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An ontology service for supporting reasoning in medical applications

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Abstract. In the past, the Semantic Web technologies have just been utilized to overcome some important challenges in different fields, such as knowledge and content management, electronic commerce, and so on. We think that the Semantic Web technologies can be also utilized in the context of medical imaging, applied both to medical practice, in order to provide a decision support, and to research about pathologies. In particular, anatomical structures can nowadays be automatically segmented, but a semantic identification of their parts remains an open question. To tackle these issues, we have applied a flexible and extendible approach, based on the integration of OWL ontologies and SWRL rules, for modelling all the relevant concepts of the anatomical knowledge. Besides, we have designed and developed an ontology-based service that i) exploits OWL ontologies and SWRL rules and ii) performs logic and reasoning mechanisms, in order to face topic issues, as the labeling and semantic identification of anatomical structures. Finally, as an application, we utilize the set of ontologies and rules achieved by O. Dameron at IDM, that model the most important anatomical structures of the brain, for giving a flavour of how rules, ontologies and the reasoning process about them might enable the labeling of unknown objects.

1. Introduction

Computer technology has been applied to almost all aspects of human activities including medicine. In particular, a frequent use of this technology is in medical image processing and analysis. As a matter of fact, systems for analysis of medical images can be on computer assisted or fully automated systems. They produced medical images that are usually obtained by X-rays, microscopes and especially in recent years by magnetic resonance (MR) and computed tomography (CT) scanners. Segmentation and labeling of various tissues types in medical imaging are important objectives in anatomical studies, diagnosis or pre-operative planning, especially when large databases are used as knowledge resources, for example a picture archiving and communication system (PACS). In particular, anatomical structures can nowadays be automatically segmented, but a semantic identification of their parts remains an open question. This task is difficult as it requires complex application-specific knowledge and because of image artifacts such as partial volume effects and noise [1].

As a result, the main challenge is modelling the medical knowledge and the diagnostic context in order to label the sought objects and close the semantic gap between low-level pixel information and high level application knowledge.

We think that the Semantic Web [2] technologies can be utilized to tackle these issues. In particular, we have applied a flexible and extendible approach, based on the integration of OWL [4] ontologies and SWRL [3] rules, for modelling all the relevant concepts of the anatomical knowledge.

Besides, we have designed and developed an ontology-based service that i) exploits OWL ontologies and SWRL rules and ii) performs logic and reasoning mechanisms, in order to face topic issues as the labeling and semantic identification of anatomical structures.

As an application, we utilize the set of ontologies and rules achieved by O. Dameron at IDM [21], that model the most important anatomical structures of the brain, for giving a flavour of how rules, ontologies and the reasoning process about them might enable the semantic identification and labeling of unknown objects.

The rest of this work is organized as follows. Section 2 presents motivations and related work. Section 3 describes an ontologies and rules based approach. Section 4 describes an Ontology Service which exploits ontologies and rules for labeling medical images. Section 5 overviews a set of ontologies and rules describing the most important brain anatomical structures and presents an applicative example. Finally, section 6 concludes the work.

2. Motivations and related work

Segmentation and labeling of various anatomical structures in medical imaging are topic issues in fields as medical diagnosis and research about pathologies. These tasks are difficult and problematic as they require application-specific knowledge and make use of images that can be characterized, as instance, by partial volume effects and noise.

As an instance, analysis of brain images has been an important task in recent years and has received a lot of attention in literature. In particular, analysis of brain stroke lesions has shown to be difficult. Stroke lesions appear somewhat darker (hypodense) in CT images, but often with no distinct boundary, so even experienced radiologists do not always agree about the extent of the lesion. Another problem for computer system is finding the position of the stroke. It is obvious that some knowledge must be used to successfully label these images.

A number of authors have used intelligent systems for segmentation of medical images [6]. Research on brain image segmentation using rule-based expert systems has been presented in [7, 8, 9, 10]. Expert systems have also been used for segmentation of lung boundaries [11].

Up to now, Semantic Web technologies have been largely used in different fields, such as knowledge and content management, electronic commerce, and so on.

In medical informatics research, the provision of controlled medical terminology services [12, 13, 14] within clinical information systems constitutes the major challenge of the Semantic Web in order to facilitate semantic interoperability [15, 16].

The desideratum of standardized, ubiquitous and logically consistent terminological knowledge repositories for clinical communication and information management is, however, not accomplished by most existing large terminologies such as MeSH, SNOMED, the Read thesaurus and UMLS.

Differently, we think that the Semantic Web and its technologies can effectively be applied to face other medical topics and, in particular, to allow labeling of a-priori unknown objects, for the following reasons:

- Ontologies can be used to enable the definition of an explicit and specific description of medical vocabularies. They can provide the definitions of concepts and semantic properties associated to these concepts in order to facilitate the interoperability between systems that store, elaborate and query biologic and medical data. and allow declarative processing of data, providing a way to share context knowledge without misunderstandings.
- Rules can be used for capturing the semantic relationships and dependencies between ontological properties or between ontologies and other domain predicates.
- RDF [10], OWL and SWRL are semantic representation languages with high degree of expressiveness that are adequate for modelling ontologies and rules.
- Ontologies and rules can be reasoned by logic inference engines. We can use ontologies and rules coupled with subsets of first order logics to perform logic mechanisms, to infer new information and to ensure that the system is always in a consistent state. All these logic mechanisms can be utilized for generating new knowledge that can be used for labeling unknown objects.

As a result, in this work, we have applied the Semantic Web technologies and, in particular, we have aimed at integrating OWL ontologies and SWRL rules in a unique system in order to model all the relevant concepts of medical vocabularies and to provide a support for the labeling process of unknown objects. A similar approach has been presented and illustrated in [22].

3. The ontologies and rules based approach

In the past, the Semantic Web has just been applied to overcome some important challenges in constructing and managing information in various applicative fields. But, all the systems that have utilized the Semantic Web technologies have adopted approaches only based on ontologies.

Ontologies are represented by using XML based languages, that provide a standard format for them. These knowledge representation languages are i) RDF and ii) Description Logic (DL) based languages, as DAML+OIL [11] and OWL DL [7]. They are characterized by rich expressive power and so they are considered well suited for modelling purposes.

Furthermore, DL based languages are mapped to formal logical models which can be submitted to DL reasoners, as Racer [12] or FaCT [13], in order to ensure logical consistency and to infer new knowledge.

However, an ontology-based approach has often been insufficient and not suited for some critical aspects of many applicative fields because:

- ontology languages are not able, for example, to chain properties (such as transfer properties from parts to wholes), to reason across domains, to map ontologies between them for data integration, to express query and so on [14];
- description logics, and so also the DL based languages, can reason about names, which can include objects and relations, but don't deal with quantitative concepts, including order, quantity, time, or rates [23]. Unfortunately, this kind of reasoning is often essential.

In order to overcome such limitations, rules can be used to extend the expressiveness of ontologies languages [15]. A rule consists of an antecedent and a conclusion. The action specified in the consequent is taken when a rule is considered and the expression in the antecedent is found to be true.

In this work, we have chosen OWL DL as ontology language and we have extended it by using the SWRL rule language [6, 16].

SWRL allows to write rules on the top of OWL ontologies, by enabling ontology concepts and roles to occur in rule consequents or antecedents as unary or binary predicates. In this way, SWRL rules resolve some drawbacks of the ontology languages.

As a matter of fact, SWRL enables to chain properties, to express the whole-part relationships, to express queries and provides a set of built-ins functionalities that allow to perform arithmetic operations and comparisons and, consequently to deal with quantitative concepts.

These two distinct languages can be reasoned by specific inference engines, and, in particular, SWRL rules can be submitted to rule engines, whereas OWL DL ontologies can be processed by DL reasoners.

A complete integration of SWRL rules with OWL DL ontologies in a unique inference engine represents the ideal solution for obtaining a sound and accurate reasoning process, but is inapplicable because of decidability issues. It is impossible to have, at the same time, decidability, soundness, completeness, performance, and expressivity.

A possible solution is represented by a layered approach, that is submitting separately SWRL rules and OWL DL ontologies to a stack of specific inference engines.

Figure 1 illustrates an application of this approach. A is a set of instances of an ontology and it can be submitted to the DL reasoner in order to obtain new knowledge. I_1 represents the inferred knowledge generated after the reasoning process. Next, I_1 can be submitted to the rule engine (in addition to A) in order to apply and execute rules and produce further knowledge. This produces a new set, namely I_2 . Now, two possibilities can occur: 1) I_2 is empty and this means that the reasoning process ends; 2) I_2 is not empty and this means that the reasoning process continues by readdressing the whole inferential set to the DL reasoner.

As a consequence, the process could take several iterations, and only when no further inferred information is generated, it is completed.

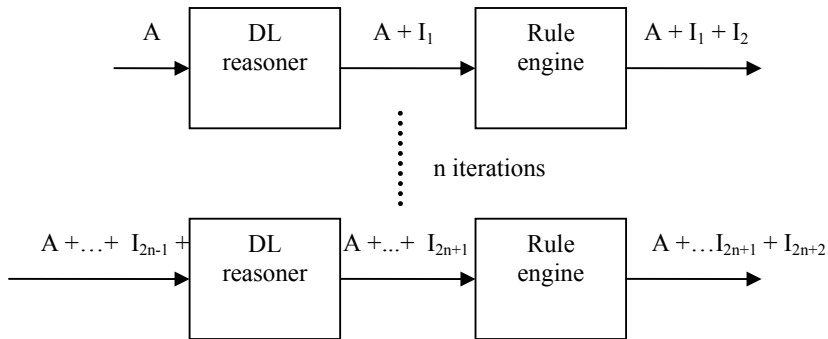


Figure 1 – The layered approach: an example of stack of inference engines

This example has shown that a stack of inference engines doesn't represent an efficient, simple, and scalable solution for obtaining a sound and complete reasoning process.

As a unique inference engine is not able to deal with both OWL DL ontologies and SWRL rules, the solution we propose consists of translating OWL DL ontologies into SWRL rules and then utilizing a rule engine.

But, OWL DL syntax constructs can not all be translated into SWRL rules, and so we have limited the expressiveness of OWL DL and, in particular, we have used the DLP OWL language [17], which represents the intersection of a description logic based language with rules. This entails that DLP OWL ontologies can be translated into SWRL rules and vice versa.

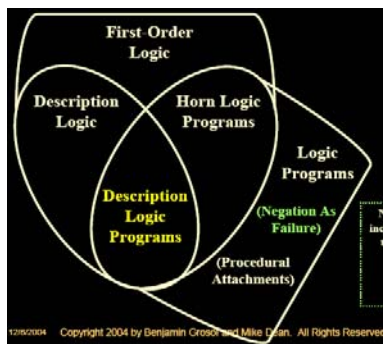


Figure 2 – Description Logic Programs

As a consequence, we are able to translate DLP OWL ontologies into SWRL rules and then to use a unique rule engine to infer and reason in a complete and sound way.

This would represent a simplified method, which would also grant a higher degree of scalability and portability.

It is worth noting that DLP OWL is less expressive than either the ontology or rule languages, but, in many cases, the complete expressiveness of OWL is not needed and a restriction such as DLP OWL is enough.

4. The Ontology Service

To tackle issues as the labeling of anatomical structures in medical images, we need a semantic service able to deal with and integrate the semantic information describing the anatomical knowledge and perform logic and automatic reasoning procedures about it.

As a result, we have designed and developed an Ontology Service that manages DLP OWL ontologies, SWRL rules and RDF statements. In particular, it implements mechanisms to load and validate ontologies and rules respectively from DLP OWL and SWRL files, compose them into a unique and persistent Knowledge Base, load RDF statements into the KB, perform reasoning and logical queries by utilizing the KB.

Our Knowledge Base is composed of two components:

- intensional: a schema defining classes, properties, relations among classes and a set of rules written on the top of these classes and properties (the terminological knowledge, termed the ‘Tbox’)
- extensional: an instantiation of the schema, containing assertions about individuals (the assertional knowledge, termed the ‘Abox’).

Basically, the Tbox is the model of what can be true and is created by loading DLP OWL ontologies and SWRL rules. The Abox is the model of what currently is true and it is composed of RDF statements. Besides the KB implements automated reasoning algorithms to prove ontologies and rules are consistent with the KB and to answer logical queries about the KB.

The Ontology Service has a Web Service interface and its components makes use of the Jena 2 Semantic Web Toolkit [18] and SweetRules tools [19].

Jena 2 Toolkit is a Java framework for building Semantic Web applications. It provides a programmatic environment for RDF, RDFS and OWL, including a rule-based inference engine.

SweetRules is a toolkit for semantic web rules, revolving around the RuleML (Rule Markup Language) and SWRL (Semantic Web Rule Language combining RuleML and OWL) emerging standards for semantic web rules. Its capabilities include translation between a variety of rule and ontology languages, backward and forward inferencing and merging of rulebases/ontologies.

The Ontology Service uses the Jena 2 Toolkit for managing and manipulating ontologies and rules. The Jena 2 Toolkit allows us to deal with OWL ontologies, but it manages rules expressed in a proprietary language and not in SWRL. Besides, the Jena 2 Toolkit has two separate inference engines and it is possible to use them together as a layered but not fully integrated system. These issues are resolved making use of the SweetRules in addition to the Jena 2 Toolkit.

Indeed, when starting up, a set of DLP OWL ontologies and SWRL rules are loaded into the Ontology Server. DLP OWL ontologies are translated into SWRL rules and then are added to the SWRL rules written on the top of these ontologies. Next, both are translated into Jena 2 ones. All these translation processes are realized exploiting the SweetRules tools.

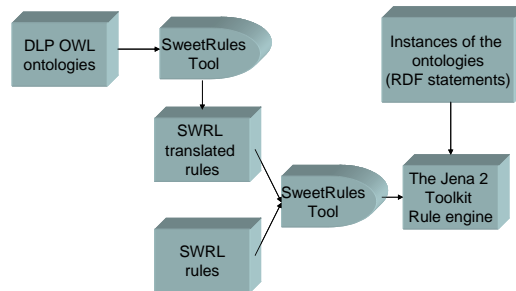


Figure 3 – Functional model

After that, the Ontology Service makes use of the Jena 2 Toolkit rule engine without any difficulty. Indeed, the rules resulting from the translation stage are submitted to the Jena 2 rule engine, which is able to build a unique knowledge base and so to infer and reason about RDF statements in a complete and sound way.

Figure 4 shows the key components of the Ontology Server:

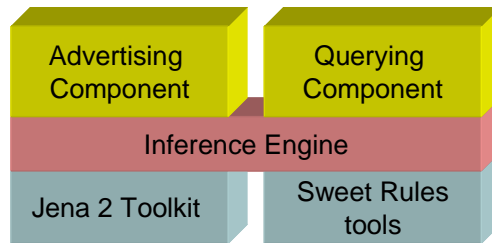


Figure 4 –Architectural model

- The *Inference Engine* is the component in charge of creating a persistent KB by loading DLP OWL ontologies and SWRL rules. It enables to perform reasoning mechanisms and to verify the KB's consistency by using a general purpose rule engine. As a result, this component hides the details of the data structures, logic engine, and KB that we have used and this makes it possible to substitute alternative implementations for them. This component also

provides functionalities for translating DLP OWL ontologies and SWRL rules into Jena 2 rules.

- The *Advertising Component* is the component in charge of loading, updating and removing RDF statements in the KB's Abox. It also allows to load RDF files in the Abox and to add further set of SWRL rules to the Tbox' model.
- The *Query Component* is the component in charge of querying the Abox. Queries can be formatted by specifying only some parts of the requested RDF statements or by using RDQL (RDF Data Query Language) [20], that is a query language for RDF statements.

5. Ontologies and rules for modelling the brain

As an application, we have utilized the set of ontologies and rules achieved by O. Dameron at IDM [21], that model the most important anatomical structures of the brain. In particular, we have represented them by using DLP OWL and SWRL languages and submitted them to the Ontology Service. Besides, we propose an example for giving a flavour of how rules, ontologies and the reasoning process about them might be used for providing a semantic identification of unknown objects.

The brain is composed of two “hemispheres”, separated by a deep fissure called “longitudinal fissure”. Each hemisphere is divided into several “lobes” separated either by fissures named “sulci” or conventional lines. Each lobe is composed of gyri bounded by sulci. A gyrus may be composed of parts, called “pars”, also separated by sulci. There are different types of connections between gyri: conventional separation, pli de passage, and operculus

These concepts are the most important brain anatomical entities, are modelled as ontology classes and organized in the following hierarchy shown in Figure 5:

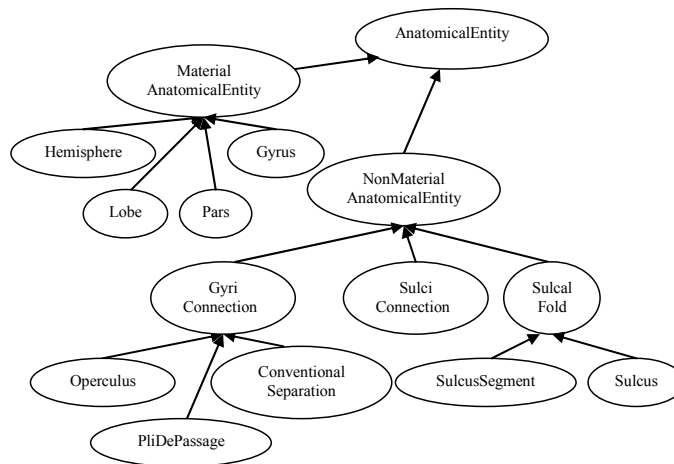


Figure 5 –The ontology concepts

The root entity in the ontology is the primitive class *AnatomicalEntity*, from which stem two subtrees: *MaterialAnatomicalEntity* (MAE) that denoting brain entities made of material opposed to *NonMaterialAnatomicalEntity*. (NMAE). MAEs are composed of several parts, separated by NMAEs, that is sulcal folds or other lines.

MAE includes several subclasses representing the main material anatomical entities: *Hemisphere*, *Gyrus*, *Lobe*, *Pars*.

NMAE includes *SulcalFold* denoting sulcal folds between material entities such as sulci; *GyriConnection*, denoting a connection between two gyri, such as *ConventionalSeparation*, and *SulciConnection*. All siblings classes such as Gyrus, Lobe, Hemisphere, etc. are disjoint.

In addition to the subsumption relationships shown in Figure 5, mereological and topological properties are defined in the ontology. Mereological properties concern part-whole relations between anatomical entities, whereas topological properties concern neighbourhood relations. As instance, mereological properties are *hasAnatomicalPart*, that relates material anatomical entities, and *hasSegment*, that expresses a relation between non material anatomical entities. Instead, an example of topological property is *isMAEBoundedBy*, that expresses that a MAE is bounded by a *SulcalFold* or by a *GyriConnection*.

Besides, rules have been realized to capture the relationships between the mereological and topological properties, the relationships to other domain properties and the semantics of the part-whole relations related to the topological propagation.

An example of rule is the following and it has been used for inferring connected entities from a common *SulciConnection*:

$$isSFConnectedTo(n1,n2) \leftarrow isSFBoundedBy(n1,s) \wedge isSFBoundedBy(n2,s) \wedge SF(n1) \wedge SF(n2) \wedge sulciConnection(s)$$

Next, we show how semantic identification of unknown anatomical entities might be obtained by exploiting the reasoning with the rules and the ontology.

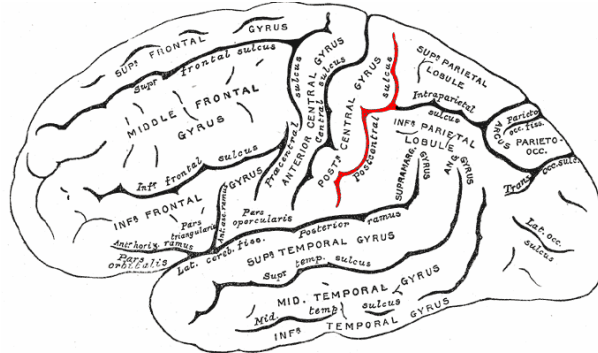


Figure 6 –The brain anatomical structures

Let be *pcs1* the sulcus to be labeled. We want to verify if this sulcus is a PostCentral Sulcus (shown in Figure 6).

The instances of the structures specific to the brain image under study are denoted by mark 0, for example the particular instance of inferiorPostCentralSulcus for the considered image is *ipcs0*, of intraParietalSulcus is *ips0* and so on.

Assume that at the current step of resolution we know that *pcs1* and *spsc0* are sulci, *ipcs0* is the inferiorPostCentralSulcus, *ips0* is the intraParietalSulcus, *spg0* is the superiorParietalGyrus, *sppcg0* is the superiorParsofPostCentralGyrus and *sc* is a sulciConnection.

Besides, the sulcus under study is composed by two segments, that are *spsc0* and *ipcs0*. Both the superiorParsofPostCentralGyrus and superiorParietalGyrus are bounded by the sulcus *spsc0*, whereas the sulciConnection *sc* bounds the two sulcal folds *ips0* and *spsc0*.

These current facts are the following:

- F1: sulcus(pcs1)*
- F2: inferiorPostCentralSulcus(ipcs0)*
- F3: sulcus(spsc0)*
- F4: intraParietalSulcus(ips0)*
- F5: hasSegment(pcs1, spcs0)*
- F6: hasSegment(pcs1, ipcs0)*
- F7: superiorParietalGyrus(spg0)*
- F8: isMAEBoundedBy(spg0, spcs0)*
- F9: superiorParsofPostCentralGyrus(sppcg0)*
- F10: isMAEBoundedBy(sppcg0, spcs0)*
- F11: sulciConnection(sc)*
- F12: isSFBoundedBy(ips0,sc)*
- F13: isSFBoundedBy(spsc0,sc)*

We focalize our attention about the definitions of both the concepts PostCentral Sulcus and superiorPostCentral Sulcus in the ontology:

- O1: PostCentralSulcus ≡ Sulcus □ □ hasSegment inferiorPostCentralSulcus □ □ hasSegment superiorPostCentralSulcus.*
- O2: superiorPostCentralSulcus ≡ Sulcus □ □ bounds superiorParietalGyrus □ □ bounds superiorParsofPostCentralGyrus □ □ isSFConnectedTo intraParietalSulcus.*

and about the following rule:

- R1: isSFConnectedTo(n1,n2) ← isSFBoundedBy(n1,s) ∧ isSFBoundedBy(n2,s) ∧ SF(n1) ∧ SF(n2) ∧ sulciConnection(s).*

In this example, the concepts' definitions O1 and O2 in the ontology and the rule R1 represent our integrated KB, created and managed by the Ontology Service.

Then, we illustrate the reasoning process step by step. First, from the facts F3, F4, F11, F12, F13, the rule R1 infers that the sulcus *spsc0* is connected to the intraParietal Sulcus *ips0*. Then, from this new inferred fact and the facts F3, F7, F8, F9 and F10 (the inverse property is applied in the reasoning process for F8 and F10) the definition

O2 derives that `spcs0` is a superiorPostCentral Sulcus. Finally, from this last inferred fact and facts `F1`, `F2`, `F5`, `F6`, the definition `O1` derives that the sulcus `pcs1` is a PostCentral Sulcus, and, as a result, the semantic identification of the unknown sulcus has been realized.

6. Conclusions and directions for future works

In this paper we show the appliance of an ontologies and rules based approach in the medical imaging.

In particular, we have presented i) a flexible and extendible approach, based on the integration of OWL ontologies and SWRL rules, for modelling all the relevant concepts of the anatomical knowledge; ii) an Ontology Service that exploits OWL ontologies and SWRL rules and performs logic and reasoning mechanisms, in order to face topic issues as the labeling and semantic identification of anatomical structures.

Future works will aim to extend and detail the brain ontology and rules illustrated in the work and provide a model for other anatomical structures of the human body. Besides, we'll aim to consider i) new and more powerful reasoning systems in order to enhance the logic inferences of the Ontology Service, without the limitations of expressiveness due to the use of DLP OWL; ii) the possibility of using also a rule language able to support n-ary predicates and negation in the rule body.

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