Development of a Simple Ontology Definition Language (SOntoDL) and Its Application to a Medical Information Service on the World Wide Web

Rolf GRÜTTER and Claus EIKEMEIER
Institute for Media and Communications Management, University of St. Gallen, Blumenbergplatz 9, CH-9000 St. Gallen, Switzerland

Abstract. It is the vision of the protagonists of the Semantic Web to achieve a set of connected applications for data on the World Wide Web (WWW) in such a way as to form a consistent logical web of data. Therefore, the Semantic Web approach develops languages for expressing information in a machine-processable form. Particularly, the Resource Description Framework (RDF) and RDF Schema (RDFS) are considered as the logical foundations for the implementation of the Semantic Web. This paper documents the development of a Simple Ontology Definition Language (SOntoDL). The development is part of a project aimed at the implementation of an ontology-based semantic navigation through a glossary of an evidence-based medical information service on the WWW. The latest version of SOntoDL is integrated with the RDF/RDFS framework thereby providing for the foundation of a Semantic Web of evidence-based medical information.

1. Introduction

One of the hot topics in medical informatics is the handling of medical terminologies. Thereby, the challenge is twofold. Firstly, the conflicting targets of a concept representation that is close to the real-world and, at the same time, easy to handle by healthcare professionals, e.g., for the coding of diagnoses or indexing of medical subjects, must be solved [17]. Secondly, as existing terminologies like MeSH (Medical Subject Headings), SNOMED (Standardized NOmenclature of MEDicine [19]), and UMLS (Unified Medical Language System [18]) show, it is not evident that medical terminologies provide enough conceptual expressiveness to allow for an easy handling from a formal-logical point of view. Particularly, the mentioned terminologies do not distinguish between generic (IS-A) and partitive (PART-OF) relations [15]. However, a formal-logical foundation is pivotal if terminologies should be processable by computers.

Whereas in closed healthcare settings knowledge engineering, i.e., the discipline that deals with the formal representation of terminologies, has quite a tradition, the advent of the World Wide Web (WWW) brought about an increase in its scale and scope. The increased scale refers to the spread of the WWW which is literally world-wide. The increase in scope refers to the extension of knowledge engineering methods to non-medical domains, such as corporations. This trend is reflected by the recent Semantic Web initiative [1].

It is the vision of the protagonists of the Semantic Web to achieve „a set of connected applications for data on the WWW in such a way as to form a consistent logical web of data“ ([1], p. 1). Therefore, the Semantic Web approach develops languages for expressing information in a machine-processable form. Particularly, the Resource Description Framework, RDF [14] and RDF Schema, RDFS [5] are considered as the logical foundations for the implementation of the Semantic Web.

This paper documents the development of a Simple Ontology Definition Language (SOntoDL). The development is part of a project aimed at the implementation of an ontology-based semantic navigation through a glossary of an evidence-based medical information service on the WWW. The terminology on which the ontology is based refers to the field of clinical epidemiology. The latest version of SOntoDL is integrated with the RDF/RDFS framework thereby providing for the foundation of a Semantic Web of evidence-based medical information.

2. Application Domain: The Evimed Project

The Evimed project (www.evimed.ch) was initiated in April 1998. It aims at providing general practitioners in the German speaking countries with relevant and reliable information for daily practice, thereby supporting the practitioners in making appropriate medical decisions. To achieve this, a group of physicians who are trained in evidence-based medicine systematically reviews published studies with respect to practical relevance and trustworthiness. These reviews are published together with the links to the original articles in the Journal Club of Evimed (Figure 1).

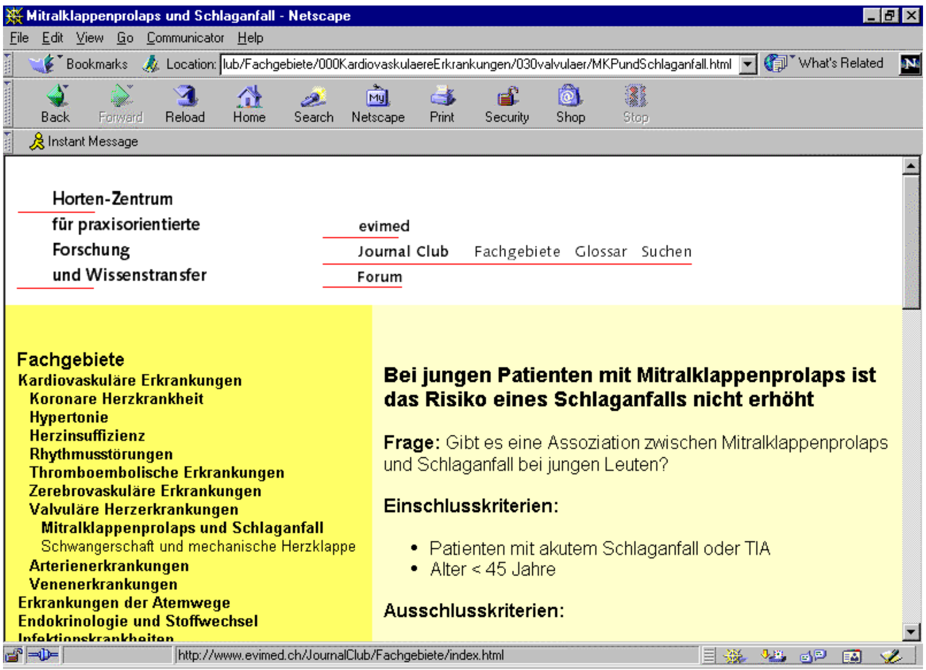

Figure 1. Journal club of the Evimed website

The Journal Club is the core of the Evimed website. Currently (i.e., in July 2001) it stores 400 reviews of selected articles published in various biomedical journals. The reviews are categorized according to 22 specialties and can either be browsed or be accessed via a (syntactic) search engine. The website includes a separate section with articles on the subject of Evidence-Based Medicine (EBM). Evimed also includes a glossary with currently 25 definitions of EBM-specific terms from the field of clinical epidemiology. Further services include free access to Medline, links to literature and other EBM-related sources (e.g., a calendar of events related to further education in EBM), and a guest book offering the possibility to post comments and to subscribe for a free newsletter. Taking into account the objection of practitioners who do not feel comfortable with translating foreign languages, all text of the website is in German.

As mentioned in the Introduction, the development of SOntoDL is part of a project aimed at the implementation of an ontology-based semantic navigation through the glossary of Evimed. The ultimate vision of the project is to effectively meet the information needs of the practitioners who are often not familiar with terms from the domain of clinical epidemiology.

3. Methods: The Concept of the Ontology

The applied conceptual framework refers to the concept of the ontology as defined by Gruber [8]. According to Gruber, an ontology is a specification of a conceptualization, i.e., a formal description of the concepts and their relations for a „universe of discourse“. The universe of discourse refers to the set of objects which can be represented in order to represent the (propositional) knowledge of a domain. This set of objects and the describable relations among them are reflected in a representational vocabulary. In an ontology, definitions associate the names of objects in the universe of discourse with human-readable text, describing what the names mean, and formal axioms constrain the interpretation and well-formed use of the ontology. In short, an ontology consists of the triple (vocabulary, definitions, axioms). Formally, an ontology is the statement of a logical theory.

This conceptual framework has been applied in order to define a simple ontology definition language. The three major steps of the development process are described in the following section.

4. Results: Towards A Simple Ontology Definition Language

The development process of SOntoDL was guided by the following requirements:

(1) The language format must allow for an easy integration with the WWW.

(2) Since the knowledge base of the given domain evolves with time, the language must allow for an easy extension of the ontology without requiring the modification of the program that processes the ontology (i.e., the inference engine).

(3) The language should be applicable not only to the given application domain but also to additional domains.

(4) The language should support the physically dissociated (i.e., distributed) maintenance of representational vocabulary and human-readable definitions, as the two do not require the same frequency of updates and the respective editors may not be the same.

(5) The language should allow for the representation of complex, non-hierarchical knowledge structures.
(6) The language should allow to distinguish between generic (IS-A) and partitive (PART-OF) relations thereby providing enough conceptual expressiveness to support an easy handling of the representational vocabulary from a formal-logical point of view.
(7) The language should be integrated in a common logical framework for connected applications on the WWW thereby taking advantage of a range of tools (hopefully) being developed.

Not all of these requirements were specified prior to the development of SOntoDL. Instead, the list was completed during the development process. The latter can be structured according to the three basic approaches, i.e., intuitive approach, generic approach, and integration into the RDF/RDFS framework.

4.1. Intuitive Approach

The intuitive approach builds on a simple mapping of the paper-based concept hierarchy as provided by the healthcare professionals of Evimed onto an Extensible Markup Language (XML) document tree, whereby the names of the concepts are represented as tag names (Figure 2). By using the emerging WWW standard XML as the core technology for SOntoDL the requirement (1) is met.

```
<EBM_Ontologie>
      ...
      <Stat_Kennzahlen>
            <Kennzahlen_Therapie>
                  <Ereignis>
                        <Ereignisrate>
                              <EER>
                                    <CER>
                                          <ARR/>
                                          <RRR>
                                                <NNT/>
                                          </RRR>
                                    </CER>
                              </EER>
                        </Ereignisrate>
                  </Ereignis>
            </Kennzahlen_Therapie>
            ...
      </Stat_Kennzahlen>
</EBM_Ontologie>
```

Figure 2. Intuitive approach to a simple ontology definition language. Note that the XML representation is incomplete and not ready for a processing by a software program. For complete XML documents cf. Figures 4 and 6.

The intuitive approach yields the advantage of comprehensiveness and easy readability by men. As a main disadvantage the representational vocabulary is fixed and cannot be extended without modifying the Document Type Definition (DTD) (not shown). In other words, it makes no sense to define a DTD at all (the definition of a DTD for a class of XML documents is optional). The intuitive approach defines a particular ontology language, namely for the domain of evidence-based medicine, rather than an ontology *definition* language and cannot be applied to other domains. Due to its obvious weakness, the intuitive approach – while initially intended – has not been implemented.

4.2. Generic Approach

The generic approach defines an ontology definition language by an XML DTD (Figure 3). Different from the intuitive approach, most of the element types denote generic concepts such as `item`, `identifier`, and `description`. „Generic“ means in this context that the concepts do not refer to a particular application domain (e.g., EBM). In addition, since the `ontology` includes zero, one or more `item` (denoted by the symbol `*`) and each `item`, in turn, includes zero, one or more `item`, the ontology can be arbitrarily extended. This way, the requirements (2) and (3) are met in addition to (1).

```
<!ELEMENT ontology (item*)>
<!ATTLIST ontology version CDATA  #FIXED "1.1">
<!ELEMENT item (identifier+,description, item*)>
<!ATTLIST item myID NMTOKEN #IMPLIED>
<!ELEMENT identifier (#PCDATA)>
<!ATTLIST identifier language (english | german | french) #REQUIRED>
<!ATTLIST identifier format (short | long) #REQUIRED>
<!ELEMENT description (#PCDATA)>
<!ATTLIST description language (english | german | french) #REQUIRED>
<!ATTLIST description implementation (html|url) #REQUIRED>
```

Figure 3. Generic approach to a simple ontology definition language

The generic version of SOntoDL has been applied in order to implement a prototype of an ontology-based semantic navigation through the glossary of an evidence-based medical information service on the WWW [9]. At this small scale, it proved to be well defined. However, when the language was applied to a different domain, i.e., the NetAcademy (www.netacademy.org), its limitations became clear: Since the ontology is implemented as a single XML file, the document soon gets very large and hard to handle. Worse, all updates, be it of the representational vocabulary (element type `identifier`) or of the human-readable definitions (element type `description`), must be made in this central document. In order to anticipate these disadvantages (and to meet requirement 4), the option to implement the description as an URI reference [2] instead of a CDATA section was added to the language (attribute `implementation`). Even though this extension of SOntoDL is marginal, it has a major impact on its applicability. Particularly, it is now possible to integrate external resources into the domain ontology. In other words, external resources can be annotated with a representational vocabulary by simple URI references.

4.3. Integration into the RDF/RDFS Framework

The generic version of SOntoDL takes advantage of the intrinsic structuring capabilities of XML documents, i.e., the representational vocabulary is implemented as a hierarchy of items together with their identifiers and descriptions, whereby each item corresponds to a node of the document tree. This pragmatic approach has been chosen since the Evimed vocabulary was provided as a hierarchy of concepts. The disadvantage of this approach is its limitation to a (mono-) hierarchical representation and the inability to represent more complex knowledge structures. In order to overcome this limitation, SOntoDL is integrated into the RDF/RDFS framework, that is the potential de facto standard for connected applications on the WWW. Thus, along with this third development step, the requirements (5), (6) (see below), and (7) are met in addition to (1) - (4).

```
<?xml version='1.0' encoding='ISO-8859-1'?>
<rdf:RDF
 xmlns:rdf="http://www.w3.org/1999/02/22-rdf-syntax-ns#"
 xmlns:rdfs="http://www.w3.org/2000/01/rdf-schema#"
 xmlns:o="http://ontoserver.aifb.uni-karlsruhe.de/schema/rdf">
  <rdf:Description ID="Item">
    <rdf:type resource="http://www.w3.org/2000/01/rdf-schema#Class"/>
    <rdfs:subClassOf resource="http://www.w3.org/2000/01/rdf-
     schema#Resource"/>
  </rdf:Description>
  <rdf:Description ID="childOf">
    <rdf:type resource="http://ontoserver.aifb.uni-
     karlsruhe.de/schema/rdf#Irreflexive"/>
    <rdf:type resource="http://ontoserver.aifb.uni-
     karlsruhe.de/schema/rdf#Asymmetric"/>
    <rdfs:domain rdf:resource="#Item"/>
    <rdfs:range rdf:resource="#Item"/>
    <o:isInverseRelationOf rdf:resource="#parentOf"/>
  </rdf:Description>
  <rdf:Description ID="siblingOf">
    <rdf:type resource="http://ontoserver.aifb.uni-
     karlsruhe.de/schema/rdf#Irreflexive"/>
    <rdf:type resource="http://ontoserver.aifb.uni-
     karlsruhe.de/schema/rdf#Symmetric"/>
    <rdf:type resource="http://ontoserver.aifb.uni-
     karlsruhe.de/schema/rdf#Transitive"/>
    <rdfs:domain rdf:resource="#Item"/>
    <rdfs:range rdf:resource="#Item"/>
  </rdf:Description>
  <rdf:Description ID="parentOf">
    <rdf:type resource="http://ontoserver.aifb.uni-
     karlsruhe.de/schema/rdf#Irreflexive"/>
    <rdf:type resource="http://ontoserver.aifb.uni-
     karlsruhe.de/schema/rdf#Asymmetric"/>
    <rdfs:domain rdf:resource="#Item"/>
    <rdfs:range rdf:resource="#Item"/>
  </rdf:Description>
</rdf:RDF>
```

Figure 4. Integration of SOntoDL into the RDF/RDFS framework

In view of its conception, the generic approach to SOntoDL is closely related to RDF/RDFS, and a re-definition mainly required the effort to become familiar with RDF/RDFS. The concept corresponding to the XML DTD is the RDF *Schema*. Therefore, SOntoDL is re-defined as an application-specific extension to RDFS (Figure 4). In addition to the generic namespaces `rdf` and `rdfs` (referring to the RDF Schema), the namespace `o` [16] is used. The latter refers to a schema that provides an ontology meta-layer for the representation of axioms. These can be used to type relations (i.e., properties in terms of RDF/RDFS) thereby providing the basis for the implementation of integrity constraints for the ontology. While the re-definition of the so far latest version of SOntoDL has been completed, it has only been partially applied to the EBM domain in order to test its applicability. Figure 5 shows the Directed Labeled Graph (DLG) representation of a sample ontology item defined by SOntoDL.

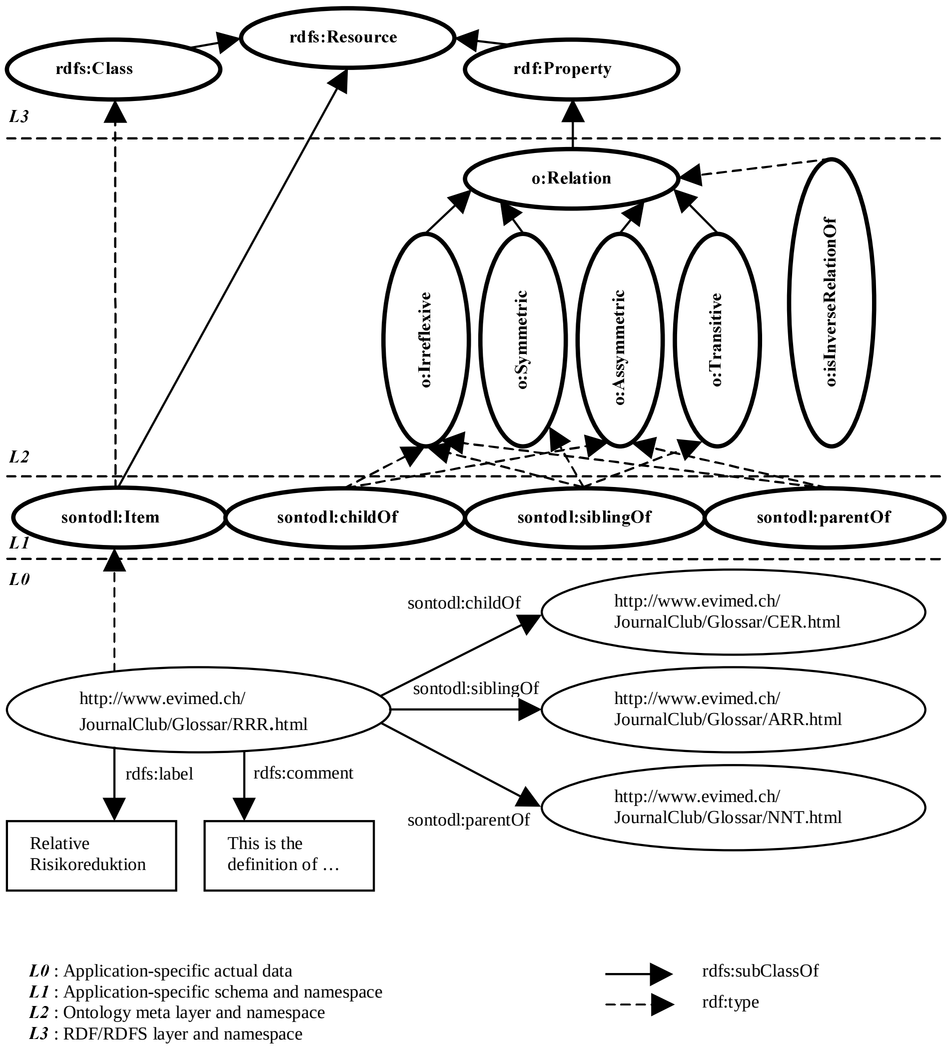

Figure 5. DLG representation of an Evimed ontology item defined by SOntoDL. Note that the `sontodl` namespace refers to SOntoDL which is defined as an application-specific extension to RDFS as shown in Figure 4. The resources on the application-specific actual data layer refer to four glossary items of the Evimed website. The serialization of the DLG representation in RDF syntax is shown in Figure 6.

```
<?xml version='1.0' encoding='ISO-8859-1'?>
<rdf:RDF
 xmlns:rdf="http://www.w3.org/1999/02/22-rdf-syntax-ns#"
 xmlns:rdfs="http://www.w3.org/2000/01/rdf-schema#"
 xmlns:dc="http://dublincore.org/documents/1999/07/02/dces/"
 xmlns:odoc="http://ontoserver.aifb.uni-karlsruhe.de/schema/ontodoc"
 xmlns:o="http://ontoserver.aifb.uni-karlsruhe.de/schema/rdf"
 xmlns:sontodl="http://.../schema/sontodl">
  <rdf:Description about="">
    <dc:Title>Evimed Ontology</dc:Title>
    <dc:creator>
      <rdf:Bag>
        <rdf:li>Rolf Gruetter</rdf:li>
        <rdf:li>Claus Eikemeier</rdf:li>
      </rdf:Bag>
    </dc:creator>
    <dc:date>2000-10-25</dc:date>
    <dc:format>text/xml</dc:format>
    <dc:description>An ontology on evidence-based medicine.</dc:description>
    <dc:subject>Ontology, Evidence-based medicine</dc:subject>
    <odoc:url>http://...</odoc:url>
    <odoc:version>2.0</odoc:version>
    <odoc:last_modification>2000-10-25</odoc:last_modification>
  </rdf:Description>
  <rdf:Description about="http://www.evimed.ch/JournalClub/Glossar/RRR.html">
    <rdf:type resource="http://.../schema/sontodl#Item"/>
    <rdfs:label xml:lang="de">Relative Risikoreduktion</rdfs:label>
    <rdfs:comment xml:lang="en">This is the definition of the labeled concept
     by Evimed.</rdfs:comment>
    <sontodl:childOf
     resource="http://www.evimed.ch/JournalClub/Glossar/CER.html"/>
    <sontodl:siblingOf
     resource="http://www.evimed.ch/JournalClub/Glossar/ARR.html"/>
    <sontodl:parentOf
     resource="http://www.evimed.ch/JournalClub/Glossar/NNT.html"/>
  </rdf:Description>
</rdf:RDF>
```

Figure 6. Serialization of the DLG representation of an Evimed ontology item defined by SOntoDL in RDF syntax. Note that additional schemata, referred to by the namespaces `dc` and `odoc`, are used in order to represent ontology meta-data.

5. Discussion

There is quite a number of related approaches to ontology definition languages (cf. Section 6). Nevertheless, the presented approach follows its own conception. The reason therefore is that the need for an ontology definition language arose during a particular project in a particular application domain (cf. Section 2). As a consequence, initially a pragmatic approach was chosen which was step by step refined as reported in Section 4. The core of the initial conception was to keep the language as simple as possible and to avoid unnecessary overkill (therefore, an unreflected application of one of the languages mentioned in Section 6 was not considered).

During the project, this conception was partially weakened, primarily in favor of a more general applicability.

As mentioned, the latest version of SOntoDL resulting from the integration into the RDF/RDFS framework unfolds its full potential only if increasingly tools are available that support RDF/RDFS. In case another technology for the implementation of the Semantic Web, such as Topic Maps [3] outsmarts RDF/RDFS, SOntoDL has to be adapted.

6. Related Work

SHOE (Simple HTML Ontology Extensions) provides distributed ontologies consisting of categories and relationship rules [10]. Thereby, the categories provide for the classification of instances. They are organized hierarchically and support multiple inheritance. The relationship rules are implemented as Horn clauses. The instances (i.e., individual constants in terms of Horn-rules) are represented as URLs/URIs. This is similar to the approach as presented in this chapter, where the human-readable definitions, as part of the ontology, are likewise represented by URI references. SHOE was originally specified in SGML (as is HTML) (before the definition of XML) but is meanwhile also specified as an XML DTD.

XOL (XML-based Ontology Exchange Language) is a language for specifying and exchanging ontologies [13]. XOL is specified in an XML-based syntax (kernel DTD). It uses a frame-based semantic model, i.e., OKBC-Lite. An XOL file consists of a module-header definition and one or more class, slot and individual definitions. The module-header definition provides meta-information of the ontology, such as the name and version. The class definitions provide the classes and subclasses of the defined individuals. The slot definitions are strings that encode the official names of the entities. Each slot definition refers to a class name. The individual definitions provide the names, documentations, instance-of information, and slot-values of the defined individuals. As a disadvantage, XOL does not re-use the core semantics of RDF/RDFS. Hence, „pure“ RDF/RDFS applications cannot process even the core object-model definitions.

The Ontobroker application answers queries based on a facts base and an ontology base [7]. The facts base stores instance information („values“ in terms of the query interface) which is extracted from annotated HTML pages, for instance, of a corporate Intranet, which is different from the hereby presented approach, where the HTML pages, i.e., the resources, are externally annotaded by RDF descriptions. The ontology base stores a set of ontologies. Each ontology includes a concept hierarchy („classes“), a set of slot definitions („attributes“), and a set of rules. The rules implement integrity constraints for the ontology. Ontologies are defined as F-logic statements.

An approach to representing ontologies in RDF/RDFS, similar to [16] (and to the latest version of SOntoDL), is pursued with OIL [12, 6]. OIL uses description logics for the definition of concepts and relations and proposes an ontological meta-layer that is combinable with the herein applied axiom categorization proposed by [16].

Closely related is the recent approach as pursued by DAML (DARPA Agent Markup Language) [11]. The goal of the DAML program is to create technologies that enable software agents to dynamically identify and understand information sources, and to provide interoperability between agents in a semantic manner. Particularly, an agent markup language developed as an extension to XML and RDF should allow users to provide machine-readable semantic annotations for specific communities of interest. According to the initiators of the DAML program and similar to Ontobroker, objects in the Web will be marked to include descriptions of information they encode, of functions they provide, and/or of data they can produce. In addition, DAML should allow for an „ontology calculus“ similar to the relational calculus that makes DataBase Management Systems (DBMS) possible.

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