MEDICAL SYSTEMS

Clinical Practice Guidelines: a Case Study of combining OWL-S, OWL, and SWRL

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Abstract

As the number of available Web services increases there is a growing demand to realise complex business processes by combining and reusing available Web services. In this context, the Ontology Web Language for Services (OWL-S) can be used to specify semantic types of the input and output data of a Web service and its functionality. This paper uses OWL-S to describe Web services and takes advantage of a XML syntax based on the OWL Web Ontology Language to encode OWL domain ontology fragments and SWRL rule fragments as the inputs and outputs of Web services. The approach presented outlines the use of the OWL's XML presentation syntax to obtain Web services that provide reasoning support and easily deal with facts and rules. To validate the proposal, the research has focused on *Clinical Practice Guidelines* (GLs) related to the biomedical field. This paper highlights the benefits and drawbacks found when applying the approach to obtain Web services that are intended to be used in clinical decision-making and rely on GLs. As an example of use, this paper concentrates on a services-based application for diagnosis and clinical management of Diabetic Retinopathy, where the end-users are health professionals who are not familiarized with Semantic Web technologies.

1. Introduction

A Web service is a set of related functionalities that can be programmatically accessed through the Web[1]. A growing number of Web services are implemented and made available internally in an enterprise or externally for other users to invoke. These Web services can be reused and composed in order to realize larger and more complex business processes. The Web service proposals for description (WSDL[2]), invocation (SOAP[3]) and composition (WS-BPEL formerly known as BPEL4WS[4]) that are most commonly used, lack proper semantic description of services. This makes hard to find appropriate services because a large number of syntactically described services need to be manually interpreted to see if they can perform the desired task. Semantically described Web services make it possible to improve the precision of the search for existing services and to automate the composition of services. Semantic Web Services (SWS) [5] take up on this idea, introducing ontologies to describe, on the one hand, the concepts in the service's domain (e.g. flights and hotels, tourism, e-business), and on the other hand, characteristics of the services themselves (e.g. control flow, data flow) and their relationships to the domain ontologies (via inputs and outputs, preconditions and effects, and so on). Two recent proposals have gained a lot of attentions: 1) the American-based OWL Services (OWL-S) [6] and 2) the European-based Web Services Modelling Language (WSML) [7]. These emerging specifications overlap in some parts and are complementary in other parts. They are both described by low-level lexical notations. WSML uses its own lexical notation, while OWL-S is XML-based.

The low-level XML code is a universal metalanguage for defining markup. It provides a uniform framework, and a set of tools like parsers, for interchange of data and metadata between applications. However, XML does not provide any means of talking about the semantics (meaning) of data. A XML document type definition can be derived from a given ontology as pointed out in [8]. The linkage has the advantage that the XML document structure is grounded on a true semantic basis.

This paper uses OWL-S to describe Web services and takes advantage of the OWL's XML presentation syntax [9] to encode OWL [10] domain ontology fragments and SWRL [11] rule fragments as the inputs and outputs of Web services. The approach presented outlines the use of the OWL's XML presentation syntax [9] to obtain Web services that provide reasoning support and easily deal with facts and rules. To validate the proposal, the research has focused on *Clinical Practice Guidelines* (GLs) related to the biomedical field. This paper highlights the benefits and drawbacks found when applying the approach to obtain Web services that are intended to be used in clinical decision-making and rely on GLs. As an example of use, this paper concentrates on a services-based application for diagnosis and clinical management of Diabetic Retinopathy, where the end-users are health professionals who are not familiarized with Semantic Web technologies.

This paper is organised as follows. Section 2 provides an approach overview. The medical ontology for clinical guidelines is presented is section 3. The details about how to encode OWL domain ontology fragments and SWRL rule fragments as the inputs and outputs of Web services are described in section 4. Section 5 shows a services-based application that health professionals use for diagnosis and clinical management of Diabetic Retinopathy. Conclusions are in section 6.

2. Approach overview

The OWL Web Ontology Language for Services (OWL-S) [6] provides developers with a strong language to describe the properties and capabilities of Web Services in such a way that the descriptions can be interpreted by a computer system in an automated manner. The information provided by an OWL-S description includes: a) ontological description of the inputs required by the service, b) outputs that the service provides, and c) preconditions and postconditions of each invocation. Dynamic use of Web Services is a very complicated task and currently OWL-S is not ready to support the dynamic discovery, composition, and invocation of services. However, OWL-S has tremendous potential, and being able to define the inputs and outputs of a service in terms of an ontology is a huge step towards dynamic discovery, composition, and invocation without user intervention.

A service in OWL-S is described by means of three elements [6]: 1) the *Service Profile* describes what the service does; 2) the *Service Process Model* describes how to use the service; and 3) the *Services Grounding* specifies the details of how to access/invoke a service. The current approach pays special attention to the *Service Process Model* because it includes information about inputs, outputs, preconditions, and results and describes the execution of a Web service in detail by specifying the flow of data and control between the particular methods of a Web service. The execution graph of a *Service Process Model* can be composed using different types of processes and control constructs. OWL-S defines three classes of processes. Atomic processes (`AtomicProcess`) are directly executable and contain no further sub-processes. From the point of view of the caller atomic processes are executed in a single step which corresponds to the invocation of a Web service method. Simple processes (`SimpleProcess`) are not executable. They are used to specify abstract views of concrete processes. Composite processes (`CompositeProcess`) are specified through composition of atomic, simple and composite processes recursively by referring to control constructs (`ControlConstruct`) using the property `ComposeOf`. Control constructs define specific execution orderings on the contained processes.

Protégé 3.2 beta [12] has been chosen as the ontology-design and knowledge acquisition tool to: 1) build ontologies in the Web Ontology Language OWL using the *Protégé-OWL Plugin* and 2) to create OWL-S ontologies using the *OWL-S Editor* that is implemented as a Protégé plugin.

The current implementation considers three Web services:

- *Patient identification service*: is a basic service that provides functionality to gather patient identification data.
- *GL clinical information service*: is a service that provides functionality to find a relevant *Clinical Practice Guideline* and gather the necessary clinical information about the patient.
- *GL recommendation service*: is a service that provides functionality to evaluate the patient condition (establish a diagnosis) and make recommendations about the clinical management based on the evidence available.

Each service considers different kinds of activities. It is necessary to detail each activity and consider if the activity can be related to an atomic process or to a composite process that can be further refined into a combination of atomic processes. Furthermore, it is essential to decide what are the inputs and outputs for each of the atomic processes. Figure 1 shows the inputs and outputs for five atomic processes. The name of each input or output is specified in (bold black) as well as a type is defined for each input and output (in brackets, written in grey). Inputs' and outputs' types are classes/concepts of ontologies that appear in figure 2.

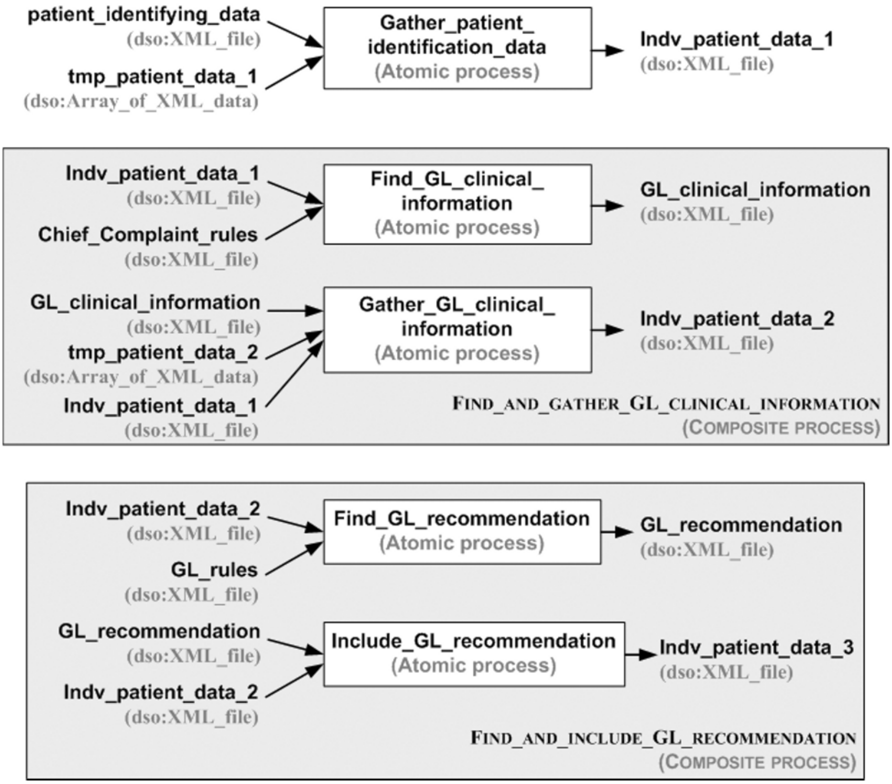

Figure 1 Inputs and outputs of atomic process

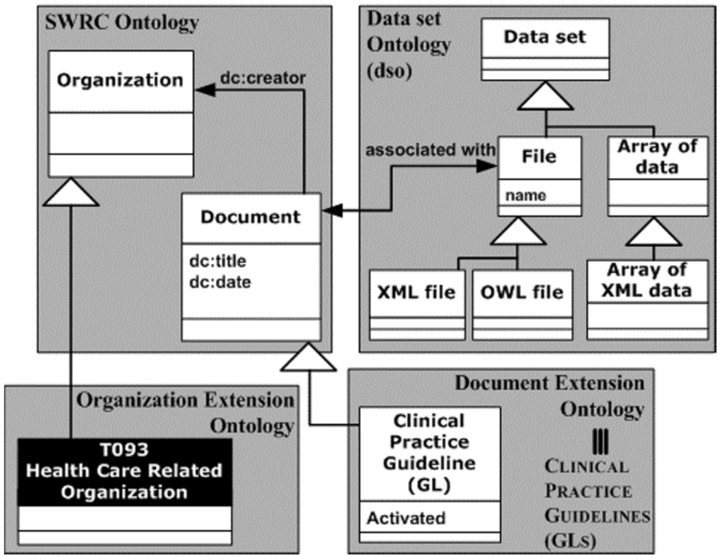

Figure 2 Concepts in the services' domain and their relations to the domain ontology

Figure 2 shows the concepts in the services' domain and their relations to the domain ontology. Four ontologies have been considered:

- The *SWRC ontology* [13] which generically models key entities relevant for typical research communities and the relationships between them. Two main top-level concepts and one relation have been reused. The relation and the two attributes reused belong to the Dublin Core Metadata Element Set [14].
- The *Organization Extension ontology* which is an extension of the *SWRC ontology* and which reuses a *semantic type* from the *Unified Medical Language System* (UMLS) [15].
- The *Document Extension ontology* which is an extension of the *SWRC ontology* to include *Clinical Practice Guidelines* that are "systematically developed statements to assist practitioner and patient decisions about appropriate health care for specific clinical circumstances" [16].
- The *Data Set ontology* which is introduced to linkage with a OWL's XML presentation syntax [9] to encode OWL [10] domain ontology fragments and SWRL [11] rule fragments as the inputs and outputs of Web services.

Figure 3 shows the control flow and data flow of a composite process that is constructed from 3 subprocesses: one atomic subprocess and two composite subprocesses that can be further refined into a combination of two atomic subprocesses (see figure 1). According to figure 3 only one Web service may be needed. However, the current implementation considers three Web services and split the service functionality shown in figure 3 into three.

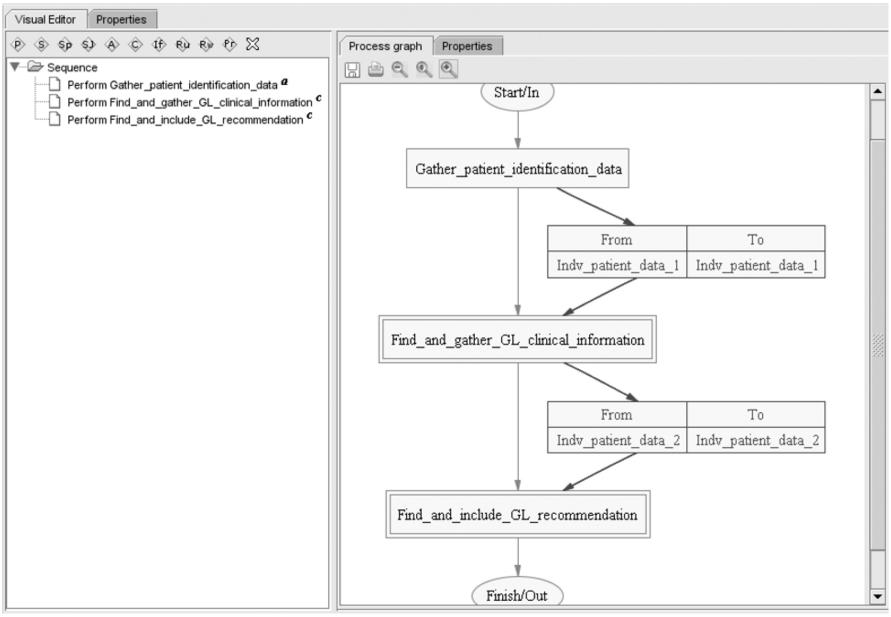

Figure 3 Control flow and data flow

The decision of considering three Web services instead of only one is to validate the approach proposed: Web services that exchange documents with a XML syntax based on a OWL's XML presentation syntax [9] and where a XML document contains OWL [10] domain ontology fragments and/or SWRL [11] rule fragments that are useful to be passed between the services and that may be needed by other components in the same workflow.

The medical ontology for *Clinical Practice Guidelines* (GLs) that is presented in the next section expands the *Document Extension ontology* that appears in figure 2.

3. The medical ontology for clinical guidelines (GLs)

Ontologies range in abstraction from very general concepts that form the foundation for knowledge representation for all domains to concepts that are restricted to specific domains [17]. The *Unified Medical Language System* (UMLS) [15] contains two separate but interconnected knowledge sources, the *Semantic Network* (upper level) and the *Metathesaurus* (lower level). The *Semantic Network* is a high-level representation of the biomedical domain based on *semantic types* under which all the *Metathesaurus* concepts are categorised, and which "may be considered a basic ontology for that domain" [18]. The UMLS *Metathesaurus* is the UMLS knowledge source that represents biomedical concepts derived from a variety of controlled vocabularies, classifications, and other biomedical terminologies, such as collections of terms used in ambulatory care or clinical record systems. *Semantic types* are assigned to concepts based on the intrinsic and functional properties of each concept, and help to distinguish different meanings associated with a single name.

The UMLS can be used to overcome problems caused by discrepancies in different terminologies. However, the UMLS's enormous size and complexity (more than 730 000 concepts in the UMLS *Metathesaurus*) can pose serious comprehension problems for potential users. The research study presented in this paper is aligned with [19], i.e. the cohesive partition of the UMLS *Semantic Network* into collections of *semantic types*. For an effective partitioning of the UMLS *Semantic Network*, the groups of *semantic types* have to be not just uniform in their structure but also cohesive. For a group of *semantic types* to be cohesive, it should have a unique root, i.e. one *semantic type* which all other s*emantic types* in the group are descendants of. The cohesiveness is a result of the fact that each of the *semantic types* in the group is a specialisation of the unique root. Hence, by naming the *semantic-type* group after the root, this name properly reflects the overarching semantics of the group. In [19] 28 *semantic-type* collections of the UMLS *Semantic Network* were identified.

The research approach presented in this paper has reused 6 *semantic-type* collections from the 28 *semantic-type* collections identified in [19]. Figure 4 shows a table that lists the *semantic-type* collections reused in alphabetic order as well as their size (number of *semantic types* in the collection according to [19]) and the *semantic types* reused for each *semantic-type* collection.

Semantic-type Collection	Size	*Semantic Types* in Collection
T190 Anatomical Abnormality	3	T190 Anatomical Abnormality; T020 Acquired Abnormality
T033 Finding	3	T033 Finding; T184 Sign or Symptom; T034 Laboratory or Test Result
T058 Health Care Activity	4	T058 Health Care Activity; T059 Laboratory Procedure; T060 Diagnostic Procedure; T061 Therapeutic or Preventive Procedure
T078 Idea or Concept	14	T078 Idea or Concept; T169 Functional Concept
T092 Organization	4	T092 Organization; T093 Health Care Related Organization
T046 Pathologic Function	6	T046 Pathologic Function; T047 Disease or Syndrome

Figure 4 *Semantic-type* collection list

Clinical Practice Guidelines (GLs) are "systematically developed statements to assist practitioner and patient decisions about appropriate health care for specific clinical circumstances" [16]. Their use in clinical-decision making is intended to improve the outcomes of clinical care. Most *Clinical Practice Guidelines* (GLs) are free texts or simple flow charts. Most of the actions referred to in GLs can be mapped into parts of the UMLS terminology that is associated with *semantic types*. While the UMLS provides its terms with associated *semantic types*, one needs a more elaborate ontology in order to use the latter in *Clinical Practice Guidelines*.

The research study presented in this paper is adhered to a modular ontology design. Existing methodologies and practical ontology development experiences have in common that they start from the identification of the purpose of the ontology and the need for domain knowledge acquisition [20], although they differ in their focus and steps to be taken. In this study, the three basic stages of the knowledge engineering methodology of CommonKADS [21] coupled with a modularised ontology design have been followed:

1. Knowledge Identification: in this stage, several activities have been included: explore all domain information sources in order to elaborate the most complete characterisation of the application domain, and list potential components for reusing. The following knowledge sources have been identified: a) the SWRC ontology [13] which generically models key entities relevant for typical research communities and the relationships between them, b) the UMLS [15] *Semantic Network* where a cohesive partition of the UMLS *Semantic Network* into collections of s*emantic types* was considered, c) the UMLS [15] *Metathesaurus*, and d) some *Clinical Practice Guidelines*.

2. Knowledge Specification: in this second stage the construction of a specification of the domain model has been made. Protégé 3.2 beta [12] has been chosen as the ontology-design and knowledge acquisition tool to build ontologies in the Web Ontology Language OWL [10] using the Protégé-OWL Plugin. Portions of text from *Clinical Practice Guidelines* (GLs) have been manually marked and linked to the *semantic-type* collections that appear in figure 4. The relations established appear in figure 5 that shows the medical ontology that is an extension of the ontological design from figure 2.

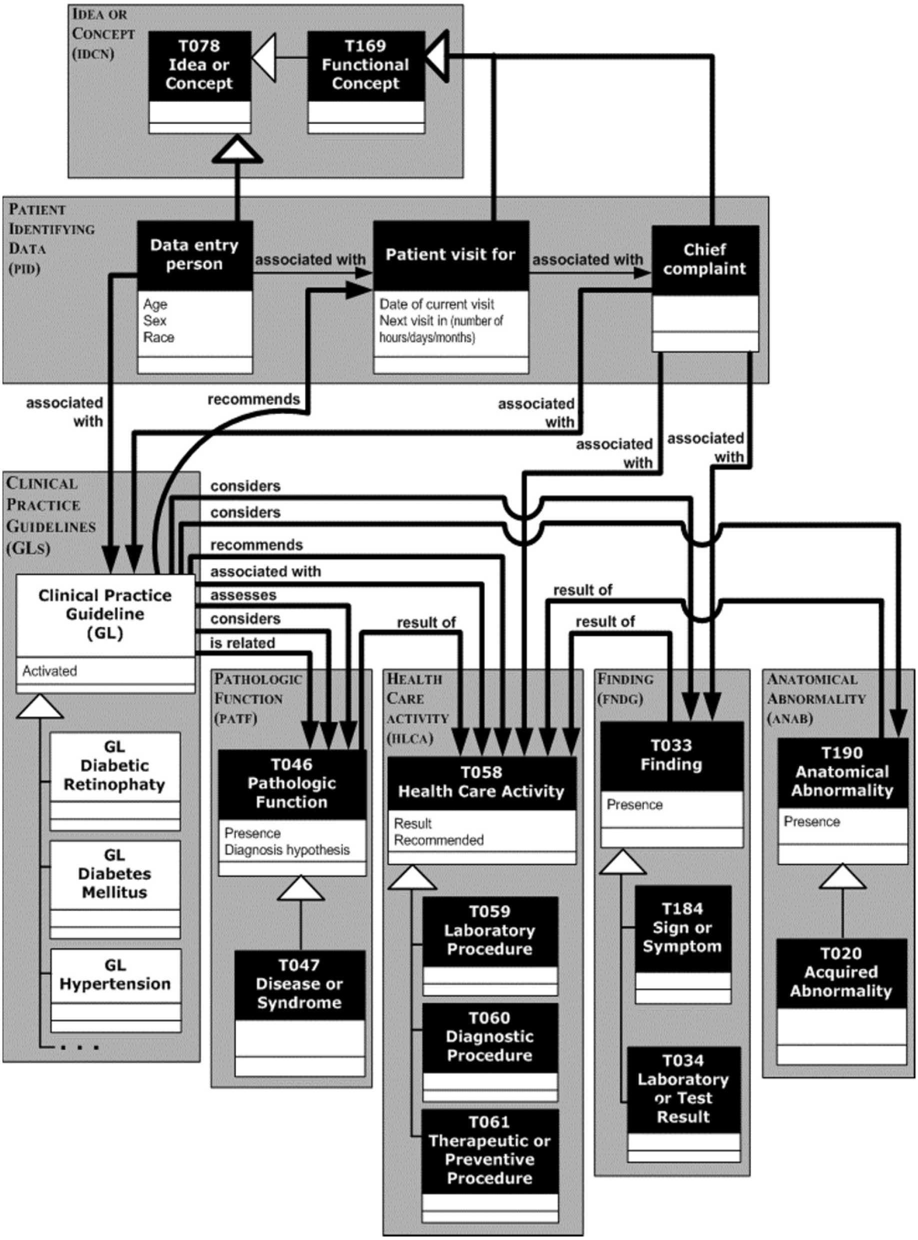

Figure 5 The medical ontology

3. KNOWLEDGE REFINEMENT: in this third stage, the resulting domain model is validated by paper-based simulation, and more concepts from the *UMLS Metathesaurus* are added to the domain model due to their linkage with the *Clinical Practice Guidelines* (GLs) for diagnosis and clinical management of Diabetic Retinopathy which are the GLs that this study focuses on. Figure 6 shows a screenshot of Protégé 3.2 beta during the OWL ontology development; the class GL_Diabetic_Retinophaty is showed.

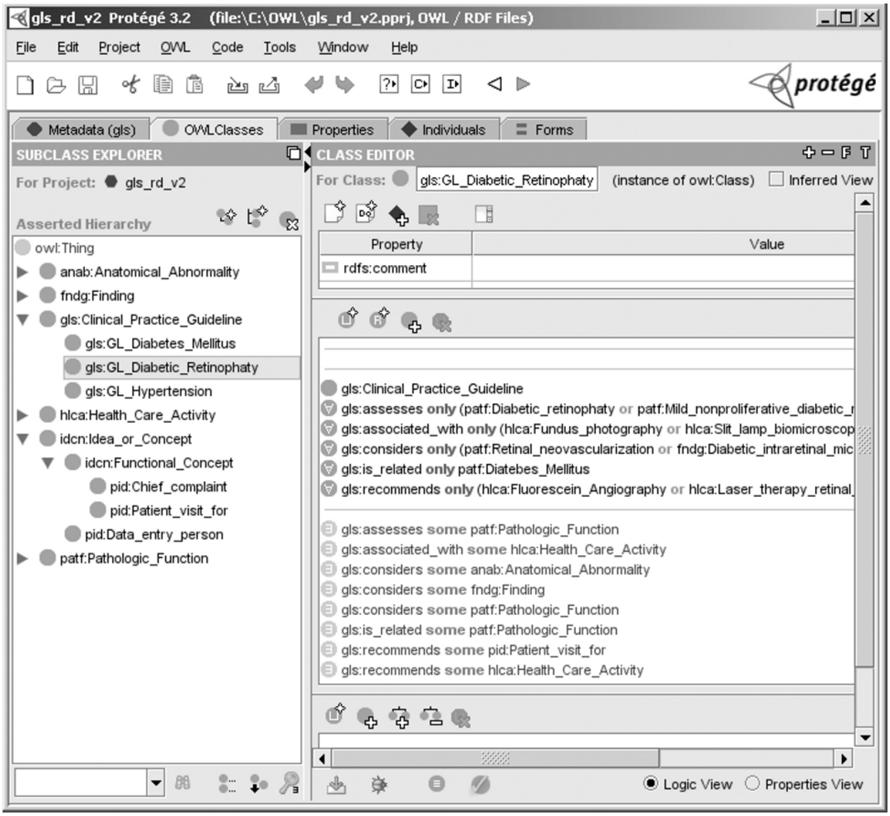

Figure 6 The class GL_Diabetic_Retinophaty in OWL edited with Protégé 3.2 beta

The next section introduces the OWL's XML presentation syntax [9] to encode OWL [10] ontologies into XML syntax and how the proposed XML syntax can be modified to deal with SWRL [11] rules. The OWL's XML presentation syntax used plays pivotal role in the approach proposed: Web services that exchange XML documents and where a XML document contains OWL domain ontology fragments and/or SWRL rule fragments that are useful to be passed between the services and that may be needed by other components in the same workflow.

4. The OWL's XML presentation syntax

Many possible XML encodings could be imagined, but the most obvious solution is to extend the existing OWL Web Ontology Language XML Presentation Syntax [9], which can be straightforwardly modified to deal with SWRL [11]. As pointed out in [22] this has several advantages: 1) arbitrary classes can be used as predicates in rules; 2) rules and ontology axioms can be freely mixed; and 3) the existing XSLT stylesheet [23] can easily be extended to provide a mapping to RDF graphs that extends the OWL RDF/XML exchange syntax.

The owlx namespace prefix should be treated as being bound to http://www.w3.org/2003/05/owl-xml, and is used for the existing OWL's XML syntax. Figures 7 to 9 show examples of the XML presentation syntax for OWL.

```
<owlx:Class owlx:name="Patient_visit_for">
</owlx:Class>
```

Figure 7 The class **Patient_visit_for**

```
<owlx:Class owlx:name="GL_Diabetic_Retinophaty">
 <owlx:ObjectRestriction owlx:property="is_related">
 <owlx:allValuesFrom>
  <owlx:Class owlx:name="#Diatebes_Mellitus"/>
 </owlx:allValuesFrom>
 </owlx:ObjectRestriction>
...
</owlx:Class>
```

Figure 8 The ObjectRestriction **is_related**

```
<owlx:DatatypeProperty owlx:name="Result">
 <owlx:domain>
 <owlx:UnionOf>
  <owlx:Class owlx:name="#Fundus_photography"/>
  <owlx:Class owlx:name="#Slit_lamp_biomicroscopy"/>
  <owlx:Class owlx:name="#Ophthalmoscopy"/>
 </owlx:UnionOf>
 </owlx:domain>
 <owlx:range>
 <owlx:OneOf>
  <owlx:DataValue owlx:datatype="&xsd;string">normal</owlx:DataValue>
  <owlx:DataValue owlx:datatype="&xsd;string">findings_and_others</owlx:DataValue>
 </owlx:OneOf>
 </owlx:range>
</owlx:DatatypeProperty>
```

Figure 9 The DatatypeProperty **Result**

The OWL's XML presentation syntax [9] is extended to add the relevant syntax for variables and rules. As in [22] the unspecified owlr namespace prefix is used for the newly introduced syntax. Figure 10 shows an example of the XML presentation syntax for a rule. Rule axioms have an antecedent (owlr:antecedent) component and a consequent (owlr:consequent) component. The antecedent and consequent of a rule are both lists of atoms and are read as the conjunction of the component atoms. Atoms can be formed from unary predicates (classes), binary predicates (properties), equalities or inequalities.

```
<owlr:Rule>
<owlr:antecedent>
 <owlr:classAtom>
  <owlx:Class owlx:name="visual_loss_in_diabetic_patient"/>
  <owlr:Variable owlr:name="x1"/>
 </owlr:classAtom>
  <owlr:datavaluedPropertyAtom owlr:property="Presence">
   <owlr:Variable owlr:name="x1"/>
   <owlx:DataValue owlx:datatype="&xsd;string">yes</owlx:DataValue>
  </owlr:datavaluedPropertyAtom>
</owlr:antecedent>
<owlr:consequent>
 <owlr:classAtom>
  <owlx:Class owlx:name="GL_Diabetic_Retinophaty"/>
  <owlr:Variable owlr:name="x2"/>
 </owlr:classAtom>
 <owlr:datavaluedPropertyAtom owlr:property="Activated">
  <owlr:Variable owlr:name="x2"/>
  <owlx:DataValue owlx:datatype="&xsd;string">yes</owlx:DataValue>
 </owlr:datavaluedPropertyAtom>
</owlr:consequent>
</owlr:Rule>
```

Figure 10 The XML presentation syntax for a rule

The next section shows a services-based application that health professionals use for diagnosis and clinical management of Diabetic Retinopathy, and where examples of individual axioms (also called "facts") - based on the OWL's XML syntax presented in this section - are provided. Facts are matched against rules (like the one displayed in figure 10) as the bases to provide reasoning support.

5. Applying the approach: Diabetic Retinopathy

The user interface of the services-based application that health professionals use for diagnosis and clinical management of Diabetic Retinopathy is comprised into three main Web pages that respectively interact with each of the Web services described in section 2. Figures 11 and 13 show the above-mentioned main Web pages, where the main asset of the Web-based Graphical User Interface (GUI) obtained is to be ease-to-use by health professionals who are not familiarized with Semantic Web technologies. As a result of the interaction with health professionals, documents with a XML syntax based on a OWL's XML presentation syntax [9] are obtained and passed between the Web services. Those XML documents contain individual axioms (also called "facts") that contain the clinical data about a patient – see figure 12 - and are matched against rules – see figure 10-.

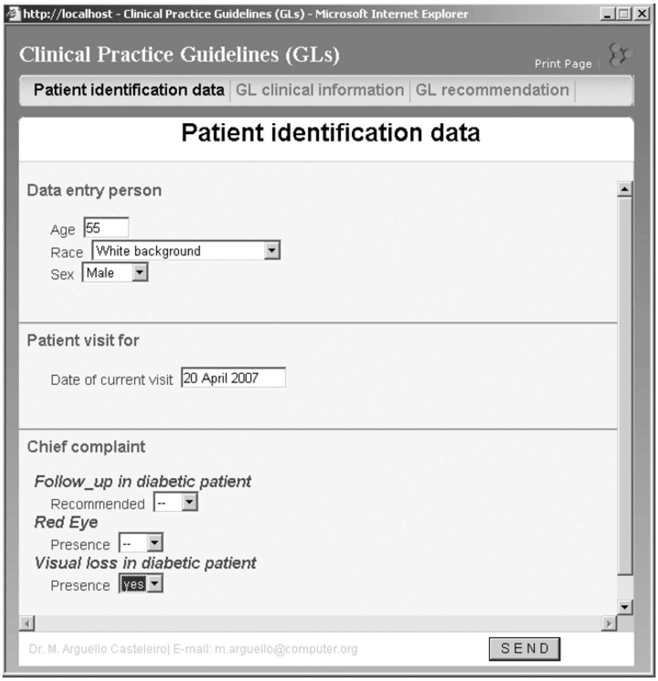

Figure 11 Patient identification data

The GUI showed in figure 11 and 13 is on-fly generated by means of Ajax [24], shorthand for Asynchronous JavaScript and XML, as the Web client development technique, where the existing XSLT stylesheet [23] and XML documents derived from the OWL XML syntax are interpreted by JavaScript functions.

```
<owlx:Individual>
 <owlx:type owlx:name="Data_entry_person">
 <owlx:DataPropertyValue owlx:property="Age">
  <owlx:DataValue owlx:datatype="&xsd;positiveInteger">55</owlx:DataValue>
 </owlx:DataPropertyValue>
 <owlx:DataPropertyValue owlx:property="Race">
  <owlx:DataValue owlx:datatype="&xsd;string">White_background</owlx:DataValue>
 </owlx:DataPropertyValue>
 <owlx:DataPropertyValue owlx:property="Sex">
  <owlx:DataValue owlx:datatype="&xsd;string">Male</owlx:DataValue>
 </owlx:DataPropertyValue>
</owlx:Individual>

<owlx:Individual>
 <owlx:type owlx:name="Patient_visit_for">
 <owlx:DataPropertyValue owlx:property="Date_of_current_visit">
  <owlx:DataValue owlx:datatype="&xsd;string">20_April_2007</owlx:DataValue>
 </owlx:DataPropertyValue>
</owlx:Individual>

<owlx:Individual>
 <owlx:type owlx:name="Chief_complaint">
 <owlx:ObjectPropertyValue owlx:property="associated_with">
 <owlx:Individual>
  <owlx:type owlx:name="Visual_loss_in_diabetic_patient">
  <owlx:DataPropertyValue owlx:property="Presence">
   <owlx:DataValue owlx:datatype="&xsd;string">yes</owlx:DataValue>
  </owlx:DataPropertyValue>
 </owlx:Individual>
 </owlx:ObjectPropertyValue>
</owlx:Individual>
```

Figure 12 Individual axioms (also called "facts")

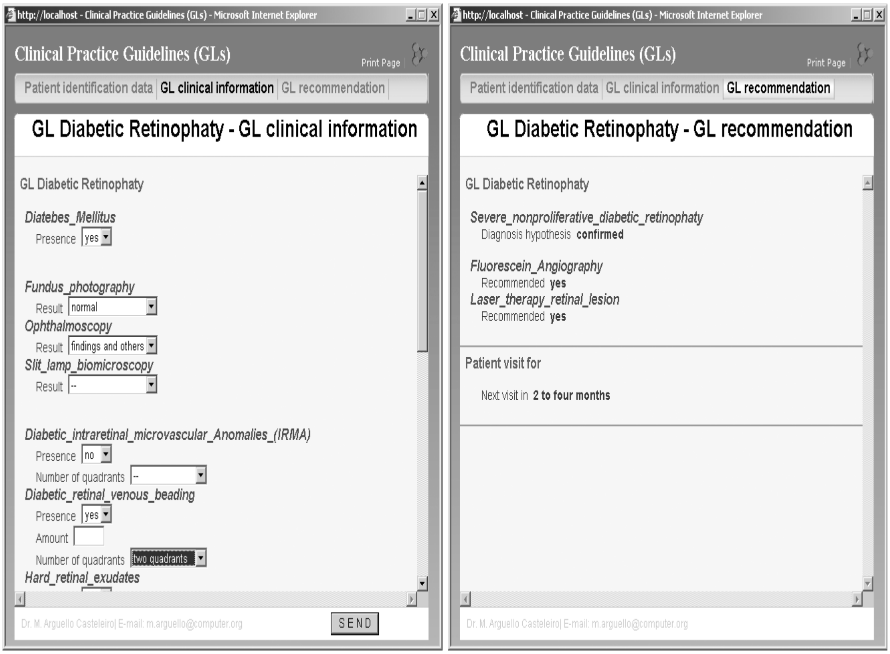

Figure 13 GL Diabetic Retinophaty

The interpretation of figures 11 to 13 is the following: as a result of the interaction between a health professional and the GUI showed in figure 11, the *Patient identification service* (see section 2) will create a XML document (`Indv_patient_data_1`) that contains individual axioms about a patient and that will pass to the *GL clinical information service* the XML document generated when the health professional presses the button SEND (figure 11). The *GL clinical information service* will match the XML document (`Indv_patient_data_1`) against rules (`Chief_Complaint_rules`) like the one showed in figure 10 and based on the matching process performed a *Clinical Practice Guideline* (GL) is selected. As a result of the interaction between a health professional and the GUI showed in figure 13 (left), the *GL clinical information service* will create a XML document (`Indv_patient_data_2`) that contains individual axioms about a patient and that will pass to the *GL recommendation service* the XML document generated when the health professional presses the button SEND (figure 13 left). The *GL recommendation service* will match the XML document (`Indv_patient_data_2`) against rules (`GL_rules`) and based on the matching process performed, the diagnosis and clinical management obtained (consequent of the activated rule) will be stored in a XML document (`Indv_patient_data_3`) and will be showed to the health professional (figure 13 right).

6. Conclusions

The case study presented here provides strong evidence of the advantages of using Semantic Web technologies. The proposal is based on Web services that exchange documents with a XML syntax based on a OWL's XML presentation syntax [9] and where a XML document contains OWL [10] domain ontology fragments and/or SWRL [11] rule fragments that are useful to be passed between the services and that may be needed by other components in the same workflow.

The approach presented outlines the use of the OWL's XML presentation syntax [9] to obtain Web services that provide reasoning support and easily deal with facts and rules. The proposal has been validated by focusing on *Clinical Practice Guidelines* (GLs) related to the biomedical field. As an example of use, this paper concentrates on a services-based application for diagnosis and clinical management of Diabetic Retinopathy, where the end-users are health professionals who are not familiarized with Semantic Web technologies.

The current study has found that OWL has expressive limitations. As pointed out in [22] these restrictions can be onerous in some application domains, for example in describing web services, where it may be necessary to relate inputs and outputs of composite processes to the inputs and outputs of their component processes. A way to overcome some of the expressiveness restrictions of OWL would be to extend it with some form of "rules languages".

The services-based application presented works well with small examples (see sections 4 and 5), and it successes with up to 50 SWRL rules and facts. However, the effectiveness of the approach presented must be open to question with larger number of SWRL rules and facts.

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