Web rules for Health Care and Life Sciences: use cases and requirements

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ABSTRACT
This paper focuses on use cases and requirements for a Web rule language in the domain of Health Care and Life Science. Most cases are based on our experience with real applications. The paper classifies rules, identifies different use cases where rules are necessary, and summarizes some requirements of important classes of applications in that area. It appears that for most of them a loose interoperation between ontology and rules is not sufficient and a close integration between the two components is highly desirable. Finally, the paper provides results of experiments achieved with the tools currently available.

1. INTRODUCTION
This paper outlines different use cases in Life Sciences where rules are necessary and summarizes requirements for rule languages in this area. For each case, a short overview is given with its requirements, but a more detailed description can be found in related references. Compatibility with OWL and close integration with function-free Horn rules (or a restricted form) appear to be the most recurrent requirements. Several different approaches e.g.; [23] [24] [25] [26] have recently been proposed for integration of OWL with Horn rules (see the papers for a survey and technical discussion of the approaches). We experimented the prototype implementations that are currently supported. First, section 2 motivates why in this area a basic requirement to be satisfied by a Web rule language is compatibility with OWL. The next section classifies rules according to their role in reasoning (§3). Section 4 mainly focuses on the requirement of tight integration between ontology and rules. Section 5 gives examples illustrating needs of fuzzy extensions. Next, section 6 introduces major classes of application in the biomedical domain (illustrated by real examples) that require Web rules: information integration, diagnosis, and decision support. Finally, section 8 provides results of experiments achieved with the tools currently available.

2. COMPATIBILITY
First, compatibility of a Web rule language with OWL DL is highly desirable.

A Web rule language compatible with OWL DL [UC-1]

2.1.1 Outline: library of huge biomedical ontologies in OWL e.g. the FMA [20].
Life Sciences have a long tradition of controlled vocabularies. Large-scale terminologies, classifications and ontologies have been developed for many years in various biomedical domains. These resources have the potential to contribute to the Semantic Web for the Life Sciences. Several actual biomedical ontologies, e.g. The Foundational Model of Anatomy (FMA), the Medical Subject Headings (MeSH), the Gene Ontology™, the National Cancer Institute Thesaurus are converted to OWL. The conversion of other ontologies to OWL is also been investigated, including the UMLS® Metathesaurus® and Semantic Network and other ones e.g., SNOMED-CT, GALEN are represented in other DLs. New biomedical ontologies are now directly developed in OWL e.g.; BioPax an ontology for biological pathway information. For example, the FMA is the most complete ontology of human canonical anatomy. It contains more than *72,000 concepts and more than two million instantiations of 150 relations. 2/3* of the FMA (i.e. a subset of 40000 concepts) including about 40000 subclass axioms, with existential and union in rhs and other OWL constructors has been represented in OWL DL [20]. The FMA is used in several actual applications e.g. the Virtual Soldier project Virtual Soldier project [19].

DLs are more suited for 'structural' knowledge (ontology) than LP language. Using LP rules for representing large-scale biomedical ontologies is not realistic, nor desirable. In conclusion, as many huge biomedical ontologies are represented in OWL DL, a Web rule language should be compatible with OWL DL.

<u>Implications</u>:

- Interoperability between ontologies through OWL standard is necessary to allow reasoning across connected domains e.g. pathology, genes, anatomy
- As most of the ontologies use union or existential in rhs, OWL-DLP expressiveness is not enough.
- OWL DL reasoning services (consistency checking, classification) are crucial for the quality assurance of large-scale biomedical ontologies.

3. CLASSIFICATION OF RULES
Rules are needed to represent the knowledge required for various reasoning or meta-reasoning tasks. The classification below is a non exhaustive classification of rules depending on their use at a conceptual level. Rules are classified according to the role they play in the different tasks [1]: deductive rules, meta-reasoning rules, connecting rules, mapping rules, querying rules. Each category refers to the uses cases (indexed by [UC]) presented thereafter in sections 4 and 5.

Note that this classification is domain independent, e.g. mapping rules can be used in different applications, for example concerning medical data integration or travelling as well. Besides, the same rules can be used for different tasks and play different roles, e.g. the same rule can serve as a normative rule (constraint), deductive rule, or other. For example, meta-rules [UC-7] can be used to build an ontology or as constraints to check its consistency and for debugging.

1. "deductive rules" are needed for inferences based on dependencies between some ontology properties, such as the transfer of properties from parts to wholes (e.g. a disease located in an organ part, is located in the organ), or dependencies between topological and mereological properties in the brain-cortex [4] http://idm.univ-rennes1.fr/~obierlai/anatomy/annexes/annexes.pdf [UC-3] For a long time, rule-based expert systems have shown the usefulness of deductive rules in health care e.g. for diagnosis [UC-6], decision making [UC-7] etc.
2. "connecting rules" are required for connecting ontologies to allow reasoning across several domains such as Genomics, Proteonomics, Pathology, for example when searching for correlations between diseases and the abnormality of a function of a protein coded by a human gene.
3. "mapping rules" for mapping ontologies in information integration, to allow answering queries over heterogeneous sources e.g., patient data scattered in many Hospital Information Systems [12] -[14]-[17]-[18] [UC-5]
4. "querying rules" expressing complex queries upon the Web or querying heterogeneous sources [UC-5] etc.
5. "meta-reasoning rules" are needed to facilitate meta-reasoning, either to support ontology engineering e.g., acquisition, validation, maintenance of huge reference ontology [1] [11] [UC-1].or control of reasoning [UC-8].

The cases description and their requirements follow below (for more details about each case see the referenced paper).

4. CLOSE INTEGRATION

The two following use-cases illustrate that a loose interoperation between ontology and rules is not sufficient and that a tighter integration is needed.

Tight integration between the ontology and rule languages

[UC-2] case is more detailed in [2] [3]. It describes a "mapping" approach between the SWRL rule extension of OWL and the language Jess that has been used for a simple example, the Family use-case, and outlines the limits of such a loose interoperation. The next case [UC-3], more detailed in [4] [5] [6], motivates the requirement of tighter integration between the ontology and the rule language on a real application in the field of medical imaging dedicated to Brain Anatomy images interpretation.

4.1 Limitation of "mapping" [UC-2]

4.1.1 Outline: The Family rules example and the SWRLJessTab Protégé plugin [2] [3]

The Family example is a simple 'pedagogical' example including an ontology representing the usual family relationships, and a rule base representing their dependencies. It was initially designed to illustrate the needs of rules, and the results obtained with the SWRLJessTab plugin.

The SWRLJessTab Protégé plugin presented at the 7th Protégé 2004 Conference and the RuleML WS [2] [3] shows how some reasoning support might be provided to interoperate between SWRL and OWL, and exhibits why a loose interoperation between ontology and rules may be not satisfying. SWRLJessTab was intended to bridge between OWL, SWRL, Racer and Jess, for supporting reasoning with SWRL. The approach was experimented on the 'family' SWRL rules (Rbox) and ontology (Tbox and Abox). Individuals and role instances of the Abox are mapped to Jess facts. SWRL rules are mapped to Jess rules. After the Jess engine execution, the new Jess facts derived by Jess are converted into assertions on concepts and roles in Racer Abox by an inverse mapping process. Iterative calls of Racer and Jess are done until an inconsistency is detected or no new fact is inferred. Applying the SWRLJessTab approach to the case described in [2] provides the right solution. But this example should not be generalized. As pointed out in [5] and thereafter, the limitations of this method should be carefully investigated. This example highlights 1) that an OWL ontology alone is not sufficient because some triangle rules like the Uncle rule cannot be expressed in OWL 2) reasoning with incomplete knowledge, that is with one component alone, either rules or ontology, or incomplete knowledge leads to incorrect inferences. Ontology and rules should be integrated for reasoning. Besides, as exhibited with [UC-3] and next illustrated §8, if reasoning is performed with both components but *separately* by an OWL reasoner and a classical rule engine like Jess, some inferences may be missed and some results may even be incorrect.

The Protégé environment including a SWRL Editor and a plugin mechanism for integrating third party rule offers more or less similar functionalities. The loose integration of the "mapping" approach proposed in [7] [8] has similar limitations. With Protégé SWRLTab and SWRL factory, the rule inferences are based on the SWRL rules mapped to Jess, possibly added with knowledge issued from a partial translation of the OWL ontology (Tbox) into Jess. But all the knowledge of the OWL ontology component is not integrated into the rule (Jess) knowledge base. Hence, as Jess knowledge base is incomplete, some inferences may be missed or be incorrect. In fact, as DL and production rule languages such as Jess (or LP languages) have basically different expressiveness and different semantics, an OWL ontology, e.g.; the family ontology, *cannot* be converted into an equivalent Jess knowledge base. First, an OWL ontology cannot be mapped to Jess, unless the ontology uses a sublanguage much less expressive than OWL e.g., DLP fragment of OWL [9]. Besides, the reasoning method consisting in deriving all consequences with a DL reasoner e.g., Racer for the OWL (DLP) ontology, then all consequences with a rule engine e.g.; Jess for the DLP rules separately, and iterating until saturation or inconsistency, may still be not satisfying. One reason is that OWL and DLP are not really compatible from a semantic point of view. With the DL component DLP is viewed as a fragment of OWL, while with Jess DLP is treated with a different semantics ("LP semantics underlies in a large part four families of rule systems that are currently most commercially important –SQL relational databases, OPS-5 heritage production rules, Prolog …".). And as pointed out in [10], OWL-DLP being a fragment of OWL is a subset of first order Horn clauses as proposed in [9] and has open world assumption, while DLP treated as having Datalog semantics has closed world assumption. The soundness and completeness of this method for the key inference problems (satisfiability and subsumption) should be carefully investigated. Thus, although this simple approach is quite attractive, such loose integration has some limitations. The risks of processing so should be clearly identified and the users informed of it.

Implications:

- A loose interoperation of SWRL rules with existing rules engine is not satisfying, interoperating between ontology and rules requires a close integration
- A Web rule language should have a clear semantics that enables OWL DL and rules to safely interoperate.

4.2 Extending OWL DL with rules [UC-3]

4.2.1 Outline: Brain Anatomy images interpretation [4] [5]

This use case on Brain Anatomy digitally processed images[1] (MRI) reported at Washington 2005 W3C Workshop on Rule Languages for Interoperability [4] and at the OWL Experiences and Directions Workshop collocated with the International Conference on Rule Markup Languages for the Semantic Web [5] presents some requirements of a Web Rule language expected for the Semantic Web Health Care and Life Sciences[2].

Sharing anatomical knowledge (ontology and rules) and tools (services) are important needs in the context of neuroimaging, applied both to medical practice in neurology and neurosurgery and clinical research. Indeed a precise identification of the cortical structures surrounding a lesion in brain MRI images is important for the preparation of neurosurgery interventions to determine a strategy leading to the complete resection of the lesion while preserving normal brain tissue and function. On the other hand, large-scale clinical studies about major brain pathologies such as Alzheimer disease, multiple sclerosis or epilepsy need to query large amounts of brain digital images distributed in various sources, including anatomical (e.g. MRI) and functional (e.g. PET, SPECT) images.

The application aims at developing new methods for assisting the labeling of the brain cortex structures in MRI images. The proposed approach relies on a brain ontology storing the a priori "canonical" knowledge about the most important brain cortex structures, combined with rules describing the dependencies between their properties. A simplified example illustrates the need for supplementing OWL with rules, for reasoning over the ontology complemented with rules [4][5]. This example illustrates that solutions are missed if the Web ontology and the rule languages are not closely integrated. Indeed, as clearly identified by earlier theoretical works [15], usual inference mechanisms are inadequate for hybrid languages, since as pointed out in [16] "a KB may entail the antecedent of a rule without the antecedent being instantiated in the KB" and "a KB may entail the disjunction of antecedents of two rules without entailing either of them".

The language required for the rules might be a FOL extension of OWL with some form of function-free Horn rules. Several interesting approaches have recently been proposed for integration of OWL with Horn rules. A first trend proposes languages retaining decidability thanks to some restriction either on the form of rules or on their interaction with the ontology e.g.; [23] [24] [25]. Another option is the SWRL proposal [26] [27]. As pointed out by its authors [26] SWRL is undecidable, but reasoning support, e.g.; Hoolet [28] can be provided by a first order prover. At the moment we have only experimented KAON2, a reasoner implementing the approach extending OWL-DL by DL-safe rules proposed by [24] and Hoolet which provides support for SWRL (§8).

Implications

- A language extending OWL DL expressiveness with rules is required for Health Care and Life Sciences applications
- Some reasoning support is required to reason over a knowledge base composed of an ontology and rules
- The rule language to be devised should have a clear semantics that enables OWL DL and rules to safely interoperate (decidable, sound and complete reasoning)

In conclusion of these cases, a close integration between ontology and rules allowing safe reasoning is required.

5. FUZZY REASONING

5.1 Fuzzy Brain Anatomy [UC-4]

Nowadays the brain cortex can be automatically segmented into various parts using numerical and statistical methods, but the problem remains to identify these parts. On the one hand, as the image segmentation algorithm results or the user interpretation are not completely sure, the assertions describing the relations between the different parts need to be associated with confidence (truth) degrees, in particular for some topological relations. On the other hand, the OWL ontology is a simplified view of canonical anatomy representing the most frequent relations between the entities. The reality is slightly different. For example, most often centralSulcus(CS) and precentralSulcus (PCS) are contiguous (rate 0.92) but a few times they are connected (rate = 0.08):

`A1: centralSulcus ⊂ ∃ isSFContigous precentralSulcus ≥ (0.92)`

`A2: centralSulcus ⊂ ∃ isSFConnected precentralSulcus ≤ (0.08)`

Different types of fuzzy extensions of OWL might be useful, both fuzzy OWL and fuzzy SWRL extensions.

Implications

- A language extending OWL DL expressiveness with fuzzy expressions
- A language extending SWRL expressiveness with fuzzy rules

6. MAIN USAGES

Many applications in Health Care and Life Sciences are faced to the needs of rules. Information Integration, Diagnosis, Decision Making are among the "hot" applications in this area. The following examples have been selected to illustrate expected usage of rules in each category.

[1] Slides http://www.w3.org/2004/12/rules-ws/slides/christinegolbreich.pdf

[2] Slides http://www.med.univ-rennes1.fr/~cgolb/Slides/OWLED-Rules-CG.pdf

6.1 Information Integration [UC-5]

6.1.1 Outline: integration of dialysis and transplantation data for strategic decisions in Health care [12] -[14]-[17]-[18]

From a biomedical standpoint, semantic integration is crucial in many biomedical domains where better patient care, as well as better understanding of diseases and sound decision making in public health or epidemiological studies require to query large amounts of data from heterogeneous sources. In the domain of terminal organ failure and transplantation, many countries are developing regional or national registries. There are also European registries e.g.; ERA-EDTA a registry for kidney, ELTR for liver transplantation. The registries are populated by data from various national or local information systems. Different terminologies are used in the different countries and in the different centers. The absence of a shared controlled vocabulary and of coordination oblige clinicians to register patient's data several times and in various forms for regional, national and international authorities. In addition, as a result encoding is not always consistent. Semantic integration may considerably facilitate the acquisition and query of reliable patient’s data at different levels.

From a technological point of view the main challenge concerns the availability of a language that allows to use OWL for the ontology and rules for the mappings and the queries, while maintaining decidability for subsumption, satisfiability and query answering. A first study was achieved two years ago with the national French agency Agence de Biomedecine (Abm). The goal was to answer queries from three district databases storing the dialysis and transplantation patients data. The approach used was a LAV mediator based on an ontology for dialysis and transplantation. The ontology (still under construction) was issued from the Abm terminological server, originally built in integrating several existing terminologies, e.g., the French Thesaurus of Nephrology and the International Classification of Diseases (ICD). A first prototype [12] (§8) was achieved using PICSEL mediator based on CARIN language [16] (i) Semantic integration is based on a *global ontology* complying with the Abm view of the domain. This ontology defines all the concepts and properties of dialysis and transplantation. It provides the vocabulary for posing queries and for defining mappings to the local sources (ii) data are stored in the existing *local sources*. Each source uses its own ontology (iii) the content of the sources is defined in terms of views over the global ontology. A set of *mappings* relate the global ontology to the sources ontologies (iv) A *query engine* exploits the global ontology and the mappings to reformulate queries in terms of the sources specific ontologies.

Implications

- OWL DL is required for the dialysis and transplantation ontology
- A rule language is needed for expressing “mappings” and “queries”
- A language extending OWL with rules that allows representing the ontology in OWL DL, expressing mappings between the local and global ontologies, queries, and answering the user queries.

6.2 Diagnosis [UC-6]

6.2.1 Outline: Diagnosis rules in Health Care

This case is based on the Clancey’s famous method ‘Heuristic Classification’ [21]. After a careful study of various knowledge base systems, Clancey has highlighted that most of them were based on the underlying method of reasoning called ‘Heuristic Classification’. According to it, Diagnosis involves three main steps: data abstraction, heuristic matching, and refinement.

- data abstraction transforms the data (e.g., finding such as temperature value of 39° C) into data abstractions (e.g., high fever), usually using abstraction rules
- heuristic matching associates the previous data to a generic hypothesis (e.g. disease), using heuristic matching rules
- refinement allows to specialize the hypothesis into more refined hypothesis, based on an ontology (e.g. ontology of diseases such as SNOMED-CT)

This case comes with various scenarios, for example including fuzzy reasoning e.g., for breast cancer diagnosis based on hybrid reasoning combining fuzzy results issued from digital image analysis with reasoning based on the ACR classification (from the American College of Radiology) of mammography images, or with the TNM Breast Tumors classification.

Implications

- Interoperating between ontology and rules possibly fuzzy reasoning

6.3 Decision making [UC-7]

6.3.1 Outline: Guidelines assisting decision making in Health Care

For complex decision support systems, expertise is furnished by a variety of different knowledge sources including rules and ontologies. The following case deals with decision support for Type 2 Diabetes. Suggesting prescriptions that follow therapeutic recommendations requires using various sources of knowledge: both ontologies of drugs (drug taxonomy, interactions, contra-indications etc.), of diseases (disease taxonomy, e.g.; nephropathy, cause, relations etc.), of food, and rules describing guidelines. It is also necessary to access the patient's data and history stored in patient's electronic medical records.

Rules representing guidelines recommendations are composed of body usually expressed as combinations of patient's past or ongoing treatments and their outcome in terms of efficacy and tolerance [22]. Head are sets of therapeutic options often expressed as therapeutic classes of drugs e.g.; Sulfamid, Glitazones etc., but sometimes otherwise, as a particular type or group of therapeutic agents e.g.; biotherapy. The therapeutic families of drugs and the classes of diseases that occur in the rules are described in ontologies of reference, e.g.; SNOMED Clinical Terms® (SNOMED CT® http://www.snomed.org/). Rule1 and Rule 2 are examples expressing guidelines recommendations:

- Rule-1. If oral monotherapy with maximal doses of sulfamide or metformin associated with lifestyle changes is not effective, then the monotherapy should be replaced by oral bitherapy.
- Rule-2. If a drug may interact with patient's medication or other conditions e.g., contraindications do not prescribe it.

The following scenario describes a real case that should be solved by a system aiming at assisting decision-making. It has been adapted for the RIF Use Cases and Requirements W3C Working Draft 23 March 2006 (http://www.w3.org/2006/01/wiki-tr/ucr).

Bob goes to his physician, Dr. Rosen for a regular control. Bob is a 54 years old patient having Diabete, diagnosed in March 2002. His blood pressure is 16/9. His doctor's goal is to maintain the HbA1c under 6.5 (HbA1c is an indicator used to evaluate diabete stage of a patient. At the beginning, his glycemy was controlled thanks to a food diet and some physical practice, of medium intensity. Then a treatment by Glucor (a inh apha-glucosidase), 50 mg 3 time per day has been prescribed from June 2003, because of a too high value of HbA1c. Four months later, this drug was replaced by Hemi-daonil (glibenclamide, a sulfamid) 1.25 mg 3 times per day, because of digestive troubles. It was well supported. But one year later it had to be replaced by Diamicron (a sulfamide) 80 mg 3 times, because the glycemy control was not satisfying. Now, the patient has a value of HbA1c > 7 for the second time since 2 months. What should his doctor prescribe him, according to the guidelines recommendations ?

His doctor was about prescribing Stagid (metformine, a biguanide) 700 mg, 3 times. Fortunately, he double checked his prescription with his system based on interpretable guidelines, patient's electronic medical records, Drug, Food, Disease ontologies. The system informed him that he should have prescribed a bitherapy (sulfamides/metformine, or sulfamides/apha-glucosidases inhibitor, or metformine/alpha-glucosidases inhibitors) instead (because of Rule1 "If oral monotherapy with maximal doses of sulfamide or metformin associated with lifestyle changes is not effective, then the monotherapy should be replaced by oral bitherapy"). So, he changed his prescription for an association of two other drugs: metformine and Avandia (rosiglitazone, a glitazones). However, unfortunately he was mistaking again. From the data registered in the electronic record of the patient, and the Disease ontology, the system warned him that this patient diabetes was complicated by a nephropathy with mild renal insufficiency. Then, hopefully the system complained again, and sent an alert from the Drug ontology indicating that the prescribed drug metformine was contraindicated because of the nephropathy, and also warned to potentially drug interaction with Bob's current Conversion Enzyme Inhibitor used for his nephropathy!

Implications

- Compatibility with OWL is necessary to allow reasoning across domains (Drug, Food, Disease)
- Reasoning with ontologies and rules

7. ONTOLOGY ENGINEERING [UC-8]

Reasoning with rules for building or validating ontology

Rules are useful for building or validating huge ontologies such as the FMA (§ 2.1.1) or even smaller such as the Brain Anatomy (4.2). For example, the rule below generates 221 relations between the classes of the the Brain Ontology [1] [11] such as CentralSulcus separates FrontalLobe and ParietalLobe.

- `Rule-1:` IF Y is part of X and Z is not part of Y and T separates X and Z THEN T separates Y and Z

```
AE(?x) ∧ AE(?y) ∧ AE(?z) ∧ AE(?t) ∧ part-of(?y,?x) ∧ not part-of(?z,?y) ∧ separates(?t,?x,?z) → separates(?t,?y,?z)
```

Similarly, the rule below expressing that if an entity has some laterality, then its parts have the same laterality, enables to verify whether laterality constraints are respected in the FMA or Brain ontologies.

- `Rule-2:` If X is part of Y and Y has side Z then X has the same side

```
isPartOf(?x, ?y) ∧ hasSide(?y, ?z) → hasSide(?x,?z)
```

Implications

- Interoperating between ontology and rules

8. EXPERIMENTS WITH EXISTING REASONERS

Several experiments have been achieved using current existing reasoners. The list below relates the most interesting features of these experiments:

- **[UC-1] Racer**. Reasoning with current OWL DL reasoners proved to be a real challenge, due to the sheer size and complexity of the FMA. Experiments were achieved with Racer 1.7. The full-fledged FMA in OWL DL raised inference problems hard to solve in terms of time and memory [20]. Even with RacePro 1.8 the memory and time needs for running the full version are prohibitive. Therefore, at the moment an incremental approach was adopted for reasoning with less complex versions.
- **[UC-2] Racer and Jess**. We tested the Family example using Racer 1.7 and Jess, thanks to the SWRLJessTab plugin. All wanted results were obtained with the SWRLJessTab plugin for the specific case under study, but we had extended the rule base with rules reflecting some knowledge of the ontology that was not translated. This plugin is not maintained and does not work any more with current Protégé OWL and RacerPro versions. Nowadays the Protégé environment includes a SWRL Editor and a plugin mechanism for integrating third party rule that offers more or less similar functionalities. But such an approach should be used with cautious. It would be worth to elicit under which assumptions, e.g.; expressiveness restrictions of DLP-OWL, DL-safe rules etc. this method might be used and to inform the users of the risks met e.g. non termination, incorrect answers or other etc. with a tool such as the current SWRLTab intended to provide inference support for rules using the Jess rule engine.

  KAON2. Different simplified versions of the Family use case have been tested using the KAON2 reasoner (http://kaon2.semanticweb.org/) implementing the DL-safe rules approach proposed by [24]. A first experiment was achieved for the simplified ontology below

Man ⊑ Human

Uncle ≡ Man ⊓ ∃isUncle Human

Parent ≡ Human ⊓ ∃hasChild Human

Domain(hasChild) ≡ Human (resp. range)

Domain(hasSibling) ≡ Human (resp. range)

Domain(hasUncle) ≡ Human (resp. range)

isUncle ≡ $(\text{hasUncle})^{-1}$

extended by the single Uncle rule:

hasChild(?z, ?x) ∧ hasSibling(?z, ?y) ∧ Man(?y) ∧ Human(?z) ∧ Human(?x) → hasUncle(?x, ?y)

For this very simple ontology, from the initial facts Man(John), Human(Bob), Human (Sam), hasChild(Bob, Sam), hasSibling(Bob, John) KAON2 derives hasUncle(Sam, John), hence that John is an instance of Uncle. In this case KAON2 provides the right answer to the query Uncle(?x) (Figure 1).

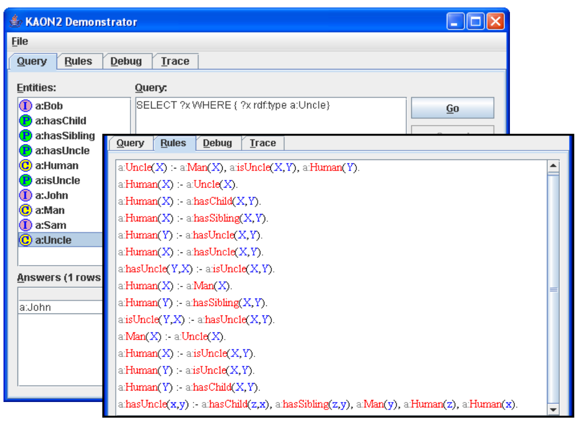

Figure 1 KAON2 answers to Uncle(?x) in test 1

The second test was achieved for the same ontology and facts, apart that Bob was asserted to be a parent, but now we don't know who is the child: Man(John), Parent(Bob), Human (Sam), hasSibling(Bob, John). As Bob has a child and Bob has a sibling John, it should logically be inferred that John is an instance of Uncle. However, KAON2 does not provide the expected answer Uncle (John). The reason is the same as for the Romulus and Remus example described in [24]. KAON2 reasoner computes the set of all the consequences that can be drawn for DL-safe rules. However the Uncle rule is a SWRL rule and is not DL-safe. When KAON2 is used, it internally interprets the SWRL rules as DL-safe rules, which means that all variables should be bound only to known individuals. Therefore, as there is no assertion connecting Bob to a known individual by the hasChild relation, the Uncle rule cannot fire for Bob, hence John is not inferred to be an instance of Uncle. Thus, John is a consequence according to the first order semantics of SWRL, but KAON2 does not draw it, as it is not a consequence under "the DL-safe semantics".

Hoolet. The same test was achieved using Hoolet [28], a prototype implementation of a SWRL reasoner that uses Vampire, a first-order prover. Obviously, Hoolet provides the expected answer John, since all inferences corresponding to the first order semantics of SWRL are derived.

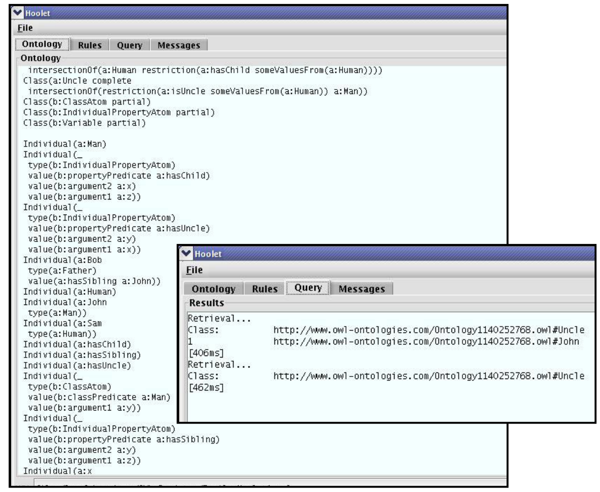

Figure 2: Hoolet answers to Uncle(?x) in test 2

- **[UC-3].** Various experiments are currently achieved with KAON2 for [UC-3]. Because KAON2 does not currently support datatypes and nominals, it was necessary to simplify the OWL ontology of the application to cope with these limitations. The OWL ontology and SWRL rules were edited with Protégé-OWL-SWRL editor (Figure 3).

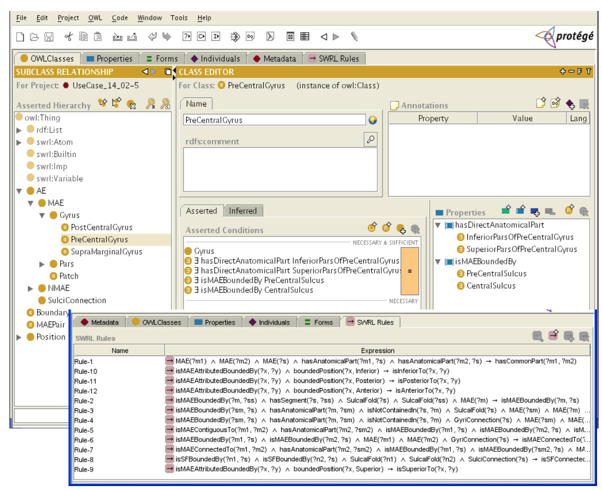

Figure 3: Editing OWL ontology and rules with Protégé

Different cases are currently experimented. KAON2 works well for simple cases similar to the Family test1. For example Figure 4 shows the result of the SPARQL query asking for the parts of a Brain MRI image that belong to the PreCentralGyrus: SELECT ?x ?y WHERE {?x rdf:type a:PreCentralGyrus; a:hasAnatomicalPart ?y }

KAON2 computes the right answers for this case. Indeed, although the rules are not DL-safe, the rules could be fired because, given the initial facts asserted, their body can be satisfied by bindings of their variable to known individuals. Therefore, just like for test 1, KAON2 infers that Gyrus_1 is a PreCentralGyrus, and next provides the three expected answers P1, P2, P3. But the same difficulty as for test2 occurs when P1, P2, P3 are asserted to be some instances of Patch without naming the instance Gyrus_1 which they are part of. Thus using KAON2 for our application is not straightforward. Various solutions and more complex cases will be further investigated with KAON2 and Hoolet. It should be noted that negation and n-ary relations occur in the rules of the real application [5].

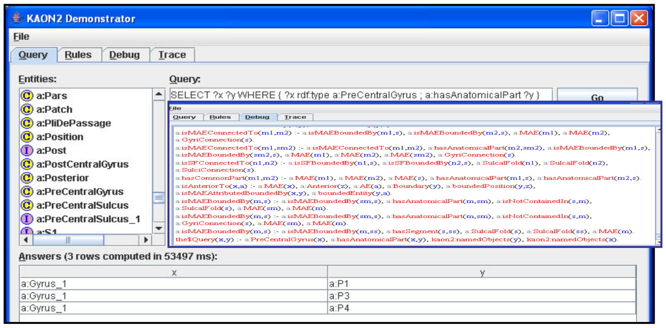

Figure 4: Running KAON2 for a simple example of [UC-3]

- **[UC-4].** Simple extensions to fuzziness were tested with the FiRE 'Beta Reasoner for the Fuzzy Description Logic f-SHI' under development**.** At the moment FiRE was runned for an OWL simplified TBox without Fuzzy-OWL definitions, but with fuzzy assertions including degrees of accuracy like:

  `isSFContigous (o4 o10) >= 0.99`

  `isMAEConnected (o4 o10) <= 0.2`

  We designed some example where FiRE inferred that the knowledge was inconsistent, from the Tbox and the example given in the Abox (Figure 5).

  However at the moment, although some use-case is available, it was not possible to run examples with triangle rules (even SWRL non fuzzy rules) because of FiRE current limitations.

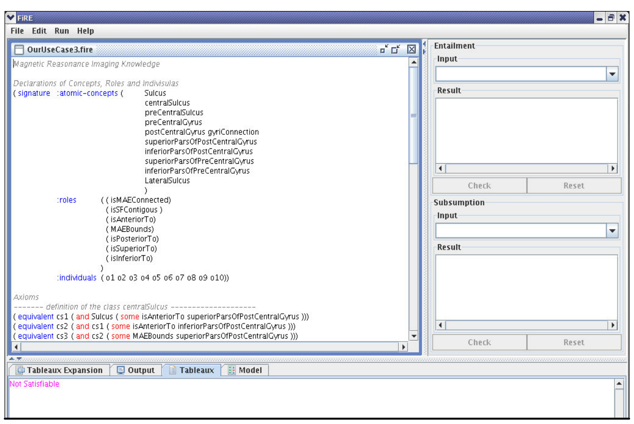

Figure 5: Fire results

- **[UC-5].** The experience aiming at integrating various sources of information about dialysis and transplant, thanks to the LAV mediator PICSEL was achieved two years ago. The global ontology was designed with Protégé OWL and Racer was used for classification and consistency checking. Then, the OWL ontology was migrated to CARIN-$\mathcal{ALN}$ [16]. As OWL DL is more expressive, some transformations were next achieved to migrate the ontology to CARIN. For example, we replaced existential restriction of the OWL class definitions by the conjunction of a universal restriction with a cardinality restriction. Some lack of expressiveness of the DL component of CARIN was compensated by the additional expressiveness provided by its rule component. For example, two rules were defined instead of a disjunction between two classes. Although the resulting knowledge base is not equivalent to the original one, it was thus possible to run PICSEL for this example. This experiment demonstrated the benefit of a LAV mediator. It should be interesting to know how the recent approaches for OWL DL extensions by function-free Horn rules e.g.; [23] [24] might be now used for answering queries with views.

9. CONCLUSION

Although simplified, the summarized examples and experiments correspond to real biomedical applications and needs. Whatever the type of rules concerned, all the presented cases require reasoning on an OWL ontology and rules. For most of them a Web rule language allowing a close integration between the two components is needed. An extension of OWL with some form of function-free Horn rules seems highly desirable. Recent researches have proposed different approaches [23] [24] [25] [26] and some prototype implementations for such integration. Further investigations are needed to determine whether the current techniques and reasoners meet the needs of the real applications presented, and in particular whether the restrictions imposed to retain decidability are acceptable or whether another approach might be proposed.

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