ABSTRACT

Traditional database resources and semantic Web technology face problems when there is a need to keep track of individuals in reality as these individuals undergo changes of various sorts. We describe an application which implements the referent tracking paradigm in which each real world entity has its own unique ID. The application is designed to be able to store relationships between tracked instances and also to be extendable to very high orders of magnitude (in principle to accept numbers of entries in the billions). Our approach is based on ontologies grounded in realism, but it can be extended also to information that is captured using the terminologies or concept-based ontologies used in traditional knowledge representation systems. The repository uses resource description framework (RDF) as representation format, and it can thus be queried with query languages such as SPARQL, SeRQL, and RQL, thereby providing support for reasoning over multiple ontologies.

Keywords: electronic health record; ontology; RDF; referent tracking; semantic interoperability; semantic Web; unqualified realism

INTRODUCTION

Electronic health record (EHR) systems are software systems that manage patient information that typically arises within a single healthcare institution. Such systems exist in various flavors and can be built up out of several different types of components and rely on different types of standards such as HL7 (Health Level Seven Inc., 2007) or openEHR (Blobel, 2006). One particular component of a modern EHR deals with the access to terminologies such as ICD-9-CM (U.S. Department of Health & Human Services, 2006) or SNOMED-CT (SNOMED International, 2007), to coding and classification systems, and, more recently, also to ontologies such as the Foundational Model of Anatomy (FMA™ University of Washington, 2006). The purpose of using such systems is to avoid the ambiguities and interpretation problems that often arise when health professionals use local terminologies (or no terminologies at all) to enter statements in an EHR (Rosenbloom, Miller, Johnson, Elkin, & Brown, 2006). Unfortunately, this goal has thus far been only partly
achieved. Using terminological systems of the sorts referred to above, in which the terms are given an intended and (so it is claimed) unique meaning, may indeed, if the system is used properly, reduce but not eliminate the risk of misinterpretation by humans. And, certainly, existing EHRs do not contain enough information of the right sort to enable correct interpretation by software agents. As an example, imagine that John consults a physician for a fracture in his left forearm at time \( t_1 \) and that the fact of John having such a fracture at that time is registered in John’s record by putting the SNOMED-CT code 91419009 in the diagnosis field of the chart describing that particular visit (which took place at a certain place, with a particular physician, on a precise date, and so forth, all data which are registered in the context of that fracture). If John, at time \( t_2 \), suffers from another fracture in his left forearm and the same procedure is used for registering this new fact, then, there is no obvious way to know whether the very same fracture is referred to, or a second one. This is because codes from terminological systems or ontologies do not identify uniquely the entities to which they are assigned in the context of clinical record keeping. They rather describe what generic category the entities to which they are assigned belong to. Referent tracking (RT) is a paradigm that was introduced in 2005 in the field of EHR systems and that is intended to avoid such ambiguities through the mechanism of assigning globally unique IDs (called IUIs for “Instance Unique Identifiers”) to the entities on the side of the patients that clinicians refer to when writing statements in an EHR (Ceusters & Smith, 2005). These IUIs are thus explicit references to the real-world entities (called particulars in the tradition of philosophical ontology) on the side of patients, including their body parts, diseases, therapies, and so forth.

Secondly, many efforts have been and are being made in developing ontologies and structured vocabularies in different domains to make data understandable by machines. Here interoperability is an indispensable requisite as EHR data are characteristically derived from different software systems. In this context various representation languages have been developed for purposes of ontology development, an example being the W3C recommended ontology Web language (OWL) (Smith, Welty, & McGuinness, 2004). In addition, there are tools such as Protégé (Gennari, Musen, Ferguson, Grosso, Crubezy, Eriksson et al., 2003), SWOOP (Kalyanpur, Parsia, Sirin, Grau, & Hendler, 2006) and OBO-Edit (OBO-Edit Working Group, 2006) which have been used in building ontologies such as the FMA and the Gene Ontology (The Gene Ontology Consortium, 2007). Reasoning with such ontologies can be done with tools such as Pellet (Sirin, Parsia, Grau, Kalyanpur, & Katz, 2006), Racer (Haarslev & Möller, 2001) and FaCT (Tsarkov & Horrocks, 2006). Some of these representation tools allow only class-level representations, while most current reasoners do not support reasoning over instances at all or in ways that mirror the relationships between the instances in reality. And finally, existing ontology tools typically fail when they are loaded with large amounts of instance data.

An additional problem exists at the level of the integration of terminologies and ontologies in EHR applications because the latter can refer to terms from different systems each of them represented in distinct language formats.

We describe a software system which implements the referent tracking paradigm. This referent tracking system (RTS) is able to contain large amounts of data pertaining to real-world entities and their relationships in a way that is consistent with the view endorsed by philosophical realism. The RTS is designed to act as a backbone for other applications such as EHRs. It uses resource description framework (RDF) as a representation language, and can be queried by means of semantic query languages thereby providing support for reasoning over multiple ontologies. The software is developed in Java and is available as a standalone server application accessible through Web services as well as a library which allows client applications to embed the RTS. The server is intended to be hosted by a health institute which serves...