A novel way of integrating rule based knowledge into a web ontology language framework

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Abstract. Web ontology language (OWL), used in combination with the Protégé visual interface, is a modern standard for development and maintenance of ontologies and a powerful tool for knowledge presentation. In this work, we describe a novel possibility to use OWL also for the conceptualization of knowledge presented by a set of rules. In this approach, rules are represented as a hierarchy of actionable classes with necessary and sufficient conditions defined by the description logic formalism. The advantages are that: the set of the rules is not an unordered set anymore, the concepts defined in descriptive ontologies can be used directly in the bodies of rules, and Protégé presents an intuitive tool for editing the set of rules. Standard ontology reasoning processes are not applicable in this framework, but experiments conducted on the rule sets have demonstrated that the reasoning problems can be successfully solved.

Keywords. Rules, knowledge, ontologies, decision support.

Introduction

Clinical practice guidelines play more and more important role in improving the everyday quality of medical care. To facilitate their sharing across different institutions and applications, including medical knowledge management and decision support systems, their representation in standardized machine interpretable forms is necessary. There exist different approaches for the implementation of computer-interpretable guidelines, including: Arden syntax, GuideLine Interchange format (GLIF), PROforma, and Asbru [1]. There are some nicely presented challenges and implementation details of intention based computerized clinical practice guidelines by the system GASTINE [2], with the focus on heart failure management, which is also the topic of this work.

The approach presented in this work is based on production rules and is, because of that, more similar to Arden syntax approach then to systems based on flowcharts and plans representation like GLIF and Asbru [1]. The decisive difference from Arden syntax is that Web Ontology Language (OWL) is used instead of Medical Logic Modules (MLMs). The main goal is to systematize the set of rules and integrate them with ontological description of the domain, and not to share individual medical logic modules. As in the GASTINE system, we are using ontological elements and Protégé based user interface, but with a completely different representation formalism [2]. The work started by constructing the ontology, now publicly available from http://lis.irb.hr/heartfaid/ontology/, which represents a relatively complete formalized description of concepts for the heart failure domain [3]. Based on the HF guidelines [4], and in cooperation with cardiologists, it was possible to define a set of about 200 rules necessary for the implementation of a decision support system for the HF patients' management. A natural choice for formalization of the rules was to use Semantic Web Rule Language (SWRL). This approach was not satisfactory for two main reasons. The first issue is a very complicated syntax of SWRL, which prohibits entering or updating rules by domain experts. The second issue is that reasoning in SWRL is done with the open world assumption that is inherited from OWL, which requires explicit definition of many additional concepts just for representing non-existing data and negations of concepts and data that are used as negative conditions in the rules.

In order to solve these problems, we have implemented a novel approach that uses Protégé as a standard user interface and represents rules as a special type of OWL classes that are, in this work, called actionable classes. Visual editing of the classes in Protégé is simple and intuitive [5], enabling domain experts to actively participate in the development of the rule base. This is relevant, because maintenance of the rule base and its adjustments to specific local needs is a never ending task. Additionally, some other advantages of the approach have been detected. This paper begins with a short description of the sets of rules collected for the heart failure domain in Section 1, and continues by defining actionable classes in Section 2. In Section 3, we present the reasoning process for this form of knowledge representation.

1. A set of rules for the heart failure domain

The guidelines regularly published by the European Society of Cardiology [3] have been the main source of information about the diagnostic and treatment procedures for the management of HF patients. The knowledge for the management of HF patients consists of eight sets of rules. It is presented on the left hand side of Figure 1. The most relevant parts are: rules for diagnosing heart failure, heart failure severity assessment, prognosis estimation, specific medication prescription and dosage, and acute decompensation of congestive heart failure. Such organization has been suggested by cardiologists, with intention to enable an overview of the current status of the collected knowledge. The sets of rules are constantly evaluated and improved. The type of the collected knowledge is illustrated by a rule from the set related to HF diagnosis:

Patient has diastolic HF if
a) he has performed echocardiography and
b) either low E/A ratio or prolonged deceleration have been detected and
c) the patient has some HF signs or symptoms.

2. Definition of actionable classes and their hierarchy

Classes in OWL may be defined by necessary and sufficient conditions that instances must satisfy in order to belong to them. A set of rules is presented so that for each rule a separately defined actionable class is introduced, whose name corresponds to the rule conclusion (consequent), and its necessary and sufficient condition corresponds to the rule condition (antecedent). The right hand side of Figure 1 presents a definition of the



Figure 1. The hierarchy of actionable classes (left) and implementation of the rule "Diastolic_positive" (right).

actionable class "Diastolic_positive", which is an implementation of the rule introduced in the previous section. The elements of the conditions for actionable classes include concepts defined by other actionable classes (e.g. "Has echo done" or "Has HF Signs") and properties from the HF ontology that may be attributed to patients (e.g. "hasCharacteristic"). In this way, the descriptive knowledge available in the HF ontology may be used directly in the decision making process. The knowledge defined in this ontology (e.g. HF symptoms include angina pectoris and weakness) influences the decision making process. This is possible because both descriptive knowledge and actionable classes use the OWL formalism.

All actionable classes for the HF decision task are created as sub-classes of the class "Patient" and, for the sake of human comprehension, they are organized as a hierarchy of classes (Figure 1). Only real patients are instances of the class "Patient" and its actionable sub-classes. Patient instances have properties "hasCharacteristic" and "hasData". The property "hasCharacteristic" connects them with the instances of the class "Patient_characteristics" from the descriptive part of the ontology. "Patient_characteristics" is the range of this property. A subset of all possible instances from this class attributed to a particular patient instance determines the factual knowledge about the patient.

Super-classes of actionable classes are also actionable because, during the reasoning process, a patient may become their instance simply because the patient became an instance of at least one of their sub-classes. This feature enables very intuitive implementation of the logical OR functions of conditions. Additionally, if a consequence of a rule is a necessary condition for another rule, then the actionable class presenting the former rule is automatically a super-class for the class presenting the latter rule. This means that, in these specific situations, the structure of the hierarchy of classes is not arbitrary, but actually determined by the logic of the implemented rule based knowledge. The structure of the hierarchy may help medical experts to understand the underlying logic of the knowledge base and, when necessary, to easily identify the position of the actionable classes which have to be updated.

3. Decision support system with actionable classes

The reasoning task, with knowledge presented in the above described way, is solved by the construction of a specialized reasoner called COSI (Closed World OWL Interpreter). Its construction did not turn out to be a major problem due to the simplicity of the syntax of OWL and description logic as well as public availability of the OWL Java API. The term "interpreter" denotes that the experimental realization does not include complete functionality expected from OWL reasoners. Its function is only to detect membership of instances in classes. The process requires that complete factual knowledge about patients and all of the intermediate reasoning results have to be presented in OWL syntax as well.

The first step in the decision support process is data extraction, transformation, and loading, which imports all relevant data about a real patient from electronic health record. The data are stored as factual knowledge in the OWL formalism so that only instances that are actually true for the patient are present in the class "Patient_characteristics". After that, the reasoning process follows, which results in placing the instance of the patient into potentially many appropriate actionable classes. The last step is the interpretation of the results. It is performed so that each actionable class is tested for including the target patient instance. If the test turns out true, then the comment part of the class is searched, and if there is a comment starting with the word "message" (see Figure 1), then a message is directed towards the user. The text of the message is defined by the second part of the "message" comment, while a group in which it will presented in the user interface (like "Diagnosis" or "Severity", as presented in Figure 2) is defined by the comment starting with the word "group". There is also a possibility to provide explanations of the resulting decisions, which is illustrated at the bottom of Figure 2.

It was noticed that a relative small part of the HF ontology is practically used by the implemented decision support system. The useful classes were those that define sets of relevant HF signs and symptoms as well as classes that define lists of medication types in relevant medication groups like diuretics, aldosterone antagonists, and β blockers. In addition to these classes, very useful were the data about abnormally low or high levels of laboratory measurements and initiating and maintenance doses for all relevant medication types.

4. Discussion

The fact that the structure of the hierarchy of actionable classes reflects, to some extent, the content of the knowledge represented by the rule base is an argument for the statement that the proposed approach represents a step towards conceptualization of this knowledge. The experiment performed within the EU project Heartfaid demonstrated that the approach can be effectively implemented with OWL, that the implementation of closed world reasoning with OWL syntax is possible, and that medically useful decision support results may be obtained. High temporal complexity is the main practical problem of the present implementation. It is the consequence of the used naïve iterative approach to computation of class membership for all given instances. Realization of more effective data extraction procedures and reasoners is a future task and a necessary step for applications with larger rule bases.

Diagnosis (6)
why? Heart failure symptoms
why? Diagnosed heart failure diastolic positive
why? Diagnosed heart failure systolic negative by echo test
why? Acute CHF by BNP
why? Patient has some signs or symptoms of acute decompensation
why? Positive diagnosis confirmed by very high BNP
Additive-diagnosis (6)
Severity (4)
Prognosis (2)
Medication-status (4)
Medication-suggestions (4)
Medication-warnings (7)
Management (3)
Other (6)

Explanation:

Diagnosed heart failure diastolic positive
Patient has:
echo test done
low EA ratio
heart failure symptoms

Figure 2. The user interface of the Heartfaid decision support service with reasoning results for a real patient.

The presented approach may be used only for medical knowledge representable by rules. The expressiveness of the rule formalism is low, especially with respect to the actions that have to be performed sequentially. This means that the methodology is not appropriate for modeling complex medical procedures. The available expressiveness was completely satisfactory for our particular application. The major advantages of the approach are logical simplicity of reasoning, which is a serious bottleneck for all of the more expressive systems, and the very acceptable man-machine interface that enables active participation of domain experts in the decision support system design.

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