A Three-Layer Domain Ontology for Guideline Representation and Sharing

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Medical Domain ontology is an important issue in guideline representation and sharing. The Guideline Interchange Format (GLIF) addresses the domain ontology problem to satisfy the needs for a variety of representation objectives: readability, machine-interpretability, integration into local information system environments, and authoring/execution support. This is accomplished in GLIF by a flexible three-layer domain ontology. The first layer, Core GLIF, defines a standard interface to medical data and concepts. The second layer, Reference Information Model (RIM), defines the basic data model for representing medical information. The third layer, Medical Knowledge, supports term encoding and semantic verification of guidelines.

Introduction

Medical terms and concepts are essential building blocks of clinical guideline documents and applications. An ontology provides a framework for the representation of and reference to terms, concepts and their relationships. For sharing of computer-based guidelines, ontology support becomes an important issue.

In the past, most guideline representation schemes had focused on the representation of logic and left the ontology issues for individual implementations to handle[1-3]. However, lack of ontology support hinders the sharing of knowledge represented in those schemes, since different applications and institutions often do not share the same vocabulary or database schema. A well-known example is the "curly bracket" problem in Arden Syntax where all implementation-specific detail is placed within curly brackets, and is thus not sharable[4]. Some recent developments in guideline representation have started to address the ontology problem[5,6,7]. For example, Quaglini et al proposed an ontology-based framework model for guidelines[5]. As a part of guideline modeling, the EON project created an explicit domain ontology that models basic concepts and relations in a medical specialty[8].

GLIF is a representation language that was developed to facilitate sharing of clinical guidelines. GLIF models guidelines as flowcharts of structured guideline steps, representing clinical actions and decisions. GLIF 2, which was published in 1998, offered little support for vocabulary and data type modeling[1]. The latest draft GLIF version, GLIF 3, aims at enabling guideline specifications to be not only readable, but also computable on virtual machines and able to be integrated into institutional applications[9]. The development of a domain ontology is part of the effort to achieve such goals.

Because of a number of differing objectives for guideline representation and sharing, the demands on ontology vary. We describe a flexible three-layer approach to meet the different demands and support incremental development.

General Approach

In GLIF, the needs of guideline sharing are classified into three levels[10]. On the conceptual level (level A), guidelines only need to be human-readable. On this level, medical terms are just free text strings that do not need to be encoded. On the computable level (level B), guidelines should be able to run on a virtual machine. To execute a guideline on a virtual machine, we need to distinguish between variable and literal data. When data items are more complex than simple types such as string or integer, it is important to define data models for sharing purposes. On the implementation level (level C), guidelines are integrated with institutional clinical applications. This requires that the data model and vocabulary used by the guideline be mapped to those used by the institutional applications, or to procedures in the institutional system that will obtain or retrieve the corresponding data elements. On both levels B and C, ontological knowledge of the attributes of medical concepts and the relationships among medical concepts that

go beyond the representation of basic data fields and concept hierarchy, is highly desirable. With such knowledge, we can perform rigorous type checking, range checking, and semantic checking (e.g., whether an expression refers to meaningful characteristics of a concept).

The support of these ontological needs for guideline modeling is separated into three layers. The first layer, *Core GLIF*, is part of the GLIF specification language. It defines a standard interface to medical data and concepts. The second layer, *Reference Information Model (RIM*), is essential for guideline execution and data sharing among different applications and different institutions. It defines the basic data model for representing medical information needed in specifying protocols and guidelines. It includes high level classification concepts, such as drugs and observations about a patient, and attributes, such as units of a measurement and dosage for a drug, that medical concepts and medical data may have. The *Medical Knowledge* layer contains a term dictionary and a medical knowledge base. Though medical knowledge is very desirable, we can only make limited assumptions about its existence because of its huge scope.

When all three layers are involved, they work closely together: Core GLIF relies on the RIM to supply the attributes of the medical concepts and to represent data values. Core GLIF relies on the Medical Knowledge layer for encoding medical concepts. The codes supplied by the Medical Knowledge layer also determine which particular data models in a RIM are used. Figure 1 is an example showing how part of a thyroid screening guideline is represented using the three-layer ontology.

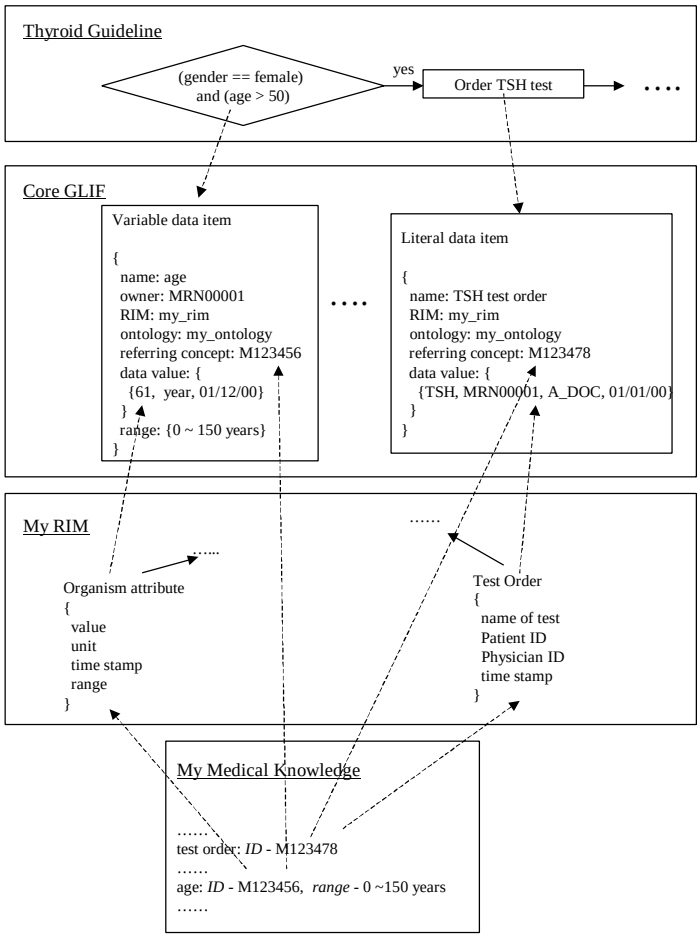

Figure 1. This figure shows how two terms (*age* and *order TSH test*), used in a thyroid screening guideline, are represented using the three-layer ontology. In the Core GLIF layer, *age* is represented as a variable data item and *order TSH test* as a literal data item. The data items employed a mock RIM **(**My_RIM) to provide data model of their values and a mock Medical Knowledge layer (My_Medical_Knowledge) to provide concept codes and range checking knowledge.

In the following paragraphs we describe the three layers of GLIF's medical ontology.

Core GLIF

Core GLIF ontology defines how medical data and concepts should be referenced by GLIF. It also defines the scope of data items and how the data items acquire their values. Core GLIF is part of the GLIF specification.

Core GLIF views all medical terms and concepts as data items. Data items are symbols that may have values. GLIF need to employ data items to describe decision and action. Data items represent data that need to be acquired from external sources, such as patient's weight, or data that are computed or assigned by guidelines, such as risk level and creatinine clearance. The data item values conform to the data objects whose structure is defined by the RIM layer of the medical ontology. Each data item has a name, a RIM identifier indicating the RIM it uses, a Medical Knowledge identifier indicating the Medical Knowledge ontology it uses, and the code of a concept to which the data item refers. By specifying the RIM and Medical Knowledge ontology at the level of an individual data item, we allow one guideline to refer to multiple RIMs and Medical Knowledge ontologies. For ease of use, a default RIM and Medical Knowledge ontology can be defined.

Core GLIF distinguishes between two types of data items: literals (constants) and variables. A variable data item can have mutable values. A variable data item has an owner and a data value attribute, which is modeled as a list of instances of a data object defined by the RIM layer. *Patient's height, weight, gender, and age* are variable data items. The variable data item *age* can be seen in Figure 1.

A literal data item is a data item that has fixed value. It is similar to a constant in programming languages. Unlike a variable data item, a literal does not have an owner and its data value is modeled by an empty list or a list of exactly one instance of the data object defined by the RIM layer. C*ongestive heart failure, female*, *smoker, and TSH test order* are all examples of literal data items. *TSH test order* can be seen in Figure 1.

The values of variable data items can be assigned at execution time and the values of literal data items can be assigned at authoring or execution time. In the example in Figure 1, the value of the variable data item *age* is assigned at execution time. The *name of test* attribute of the literal data item value *TSH test order* is assigned at authoring time to *TSH.* The rest of the attributes of the *TSH test order* literal (patient ID, physician ID and timestamp) are assigned at execution time.

Core GLIF does not require the existence of RIM and Medical Knowledge layers. When RIM and Medical Knowledge are not available, the data representation is handled by a default model. However, we do expect users to employ RIM and Medical Knowledge layers for level B and C guideline representation.

For a RIM to be acceptable for use by GLIF, it should have a class hierarchy that organizes medical concepts into classes. For each class, the RIM should provide a data model that defines the attributes or fields of the data objects that belong to that class. The Medical Knowledge ontology should provide a term dictionary that maps text strings to medical concepts.

When users simply want to create a human-readable guideline, a RIM or term dictionary might not be needed. Even when a RIM and a term dictionary are provided, not all terms may be mapped to concepts nor all concepts have a data model. When both RIM and Medical Knowledge ontology are absent, the RIM and Medical Knowledge ontology identifier fields in Core GLIF data items are marked as "UNKOWN". When a term fails to be mapped to a concept, the referring concept is automatically assigned the value "UNKOWN". When a concept or term does not have a data model specified by the RIM, the type for each instance of data value is assigned a type "string".

Some parts of a guideline can be grouped into modules that we refer to as *subguidelines.* Subguidelines may be reused and shared among different guidelines. Sometimes there is a need to pass data between a subguideline and a controlling guideline. The sharing of data items among a subguideline and a guideline is allowed only when explicitly defined.

RIM

The RIM defines the data model for medical concepts. The Health Level 7 (HL7) RIM and foundational models in SNOMED-RT are examples of RIMs[11,12]. GLIF requires the RIM to define a class hierarchy for top-level medical concepts and data attributes for each class of medical concepts.

The class hierarchy allows the medical concepts to be assigned to appropriate classes, although it does not necessarily perform the actual classification for concepts. For example, with a class hierarchy, one can classify a digoxin order as a medication order and a sodium test as a lab test. GLIF makes no

assumption about whether the class hierarchy is a multiple or single hierarchy. GLIF also does not make any assumption about the granularity of the classification. In the example given in Figure 1, *organism attribute* and *test order* are two classes in a mock hierarchy, used to classify *age* and *TSH test order*, respectively.

GLIF requires the data attributes (i.e., fields of a data item value) of each class to be defined. For example, *medication name, frequency, starting date,* and *ending date* may be data attributes of medication orders. Some classes may have no data attributes. In addition, not all attributes are data attributes. For example, *blood glucose test* may have an attribute indicating that it is a diagnostic test for diabetes. This attribute is typically not considered as a data attribute and thus not part of the RIM because the diagnostic implication of a test is normally not part of a test value. In the example given in Figure 1, the class *organism attribute* has the data fields *value, unit,* and *time stamp*. The class *test order* has the data fields *name of test, patient ID, physician ID* and *time stamp*.

Developing a RIM requires much effort. We intend to provide a default GLIF RIM with basic data types. In our study, we found both the HL7 RIM model and Unified Medical Language System (UMLS) semantic net promising, yet, not necessarily completely satisfactory for guideline authoring and execution purposes[12,13]. For example, the UMLS semantic net is relatively shallow and sometimes clusters concepts with very different data attributes into one class. HL7 RIM, with its recent Unified Service Action Model (USAM)[14], provides a model that is much closer to the needs of GLIF 3. We are studying how to adapt them for use by GLIF.

Theoretically, a RIM does not require the existence of a term dictionary. However, it is likely that some kind of term dictionary will be used with the RIM. When a term dictionary is used, it is assumed that the RIM hierarchy can be applied to the concepts in the term dictionary.

Medical Knowledge Layer

The Medical Knowledge layer consists of two parts: a term dictionary and a domain knowledge base. GLIF supports the use of a Medical Knowledge layer, but does not provide one.

GLIF does not require a specific term dictionary for use by it. If browsing is the objective for guideline representation, it is not essential to have a term dictionary. However, in order to execute a guideline, coding is required. In the example given in Figure 1, the coding of *age* and *test order* is provided by *My_Knowledge_Layer*. When integrating guidelines with other clinical applications, the term dictionary should support vocabulary mapping because applications often do not share the same coding scheme.

It is beyond the scope of GLIF to develop a comprehensive term dictionary. There are a number of vocabularies that offer term dictionaries. However, not all of them offer strong support for mapping between vocabularies. Given that GLIF is intended to facilitate sharing, we found the UMLS particularly useful, because it incorporates terms and concepts from over 50 vocabulary sources.

We would like to be able to verify the correctness of a guideline during authoring. There are different levels of correctness. A syntactically correct expression may not be semantically correct. Note that we are not addressing yet another level of correctness, medical correctness, which is beyond the scope of GLIF. The only way we can verify semantic correctness is by obtaining semantic knowledge. For example, "female > 50 years" is semantically wrong because female and 50 years are incompatible data types. Knowledge can also provide users more assistance in authoring and implementing guidelines. The expression "patient == female" can be semantically interpreted only when we know that female is a gender and that a patient has the attribute "gender". In the example given in Figure 1, the range of *age* provided by the Medical Knowledge layer supports the range check of the data item.

Discussion

The demands on domain ontology support vary, depending on the objectives of the guideline representation. The three-layer medical ontology that we describe is aimed at providing a flexible way of satisfying the needs for a variety of representation objectives: readability, machine-interpretability, integration into local information system environments, and authoring/execution-support. A readable representation can use a relatively free style while representations with rigorously structured form are needed to support computation. In the three-layered medical ontology, users have the freedom to choose the kind of representation that fits their needs.

GLIF cannot translate one RIM or Medical Knowledge ontology to another. There is no guarantee that such translation can be achieved without loss of information. For example, if one data model has a field that another lacks, the value in that field may be lost during translation. If two parties wish to share a guideline for a purpose that goes beyond human readability, great benefit may be gained by

using the same RIM and Medical Knowledge ontology.

To give users as much freedom as possible, GLIF allows the use of different data models and coding schemes in one guideline. However, not all schemes are compatible. For example, if a subguideline imports a data item using a data model different from the one the guideline uses, it must perform mapping before referencing the value. Consistency in the use of data models and coding schemes is desirable.

GLIF is an evolving guideline representation method and its medical ontology will also evolve over time. Core GLIF has been incorporated into the GLIF 3 draft specification. We are developing a RIM that fits the needs for guideline sharing. Our future research in the Medical Knowledge ontology will focus on classifying the kinds of knowledge that are needed and how GLIF can make use of them.

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