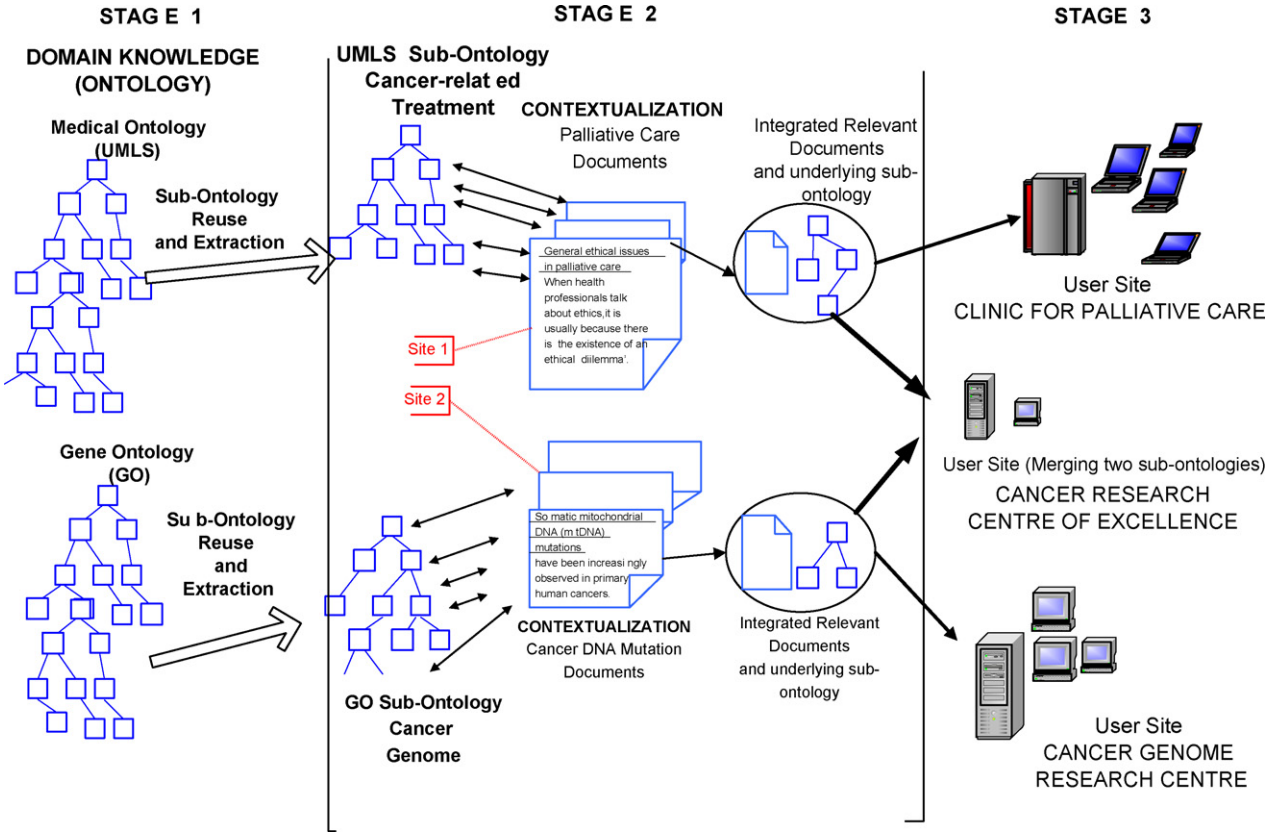
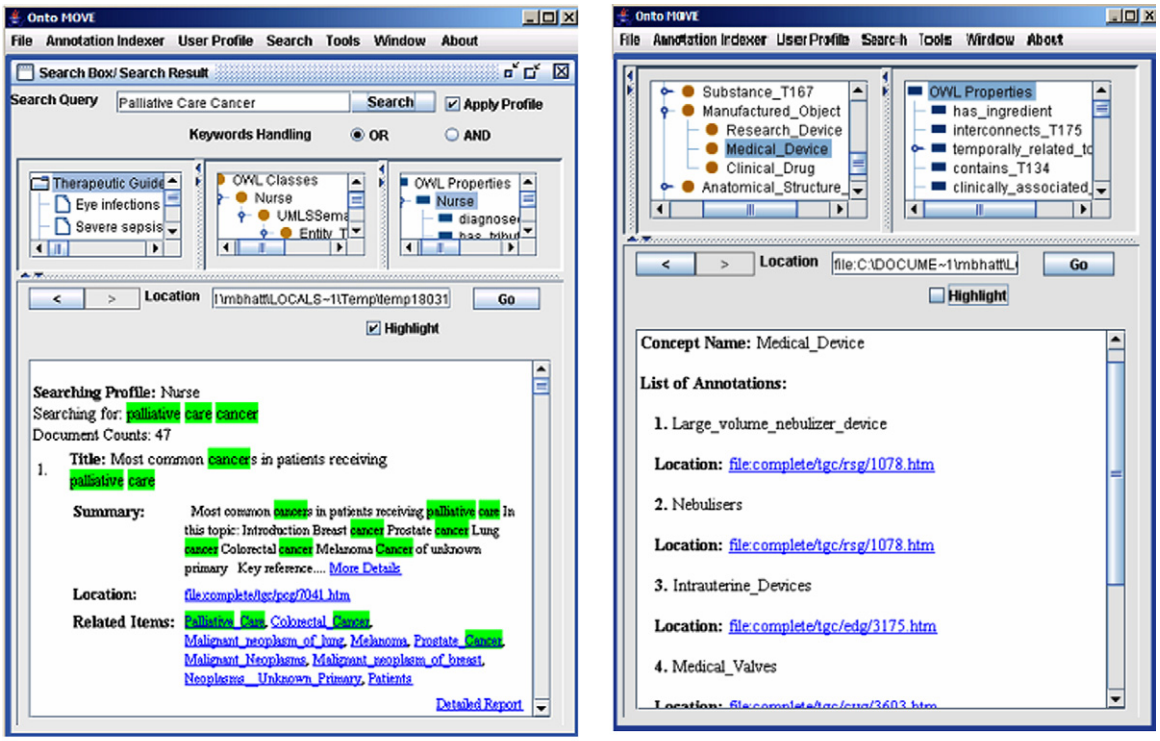


Fig. 7. OntoMOVE: a sample application in the bio-medical domain.

tations that are present in the retrieved documents. In addition to a brief summary of every document that is retrieved, the user also has access to a detailed *S* emantic Report (Fig. 9(b)) and an utility-oriented *Annotation Viewer* (Fig. 8(b)). A brief description for the two is provided in the sections to follow.

7.1. Detailed semantic report

The report consists of (semantic) information about other Metathesaurus items, which might possibly be of interest to the user, that are present in a certain document retrieved as a result of the search process. For an example, see the report in Fig. 9(b) that is obtained for the very first result/document out of a total result set of 7 documents by searching for the key-words—'*Palliative Care Cancer*'. Note that in addition to a provision for easy navigation of search results, the interface (see middle portion of Fig. 8(a)) also allows for direct browsing of the *hyperlinked* information sources directly in the form of a tree-structure. This tree-structure is obtained solely on the basis of the structural organisation provided by the information source and is derived (only once) by parsing the complete set of HTML based information sources. As a pointer to future work, it would be interesting to obtain a *dynamic taxonomy* from the set of documents that are retrieved for a search query in question. However, this is beyond the scope of our project and presently



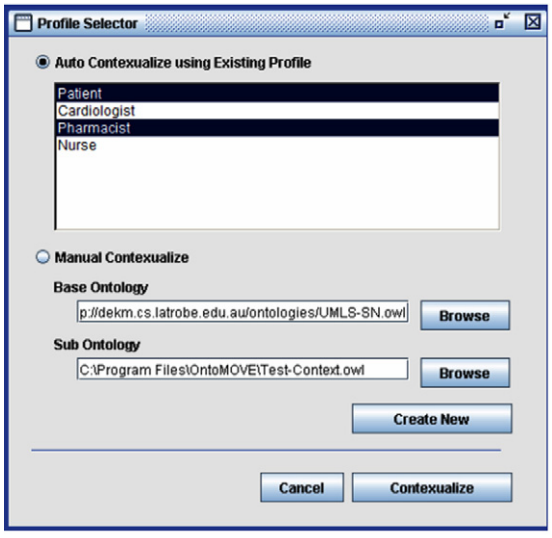
(a) TG Information retrieval (b) Annotation Viewer

Fig. 8. OntoMOVE information retrieval interface I. (a) TG information retrieval. (b) Annotation viewer.

remains a speculative note deemed worthy of further investigation.

7.2. Annotation viewer

An 'Annotation Viewer' (Fig. 8(b)) is available wherein the conceptual range of any arbitrary semantic type form the UMLS-SN ontology within the TG documents may be obtained. The viewer exploits the existing *annotation indexer* and/or a previously serialized index (if one is available). For every semantic type within the presently loaded ontology (or sub-ontology if *p* rofiles are active), the steps illustrated in Listing 2 are performed to determine the range of a selected semantic type or property.



	Keywords	IS-A	Semantic Type
1	Palliative_Care	<=>	Health_Care_Activity
2	Colorectal_Cancer	<=>	Neoplastic_Process
3	Malignant_neoplasm_of_lung	<=>	Neoplastic_Process
4	Prostate_Cancer	<=>	Neoplastic_Process
5	Malignant_Neoplasms	<=>	Neoplastic_Process
6	Patients	<=>	Patient_or_Disabled_Group

(a) Profile Selector (b) Semantic Report

Fig. 9. OntoMOVE information retrieval interface II. (a) Profile selector. (b) Semantic report.

Listing 2: Establishing semantic scope.

```
/*
        INPUT : A semantic type (concept or property)
                from the 'requirement profile' (R)
        OUTPUT: A set of concepts that are either directly or
                indirectly related to a named concept/property
                (ResultSet).
*/
    //Main Function to get Semantic Scope
1.  function getSemanticScope () : ResultSet
    {
        ResultSet = {};
        if ( R is Concept )
        {
1.1         ResultSet = ResultSet ∪ getSubConcept ( R );
        }
        else if ( R is Property )
        {
            domainConcept = domain(R);
1.2         ResultSet     = ResultSet ∪ getRelatedConcept ( domainConcept );
            rangeConcept  = range(R);
1.3         ResultSet     = ResultSet ∪ getRelatedConcept ( rangeConcept );
        }
        tempSet = NULL;
        Loop through each Concept in ResultSet
        {
            C = next(ResultSet);
1.4         tempSet = tempSet ∪ getEquivalentConcepts(C);
        }
        return ( ResultSet ∪ tempSet )
    }
    //Helper Function to get Related Concepts Result Set
2.  function getRelatedConcepts ( Concept R ) : ResultSet
    {
        PartialResultSet = NULL;
        if ( R is Primitive )
        {
2.1         PartialResultSet = PartialResultSet ∪ R;
        }
        else if ( R is UNION OR R is INTERSECTION )
        {
            Loop through each Concept in R
            {
                C = next ( R );
2.2             PartialResultSet = PartialResultSet ∪ C;
            }
        }
        return PartialResultSet;
    }
```

In the following, we briefly explain the algorithm in Listing for establishing the semantic scope of semantic type from the profile within a annotated resource repository: firsly, if a primitive concept is selected', then it is simply added to the established semantic scope denoted by *ResultSet*. If a property is selected instead, then the task is to find the *Domain* (*domainConcept*) and *Range* (*rangeConcept*) of the selected property and perform this algorithm for each of them. In the final step of the algorithm, the *ResultSet* that is obtained is essentially a set of concepts from the profile. This set is complemented with all concepts that have been defined to be *equivalent* (using *OWL* : *sameAs* property) to any concept present in the original result set. This final step (potentially) widens the semantic scope of the results by including the synonym concepts.

Once the set of concepts resulting from the application of above mentioned steps is obtained, the *annotation index* is queried to retrieve the list of *annotation objects* that exist for each of the concept included the result set. Finally, the information in each annotation object is used to dynamically generate an HTML-document that provides a list of documents represented by the list of annotations (see Fig. 8(b)).

8. Analyzing contextualization capability

OntoMOVE's performance can be evaluated along two fronts. Firstly, the sub-ontology extraction mechanism, which underlies the OntoMOVE framework, can be evaluated independently from its proposed application in the form of profiling and Contextualization for the medical information retrieval domain. Here, the focus is on OntoMOVE's capability to generate sub-ontologies that are semantically complete, optimal and independent of a base ontology. Secondly, performance of OntoMOVE's proposed application toward semantic requirement profiling and Contextualization in the medical information systems domain may be evaluated. Here, OntoMOVE's capability to contextualize or restrict an information retrieval request to only those parts of the overall repository that are semantically related to the user's area of interest is highlighted. Considering the medical domain-specific scope of this paper, we restrict ourselves to the evaluation along the latter application front.[7]

8.1. Scope of analyses

We restrict the evaluation to the Contextualization capability of the system, as opposed to the empirical investigation of the precision and recall abilities of the information retrieval phase. Here, we are primarily interested in ensuring the consistency of two-way Contextualization—specialising a context or further expanding it by the refinement of the requirements specification. The manner in which such a refined, i.e., expanded or further specialized, con-

[7] The underlying sub-ontology extraction has been qualitatively analysed with practical illustrations in [50].

Table 3
Annotation metrics.

Setup	Value	
(a) Annotation and concept coverage		
Ontology	UMLS-SN	
Total concepts	135	
Annotated documents	170	
Total annotations	1156	
Concept coverage	95/135	
Statistic	**Annotations per concept**	**Annotations per document**
(b) Annotation statistics		
Max	226	43
Mean	8.56	6.8
Standard deviation	23.41	6.67

text is exploited by a information retrieval module, and its resulting precision and recall abilities, is independent of the Contextualization module per se. Hence, here we focus on the Contextualization capability.[8]

8.2. Base setup

For evaluation purposes, we use the setup illustrated in Table 3. The base medical domain ontology (i.e., UMLS-SN; see Section 5.2) being used consists of a total of 135 concepts. Using these concepts, a total of 170 documents from the overall Medical Therapeutic Guidelines have been annotated, resulting in a total of 1156 annotations. Note that from the overall 135 concepts in the base ontology, a majority of 70% have corresponding annotations in the document-set, with the remaining concepts being those that did not have any semantically related resources in the medical TG document-set. The absence of corresponding resources to annotate results from the fact that the (test) TG document-set chosen for the analysis is a small fraction (approximately 10%) of the complete TG collection.

8.3. Annotation quality

From the viewpoint of a conventional information-retrieval centric analyses, which is not the objective here, a limitation of our prototype is that our annotations are not performed by a domain-expert. As such, the annotation process also lacks a global annotation strategy. A general measure of the optimality or correctness of a particular distribution is not quantifiable, that being governed by the nature of the document-set being annotated. As a guideline toward preventing over/under representation of some medical resources within the resource repository, we adopt the heuristic that the annotations be (approximately) normally distributed throughout the document-set. However, note that this is simply a work-in-progress heuristic and will be no more applicable once a domain expert is involved (see Section 9). The heuristic also ensures that every document in the test-set is annotated in order to provide coverage to every identifiable semantic content present in it.[9] Given the availability of annotation statistics, such as in Fig. 10(a), this, or potentially other annotation heuristics, are easily achievable within the framework.

Table 3(b) consists of basic statistical data relevant to the annotation process. Fig. 10(a) reflects the overall frequency distributions for the number of annotations per document that resulted from the annotation process for the document-set under consideration. Both, the concept and annotation coverage data as well as associated statistics are generated within the system for the singular reason that they are reflective of the qualitative aspects of the annotation process. This functionality is useful for fine-tuning the annotation strategy, which, with or without a domain expert, is generally considered to be a key component in the quality and improved accuracy of the information retrieval [13,36].

8.4. Contextualization capability

Toward the main evaluation task, we utilize a base requirement specification that approximately spans the entire UMLS-SN ontology. Based on this requirement specification, a complete and independent sub-ontology (referred to as ontology A) is created. The same process is repeated 3 times, each time using the sub-ontology from the preceding stage as the base ontology for the next stage. The result is a collection of four sub-ontologies (referred to as A, B, C and D) or user profiles that share the following relationship: $[A \supset B \supset C \supset D]$. The rationale behind this approach is that since the semantic scope (see Section 6) of the overall base ontology spans the entire test document-set, further specialization of the base ontology should result in a corresponding contraction of the semantic scope within the document-set. The results in Fig. 10(b) illustrate that this is indeed the resulting behaviour, i.e., the semantic scope of profile A, which consists of 107 concepts, spans 168 documents, whereas with profile D, the scope is narrowed down to 113 documents. Note that although multiple, non-related profiles can be applied toward a certain Contextualization task, we use the afore-discussed process of incremental specialization with one profile in order to make the results comparable since multiple profiles without non-overlapping requirements are not comparable.

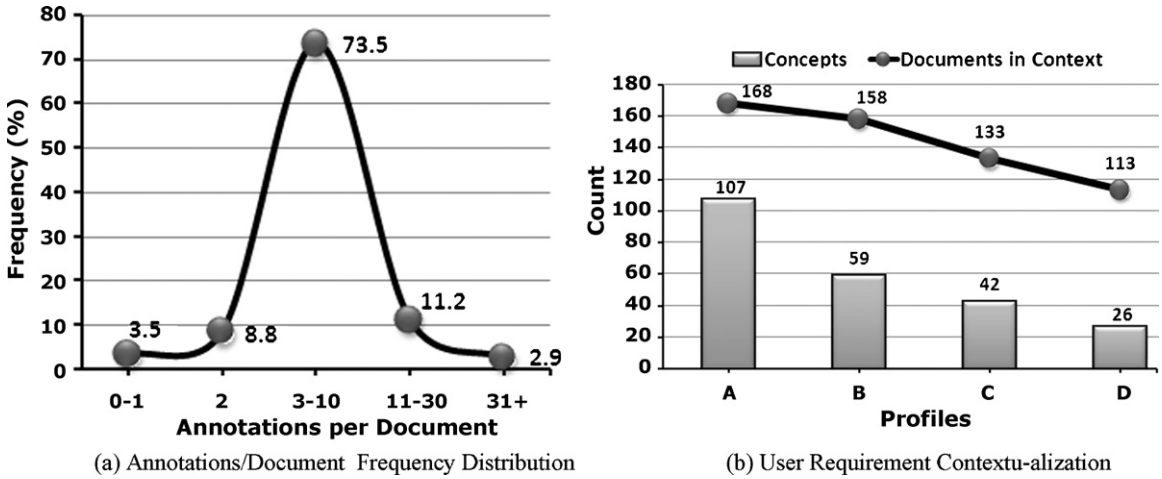
Once the semantic-scope for a profile within the overall document-set is derived, the resulting scope essentially provides a means for the retrieval of semantically valid information for the user. Table 4(a), consisting of results for individual search queries, reflects the effects of Contextualization of the user's interest or requirement for sample queries. We highlight the results for 4 queries (called $Q1$, $Q2$, $Q3$ and $Q4$) and additionally, also include the average results for a total of 21 queries. For instance, the results for profile A in row one of Table 4(a) indicate that from a total of 168 documents, queries Q_1 results in 67 relevant documents whereas for profile C, which is a specialized profile in relation to A, the result drops down to 55 documents. The overall trends for all the profiles and the respective queries are shown in Table 4(a). Similarly, the effects of Contextualization when multiple, disjoint profiles are applied are shown in Fig. 11. For ease of reference, the results obtained when comparing both related and non-related

Table 4
Effects of context specialization and expansion.

Profile	Query result				
	Q_1	Q_2	Q_3	Q_4	Mean (21 Queries)
(a) Setup I: Incremental specialization of context					
A	67	64	51	29	16.64
B	65	62	48	27	14.4
C	55	50	43	22	13.55
D	49	42	39	18	11.9
Profile	**Concepts**				**Documents in context**
(b) Setup II: Specialization and expansion of context					
E	59				158
F	26				113
G	79				117
$E \cup G$	138				169

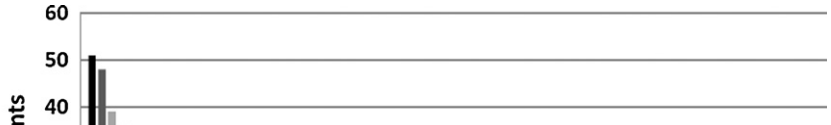
[8] Section 9 further elaborates the potentiality of a precision and recall based analysis from a conventional information retrieval viewpoint.

[9] This identification process itself is complex, given the fact that it pre-supposes intricate domain-specific knowledge that is typically available only to a qualified expert.



(a) Annotations/Document Frequency Distribution (b) User Requirement Contextu-alization

Fig. 10. Annotation frequency and document contextualization. (a) Annotations/document frequency distribution. (b) User requirement contextualization.



Profile	Queries													
	1	2	3	4	5	6	7	8	9	10	11	12	13	14
E U G	51	16	29	16	8	7	8	7	10	7	6	4	5	17
E	48	13	27	16	6	6	7	5	9	4	4	3	3	13
F	39	11	18	12	6	5	6	3	4	3	3	2	2	8
G	36	14	22	15	5	4	5	4	8	5	4	3	2	11

Fig. 11. Two-way consistency of contextualization.

(i.e., disjoint) requirement profiles are illustrated together. Here, we compare three profiles, E, F and G, which are related in the following way: $[E \supset F]$ and $[E \cap G \equiv \{\}]$. The objective of using the setup in Table 4(b) and Fig. 11 is to show that Contextualization can operate both ways, i.e., for further (incremental) specialization of interest (as exemplified with the setup in Table 3(a) and Fig. 10(b) involving profiles A, B, C and D) as well as for the expansion of the semantic scope with the document collection. Note from the setup in Table 4(b) that profiles E and G are disjoint and that $[|E \cup G| = |E| + |G|]$[10] holds, this relationship being important for our present exemplary purposes. The following should be noted from Fig. 11, which illustrates the results for 14 test queries: (a) Where a profile is a specialization of another (e.g., F is a subset of E), the resulting documents with the specialized profile are consistently lesser than those for the general profile across all queries (this was illustrate in Fig. 10(b) as well). (b) When independent profiles are applied in conjunction (e.g., $E \cup G$), the results are consistently greater than those for any of the individual profiles applied in isolation across all queries. Note that the total number of results for the combined profiles need not be a sum-total of the results for the individual profiles since a document may consist of annotations of concepts from both the profiles (this is exemplified in Fig. 6 with overlapping ovals). Obviously, as mentioned previously, disjoint profiles (e.g., E and G) are not comparable in isolation. As such, the respective data for profiles E and G in Fig. 11 are not relevant.

9. Summary and outlook

Although our methodology of sub-ontology based semantic requirement profiling and information retrieval is generally applicable in any domain where it is possible to annotate resource-sets, as an exemplar, we have designed *OntoMOVE* so as to be specifically applied in the medical information systems domain. OntoMOVE utilizes semantic web [5] standards like RDF(S), and the Web Ontology Language (OWL) in addition to medical domain standards and controlled vocabularies encompassed by the UMLS Knowledge Sources (UMLSKS), namely the UMLS Metathesaurus and the UMLS Semantic Network. The system offers advanced profiling of a users field of specialization and/or interest by exploiting materialised sub-ontologies produced by *MOVE*. Our methodology consists of making the semantic content present in the medical information sources explicit and building a system that takes advantage of the explicitly represented knowledge. At the core of our system lies

[10] This implies that the profiles under consideration do not share any common concepts. Here, cardinality of a profile is equal to the number of *distinct* concepts included in the profile.

semantic web based medical domain knowledge in the form of an ontology and its preference/user dependent sub-ontologies. For demonstration purposes, we utilize the Medical Therapeutic Guidelines [45] documents as the information source and selectively annotate a partial collection of the TG documents encompassing a few select categories form the overall collection.

9.1. Expert-driven annotation and empirical study

The current focus of our work in this paper is to utilize a sub-ontology profiling technique to perform contextual search and to filter relevant documents based on the user's profile as defined by the sub-ontology. As such, our goal is to prune document corpus based on our sub-ontology Contextualization approach. It is expected that this process will be further refined in the future, whereby the relevance of the document collections are then ranked based on their relevance to the user profile. Because our focus here is to demonstrate a novel approach of document filtering based on a sub-ontology extraction technique, our evaluation technique is currently experimental-based, and our future aim will be to perform the evaluation on real domain experts in the area. Indeed, this will also be necessary to perform a realistic empirical study from an information retrieval viewpoint since the quality and/or correctness of information retrieval and their benchmarking in terms precision and recall is not possible without domain-expert driven annotation strategy [13,36] and result ranking.

9.2. Annotation automation potentiality

As further work, we also regard it important to at least semi-automate the annotation process by utilizing the *deep annotation* technique that is suited to scientific (Medical or Bioinformatics) ontologies [20]. Toward this end, dynamic interaction via web-services with a corpus of Medical domain *t* erms, the UMLS Knowledge sources and the Medical information sources (in a semi-structured form) seems essential. Work is in progress to develop built-in functionality, similar to that utilized via OntoMat, to create manual annotations using our custom annotation vocabulary. Although essentially similar to OntoMat in methodology, this built-in functionality will differ in two regards: (a) instead of an RDF based annotation schema, we develop our custom OWL ontology based schema, (b) the annotation schema itself will not be static, as is the case with OntoMat, thereby allowing users to specify their own annotation types. Most importantly, these extensions also facilitate the provision of the entire information retrieval methodology in one single application.

9.3. Ontological reasoning capability

Other interesting extension involves exploiting the ontological reasoning facilities that are available for OWL described resources using existing tools description logic reasoners such as Racer [42]. Finally, the Contextualization in the present system is performed in a built-in sequential manner. We are working toward utilizing the distributed version (see [8]) of our framework that has been designed for use in a distributed cluster environment. We propose to achieve this goal via the medium of web-services, i.e., a web-service performs as a mediator between the distributed framework operating in a Linux cluster environment and the Java-based *OntoMOVE* application.

9.4. Integration within an information retrieval system

The present work categorically focussed on semantic requirement profiling, and resource Contextualization and its empirical investigation. We regard that any specialized ontology and annotation based information retrieval system will benefit from our Contextualization approach. For instance, as illustrated in the Contextualization analyses in Section 8, incremental specialization and expansion of the context is consistently achievable by the application of our profile derivation approach. These may in turn be utilized together or in isolation by an information retrieval system to fine-tune/control its search space and improve the quality of its results from a semantic viewpoint. In this context, an important functionality that need development is the automation of the requirements specification (Section 5.1) stage so that any arbitrary application may communicate its needs automatically to our system. Once this is achieved, in conjunction with the aforementioned task of involving expert-driven quality annotation, a conventional precision and recall study of the entire Contextualization-backed information retrieval phased would be feasible, and beneficial.

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