TOWARDS INTEROPERABILITY IN e-HEALTH SYSTEMS *A Three-Dimensional Approach based on Standards and Semantics*

Jose Manuel Gómez-Pérez¹, Sandra Kohler¹, Ricardo Melero¹, Pablo Serrano², Leonardo Lezcano³ Miguel Angel Sicilia³, Ana Iglesias⁴, Elena Castro⁴, Margarita Rubio⁵ and Manuel de Buenaga⁵ ¹iSOCO S.A, Pedro de Valdivia 10, Madrid, Spain

²Hospital de Fuenlabrada, Madrid, Spain

³Universidad de Alcalá de Henares, Madrid, Spain

⁴Universidad Carlos III, Madrid, Spain

⁵Universidad Europea de Madrid, Madrid, Spain

Keywords: Semantic interoperability in eHealth, CEN 13606, Archetypes, NLP, OWL, SNOMED.

Abstract: The interoperability problem in eHealth can only be addressed by means of combining standards and technology. However, these alone do not suffice. An appropriate framework that articulates such combination is required. In this paper, we adopt a three-dimensional (information, concept, and inference) approach for such framework, based on OWL as formal language for terminological and ontological health resources, SNOMED CT as lexical backbone for all such resources, and the standard CEN 13606 for representing EHRs. Based on such framework, we propose a novel form for creating and supporting networks of clinical terminologies. Additionally, we propose a number of software modules to semantically process and exploit EHRs, including NLP-based search and inference, which can support medical applications in heterogeneous and distributed eHealth systems.

1 INTRODUCTION

Healthcare is one of the most information-intensive sectors of European economies, expected to greatly profit from research in information and communication technologies. However, the general feeling is that, to date, health information technologies have been mostly the realm of enthusiasts and the computer wave has not yet completely arrived.

The European eHealth Action Plan¹ provides a mid-term roadmap for improvement of the Health sector. One of the most challenging issues identified addresses the interoperability problem between different e-health systems. Such problem is partially due to the exponential increase of the number of medical terminologies (SNOMED CT², MedDra³,

etc.), ontologies (GALEN⁴, FMA⁵, etc), and classifications of diseases and related medical events and concepts (ICD⁶, CPT⁷, etc.) that eventually need to interoperate with one and another. The same report highlights the need for standardization as the key piece to ensure interoperability in this roadmap. Making eHealth systems interoperable by means of consensual, standard data formats and protocols will allow for a significant step forward towards satisfactory healthcare, accomplishing a number of goals like improvement of the quality of patient care, reduction of medical errors, and therefore savings in terms both of human and economic costs. Experiences on semi-automated local health systems have shown a lack of underlying standards for data exchange, emphasizing as a result that the gap

lec.europa.eu/information_society/activities/health/policy

² www.snomed.org

³www.meddramsso.com/MSSOWeb/index.htm

⁴www.open-galen.com/index.html

⁵sig.biostr.washington.edu/projects/fm/index.html

⁶www.cdc.gov/nchs/icd9.htm

⁷www.ama-assn.org/ama/pub/category/3113.html

between consumer expectations and actual service delivery remains unabridged.

roadmap towards The leveraging the interoperability problem in eHealth has been favoured by significant advances in information technologies, especially with respect to knowledge representation and interoperability. Particularly, large effort has been invested on the field of semantic technologies, finally coming of age and entering the plateau of commercial productivity. The usage of ontologies⁸ as the main asset of semantic technologies is currently supported by a wide range of tools for ontology construction, storing, feeding, evolution and evaluation like Protégé9 or the NeOn Toolkit¹⁰. In addition, mature methodologies and standards allow for accurate scheduling of ontology and application developments in terms of effort, time, quality and finance resources.

As a consequence, the Semantic Web initiative has proved to offer a reliable solution for large scale integration and representation problems, which can contribute to significantly alleviating the interoperability problem in the Health domain. In parallel, standardization efforts are going on in various subdomains for electronic interchange of clinical, financial, and administrative information among health care oriented computer systems, for e.g. definition of communication standards (HL7¹¹) or information models for electronic health record 13606¹² (ISO/CEN norm and openEHR¹³ specifications). The work presented in this paper combines both approaches (uptake of standards and semantic technologies) to tackle the interoperability problem in the Health domain.

Complete information Health systems need to address three main dimensions (information, concept system, and inference)¹⁴, as well as their associated resources, which are developed independently. Each of these components comprises the model itself, knowledge about a given view of the domain, metadata, and interfaces to the other components. Figure 1 shows a version of this vision, instantiated with the solutions adopted in our approach, based on OWL¹⁵ as formal language for terminological and ontological health resources, SNOMED CT as lexical backbone for all such resources, and the

standard CEN 13606 (together with the ADL¹⁶ language for archetype definition). The information dimension deals with high quality, structured and timely data collection and representation, allowing building an information framework for electronic health records (EHRs). In the present work, we exploit archetypes as formalism for modelling the required structures for EHR and defining the context of the clinical domain where such records belong.



Figure 1: Components of a complete Health system (source: semantichealth.org).

In our approach, Natural Language Processing (NLP) support for analyzing patient records is part of the information dimension and uses the terminologies provided by the terminology server in the concept system dimension to identify and process the information contained in the records. On the other hand, we use NLP (Jurafsky & Martin, 2000) for extracting data and information from EHR (free text documents) for further processing. The NLP of the EHR uses the terminologies provided by the terminology server to identify and process the information contained in the records and enable inferences using the EHR.

The information dimension lies on top of the concept system dimension, which deals with all the available terminological and ontological resources and provides the other two dimensions with uniform access to such resources. We have addressed the problem of managing all these resources through the development of a terminology server, which consequently allows relating them in a terminology network.

Finally, the inference dimension exploits both the concept system and information dimension to discover new knowledge. Inference and semantic search through NLP interfaces are some of the most

⁸ en.wikipedia.org/wiki/Ontology_(computer_science)

⁹ http://protege.stanford.edu/

¹⁰ www.neon-toolkit.org

¹¹ www.hl7.org

¹²www.chime.ucl.ac.uk/resources/CEN/EN13606-1/N06-02 prEN13606-1 20060209.pdf

¹³ www.openehr.org

¹⁴www.semantichealth.org

¹⁵www.w3.org/TR/owl-ref

¹⁶www.openehr.org/drafts/ADL-12draftF.pdf

immediate functionalities on top of which applications of this vision can be implemented.

The remainder of this paper is structured as follows: Sections 2 provides an insight to the three layers of the model, section 3 illustrates the integrated use of the described infrastructure and section 4 concludes the paper by describing ongoing work.

2 THREE LAYER MODEL

2.1 Information Dimension

As introduced above, the information dimension deals with representing and structuring EHRs. In this work, we approach this dimension from three different and complementary perspectives:

1.) The aim of the ISO/CEN 13606 standard is to normalize the transfer of information between EHRs systems in an interoperable fashion, without specifying how to implement them. The reference model represents the general features of the components of the health record, how they are organized and the context information needed to satisfy both the ethical and legal requirements of the record. This model defines building blocks for a formal representation of EHRs. An archetype is the definition of a hierarchical combination of components of the reference model, which restrict it (giving the names, possible types of data, default values, cardinality, etc.), to model clinical concepts of the knowledge domain. These structures, although sufficiently stable, may be modified or substituted by others as clinical practice evolves.

The Archetype Definition Language (ADL) allows expressing archetypes. An archetype starts with a header section followed by a definition section and an ontology section. The header includes a unique identifier for the archetype, a code identifying the clinical concept defined by the archetype. The definition section contains the restrictions in a tree-like structure created from the reference information model. This structure constrains the cardinality and content of the information model instances compliant with the archetype. Codes representing the meanings of nodes and constraints on text or terms as well as bindings to terminologies such as SNOMED, are stated in the ontology section of an archetype.

2.) Processing EHRs requires handling structured, semi-structured and unstructured data. To process unstructured data, which is generally free text such as clinical notes taken by doctors and nursing staff during patient visits, tools are needed that work

automatically with the language and allow information to be extracted so it can be easily stored and consulted. This is especially important in processes related to Patient Safety.

A series of additional problems exist for working with reports in free text written by clinical personnel: heavy use of acronyms and abbreviations; spelling errors; and including information on people or organizations, which must be anonymized in order to comply with laws on health information. Furthermore, most of the works in this area focus on the English language, where specific resources in biomedicine can be found, as MESH, UMLS (Bodenreider 2004), etc. Nevertheless, in the case of other languages, such as Spanish, relevant studies dealing with hospital reports or clinical notes have not been carried out yet.

Herein we present MOSTAS: A MOrpho-Semantic Tagger, Anonymizer and Spellchecker for Biomedical Texts, in order to address these problems in the information dimension of eHealth systems. The main objective of MOSTAS is to analyze clinical reports in Spanish using the ontological and lexical resources available for the Spanish language in order to first, pre-process the clinical reports so that they can be anonymized, abbreviations and acronyms can be detected and expanded, medical concepts in the application domain can be detected. The system's output is an XML document with morpho-semantic information that will facilitate later information retrieval of these texts.

3.) The remaining module in the information dimension of our system deals with semantic search through NLP interfaces using conceptual knowledge. This module is oriented to implement main features of document indexing based on the exploitation of knowledge and ontological resources included in an integrated way in UMLS, as SNOMED and MeSH. For the design and evaluation of the NLP semantic search module, we have developed a basic system offering interconnection between health records and a set of scientific information and health news. Given a query in submitted by a person, it first retrieves a list of medical records ordered by relevance in three steps: i) the query is expanded using concepts included in a biomedical ontology (i.e.: UMLS); ii) medical records are ranked using a representation based on biomedical concepts; iii) then, the user can choose a record and the system will retrieve several lists of ranked documents in English: from Pubmed news, or from article abstracts.

2.2 Concept System Model

1.) Addressing the concept system dimension requires devising a way to deal with the interoperability problem between the available terminologies and ontological resources used by the different computer health systems. In our framework, we have approached this problem by developing a terminology server, an open platform for: 1) Normalizing pre-existing terminologies as OWL ontologies, 2) Importing available ontological resources into the server, 3) Relating all these resources with each other in a terminology network, where equivalent terms are connected, and 4) Visualizing and browsing such network. By open, we mean that the terminology server is fully extendable with new terminologies, which can be plugged in as desired. Currently the following terminologies have been integrated into the system: ICD-9, official classification of the WHO¹⁷ for diseases and health problems, CPC-2¹⁸, international classification for primary care, SNOMED CT, the most extensive terminology in medical terminologies, Local Terminologies, containing terms used by hospital clinic personnel in their patient records and notes. Using OWL (the W3C standard ontology language) for representing normalized medical terminologies is accompanied by a large number of advantages. In a nutshell, these can be summarized as: high expressivity, reasoning capabilities, inference, and a wide tool and infrastructure support favoured by its status as a standard and the ongoing contributions from the knowledge engineering community. Additionally the number of medical resources available in OWL is dramatically increasing. As a consequence, in this work, we have adopted OWL as the reference language for medical terminology. As part of our approach, it was necessary to translate legacy terminologies into OWL in order to incorporate them into the terminology server. The most relevant case is SNOMED CT, which required a specific treatment due to its size and complexity. The SNOMED CT terminology is distributed across three text files: one containing the English terms (>300,000), another for term names in other languages (Spanish in our version) plus their corresponding preferred term and synonyms, and a third one describing the SNOMED CT taxonomy and the relations between the terms (>1.000,000).

In the translation process, we neither tried to

improve the overall quality of SNOMED CT (though several errors were detected (Schulz et al, 2007), (Rector, 2007)) nor did we modify the concept names. The resulting OWL file contains the most relevant parts of the terminology without extending its semantics.

2.) The terminology server follows a three layer architecture (Figure 2). The lower level stores, maintains and provides access to the terminologies, in their OWL forms, currently stored in a Sesame repository¹⁹. The same level allows managing metadata about the terminologies, like e.g. the terminology subdomain, the authoring institution, a short description, etc, which can be useful during search. The middle layer contains engineering implement components that three basic functionalities on top of the lower layer: i) search of terms both in a single terminology or across several, ii) mapping related terms of different terminologies, and iii) term visualization and browsing in and across terminologies. The higher level of the architecture contains the GUI components of the user applications exploiting the functionalities provided by the underlying layers of the terminology server. The GUI components can access such functionalities either programmatically, via a Java API, or in a loosely coupled way, through web services.



Figure 2: Terminology server three layer architecture.

3.) Currently, the terminology server provides two different types of search: The identifier-based search, where the user types the code of the term in a before specified terminology and the name-based search, where the user types the name of the concept or a part of it. As a shared characteristic, both searches return the sought term (if any) and situate it in the terminology network, showing the terms

¹⁷www.who.int

¹⁸www.globalfamilydoctor.com/wicc/pagers/english.pdf

¹⁹ www.openrdf.org

related with it in the other terminologies. This search scenario requires the definition of a terminology network, where the different terminologies are connected by means of related terms. For example, "Acne disorder" in SNOMED CT corresponds to "Acne" in ICD-9. Currently, in order to support users in defining the mappings of a term against the corresponding terms of the remaining terminologies of the network, we first search its name in those terminologies. Then, we use SNOMED CT as a terminology gateway in case no such term name is detected, i.e. we automatically fetch its synonyms from SNOMED CT and then search the synonyms instead. If still no solution is found, the terminology server tokenizes the term name, discarding stopwords and starts a new search with these tokens and their synonyms. Future work includes using ontology matching²⁰ techniques that exploit the semantics of the terminologies in OWL.

2.3 Inference Model

This section broadly reports on an approach to convert ADL definitions to OWL and then attach rules to the semantic version of the archetypes.

Let us consider the following situation. A health care information system that receives some OBSERVATION (e.g. "blood pressure") entry (no matter where from but fulfilling an archetype specification) is able to syntactically understand such information and therefore may deliver it to a professional, who proceeds with the OBSERVATION assessment. This is clearly a great advance for the interoperability of medical systems but it would be even more interesting if the observation archetype could tell not only how to manipulate observation's values but also how to assess and evaluate them. Every task that depends on data analysis and conclusion arrival usually requires the presence of an expert with enough knowledge to make a good decision. However if we separate tacit from explicit knowledge then we could add the latter in the archetyped concept so the expert only needed to deal with the former one.

Unfortunately, the ADL language does not provide support for rules and inference which are important pieces of clinical knowledge. Besides, while one of the greatest advantages of two-level modelling (Beale, 2002) is the carrying out of archetype definition as a decentralized process, it allows for contradictory viewpoints to coexist or even false information to be provided. In addition, a higher level of normalization of clinical knowledge could be achieved, encouraging for automated means to reuse knowledge expressed in the form of rules, which follows the same philosophy of sharing archetypes.

SWRL²¹ is a W3C recommendation developed to improve OWL limitations, in terms of inference, by means of rules. In combination, they add considerable expressive power to the Semantic Web. Furthermore, by merging SWRL rules with OWL ontologies, we will be able to partially automate decision making process.

Concretely, the complete knowledge workflow, from archetypes to inference, can be summarised as follows: 1) Translating ADL to OWL, 2) Mapping clinical data to OWL instances, 3) Adding SWRL rules to the ontology, 4) Executing inference.

When translation is finished, the obtained ontology file should be filled with instances of concrete clinical data. Depending on the nature of the data source, an adequate access approach should be chosen to correctly map each field to individuals' properties. From our perspective, preferred source will be the one where supplied XML files are compatible to the Reference Model syntax. In this case, instance mapping is a straightforward process.

As a particular implementation, here we adopt an inference process based on the Jess-Java bridge provided with the Protégé ontology editor (Golbreich and Imai, 2004). The Protégé SWRL Editor is an extension to Protégé-OWL that permits interactive editing of SWRL rules. It generates OWL files that include attached SWRL expressions.

The resulting OWL file, enriched with inferred knowledge, has many possible destinations. For example it can be directly delivered to the end user through a compatible interface or stored in a repository. In the clinical domain, these results provide means for automatically improving decision making and monitoring tasks.

3 INTEGRATION

The following example, focused on preventing pressure ulcers in hospitalized patients illustrates an integrated, practical use of the three-dimensional (Information, Concept, and Inference) architecture described in this paper. Pressure ulcers are a severe problem for bedridden patients caused by many different reasons like friction or humidity, which, not treated in time can become live-threatening. The goal of the hypothetical system described in this example is to automatically produce an alarm if a risk of ulcer is detected for any patient.

²⁰ www.ontologymatching.org

²¹www.w3.org/Submission/SWRL

First, we define an archetype in the CEN 13606 standard (Information Model) by using the knowledge of the clinical personnel of this concrete domain (Inference Model), see Figure 3. The archetype contains all the information related to the field of pressure ulcers and defines rules for identifying possible risks (for example: If the patient is not able to leave the bed, the risk increases²²). The next step is the linking of the CEN standard with a reference terminology, in our case SNOMED CT (Concept System Model).

Now, the risk-detecting protocol can start: The system is periodically fed with information about the patients, contained in clinical databases. The terms contained in such information are processed by the terminology server, expanded using the mappings defined across the available terminology network. This allows the NLP systems to detect occurrences of the words in the EHRs, which are also corrected if they had been previously misspelled or abbreviated.

With the information found in the EHRs the rules built as an extension of the OWL ontologies corresponding to the ADLs resulting from implementing the archetype, are immediately triggered and, if a risk of pressure ulcer is detected the alarm is triggered.

Additionally, semantic search may offer information from different EHRs and scientific or reference documentation related with the particular case on hand, facilitating decision making to the clinical personnel.



Figure 3: Example for the interactivity.

4 CONCLUSIONS AND ONGOING WORK

The interoperability problem in eHealth can only be addressed by means of combining standards and technology. An appropriate framework that articulates such combination is required. In this paper, we adopt a three-dimensional (information, concept, and inference) approach for such framework. Based on this framework, we have proposed a novel form of relating the different terminologies with each other by means of a terminology server that supports a clinical terminology network. On top of that, we have also proposed a number of modules to semantically process and exploit EHRs, including NLP-based search and inference, which support applications like e.g. automatic detection of pressure ulcers.

Nevertheless, all this work is still preliminary, and we are addressing further tests and evaluation in real-world systems. Ongoing work lies in this direction, aiming to demonstrate our approach for e.g. personal health records. Furthermore, we will continue the integration of semantic technology in this framework, especially in the concept dimension, incorporating novel ontology modularization, mapping, and context technology in order to facilitate management of complex and large terminologies as in the case of SNOMED CT.

ACKNOWLEDGEMENTS

This work has been funded as part of the Spanish nationally funded projects ISSE (FIT-350300-2007-75) and CISEP (FIT-350301-2007-18). We also acknowledge IST-2005-027595 EU project NeOn.

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²²http://www.sanitariascaligera.com/index.php?option=com_cont ent&task=view&id=44&Itemid=69&lang=en