Towards the harmonization of Clinical Information and Terminologies by Formal Representation

Catalina Martinez-Costa¹, André Queiroz de Andrade¹,², Daniel Karlsson³, Dipak Kalra⁴, Stefan Schulz¹,⁵

¹Institute for Medical Informatics, Statistics and Documentation, Medical University, Graz, Austria
²Information Science Graduate Program, School of Information Science, Federal University of Minas Gerais, Belo Horizonte, Brazil
³Department of Biomedical Engineering, Linköping University, Linköping, Sweden
⁴The Centre for Health Informatics and Multiprofessional Education, University College London, United Kingdom
⁵Institute of Medical Biometry and Medical Informatics, University Medical Center, Freiburg, Germany

Abstract

Background: Semantic interoperability of clinical information is the capability to recognize and process semantically equivalent information in a homogeneous way, even if data instances are heterogeneously represented, i.e. differently structured and/or using different terminology systems. Ontologies and terminologies are recognized as playing an important role for achieving this task. Objectives: The aim of this paper is to present an approach for enhancing clinical information by semantic annotations that makes it semantically interoperable. Methods: We propose an ontological framework rooted in an expressive upper level ontology. It is used to add standardized semantics to clinical information by combining EHR information entities with SNOMED CT. BioTop, the upper level ontology used will help to constrain the ways in which information and clinical entities can be combined. Results: We exemplify how the same clinical statement is heterogeneously represented depending on the EHR standard or terminology used. The same statement is represented according to the ontological framework proposed. Strengths and limitations of the approach are discussed. Conclusions: Our approach constitutes a first step towards interoperable representations of patient data in health records by using a shared framework using formal ontologies based on description logics. Such framework is required for the correct exchange of clinical information from heterogeneous sources, providing clear and unambiguous meaning of medical terms and the context in which they are used.

Keywords

Semantic interoperability, EHR (electronic health record), clinical terminology, ontology, formal ontologies, archetypes

Correspondence to:
Catalina Martinez-Costa. PhD
Institute for Medical Informatics, Statistics and Documentation.
Medical University Graz
Address: Auenbruggerplatz 2
8036 Graz. Austria
E-mail: catalina.martinez@medunigraz.at

EJBI 2012; 8(3):3-10

1 Introduction

It is of increasing importance for clinical information not just to be communicated between different EHR systems, but to be richly understood by the receiving system, so that the clinical meaning of EHR data imported – or queried – from elsewhere can be interpreted by computers as well as by humans. The SemanticHEALTH report [1] defines the ultimate goal, full semantic interoperability, as the ability for received EHR data to be combined seamlessly with local EHR data and processed homogeneously.

Computable semantic interoperability is vital to ensure that clinical applications, decision support systems, guideline and care pathway systems can all consistently and correctly interpret data derived from heterogeneous EHR systems, and provide appropriate and safe recommendations and alerts to support patient care. This consistent interpretation is also needed to enable the longitudinal analysis of individual patient’s EHRs to identify trends: improvements in health states, deteriorations or safety critical health situations.

Populations of health records need to be queried to
learn how best to optimise care pathways, to evaluate interventions, to improve outcomes, to help efficiently manage health services and underpin public health surveillance and measures. Patients will increasingly benefit from smart applications that help them to expertly manage their own long term conditions, and also to prevent illness and promote wellness. Clinical research will increasingly leverage large scale repositories of EHRs to complement the findings of prospective clinical trials, which will rely upon the scientifically valid analysis of data derived from heterogeneous systems.

Although much of the investment in EHRs has been at national levels, the challenge of semantic interoperability is a global one, not only to support cross-border health care but to support large scale multi-national research and valid international comparisons. However, although multiple initiatives have been provided by different Standards Development Organizations (SDOs), major challenges remain unsolved. As it can be derived from the previous semantic interoperability definition, in order to enable semantic interoperability, it is necessary to univocally identify clinical information in the different systems, i.e. to identify its meaning. Nowadays there are three layers of artifacts to represent the meaning of clinical information:

- Generic reference models for representing EHR data such as the provided by ISO 13606 [2], openEHR [3] and HL7 RIM [4].
- Clinical terminology systems such as LOINC [5], ICD [6] and SNOMED CT [7], the latter being increasingly based on formal-ontological principles and logic.
- Archetypes and HL7 templates as instantiations of generic reference models, tailored to the needs of structured data acquisition.

In the final report of SemanticHEALTH it is stated that sharing clinical meaning does not automatically imply identical terms and data structures: different physical or logical EHR representations may have a common meaning, i.e. they may be semantically equivalent. Therefore the goal of semantic interoperability is to be able to recognize and process semantically equivalent information homogeneously, even if instances are heterogeneous represented in terms of using different terminologies and information models, or different flavors of the same system, e.g. pre- vs. post-coordinated expressions in SNOMED CT. According to the same report, the use of terminologies like SNOMED CT, together with EHR standards should provide the way of identifying equivalent clinical information. In order to achieve this goal, SemanticHEALTH recommends that formal ontologies should play an important role to univocally represent the meaning of each information item and mapping semantically equivalent expressions within EHRs, within one language and between languages, thus supporting semantic interoperability. Formal ontologies consist of logical axioms that convey the meaning of terms for a particular community [8]. The set of logical axioms that define a term is named intensional definition and there is only one intensional definition per term for each community. In this way, ontologies are based on the understanding of the members of a community and help to reduce ambiguity in communication [9].

Throughout life sciences, ontology resources have increasingly been developed in the last years, and more and more experience in ontology theory and engineering has been accumulated in the (bio-)medical informatics community. OWL, the Web Ontology Language [10], supported by the Protégé editor, and several description logics reasoners have been established in biomedical ontology research and practice.

SemanticHealthNet is a EU FP7 funded R&D network [11], which takes up the results and recommendations of the above mentioned SemanticHEALTH project. SemanticHealthNet comprises representatives of international Standards Development Organisations like HL7, openEHR, and the EN 13606 Association [12], as well as WHO and IHTSDO, together with numerous recognised experts from academia and industry. It aims to generalise and formalise the methods and best practices in how to combine and adapt informatics resources to support semantic interoperability, and how these can be developed and supported at scale.

The authors are participants of SemanticHealthNet and propose an approach for achieving semantic interoperability based on the use of a common ontological framework that consists of upper level, domain and information ontologies, for representing clinical information.

Building an ontology is a complex and demanding task. In order to mitigate this, as much as possible, we believe that a formally rich upper level framework can guide and standardize, to a certain extent, the process of developing new domain ontologies. Upper level ontologies describe very general categories that are the same across all domains, such as Process, Quality and the like. These frameworks are easily reusable across specific domain applications. Examples of upper level ontologies are the Basic Formal Ontology (BFO) [13], DOLCE [14] or SUMO [15]. Under upper level ontologies, top-domain ontologies are defined as the application of the former ones to a specific domain, especially being more elaborated in some parts and less in others. BioTop [16] is one of these top-domain ontologies. Its reduced version BioTopLite [17] is being used in SemanticHealthNet for representing clinical information and relating information model entities with clinical entities.

2 Methods

One of the barriers to semantic interoperability is the lack of a strict separation between the information represented by terminologies and by information models. On the one hand, information models represent the entities in which information is recorded. They are (immaterial) in-
information entities like documents, headings, sections, entries, etc. Information entities refer to (types of) real entities, in the same way as words or images do. Just as in human language this should also take into account a kind of discourse where some information entity about X does not imply the existence of any instance of X. Examples of this abound in medical documentation. For instance, a child is suspected of bacterial meningitis and is treated as such although, retrospectively, there was only a viral infection. Nevertheless, the medical concept of meningitis is present in the physician’s deliberations just as it is documented and coded in the record, albeit in the context of suspicion.

This shows that information entities as used in clinical documents not necessarily refer to entities of proven existence on the side of the patient. They are often about states of human knowledge (e.g. what a physician thinks, hypothesizes, plans), and subsequent speech acts that manifest themselves in record entries with words like suspected, confirmed, order etc. Other abstract entities typically represented by information models are observation results, timestamps, or URLs.

On the other hand there are clinical codes that denote types of clinical entities (concepts\(^a\)). They often come with intensional definitions and have particular entities in their extension. In SNOMED CT, e.g. code/term combinations like 372244006 [Malignant melanoma (disorder)] represent the class of all (individual) melanomas. The axioms attached convey invariant features for an entity to qualify as a melanoma.

It is tempting to draw, in theory, a clear line between these two types of semantic artifacts. In reality however, clinical terminologies and classifications include code with an intended meaning that conveys how the clinical reality is perceived, measured, and understood by health professionals (i.e., they blend ontological with epistemic aspects) [18, 19]. An example of such overlapping is the SNOMED CT concept 395099008 [Cancer confirmed (situation)]. In that case cancer is a disease (i.e., a clinical entity), whereas the information that it is confirmed should be an expression of the diagnosis of such disease (i.e., an information entity). Another example would be the SNOMED CT concept 160244002 [No known allergies (situation)] in which allergy is a disease (clinical entity) and the fact that they are unknown should be represented by information entities.

Both concepts are defined in SNOMED CT under the situation with explicit context concept model category. In this segment of SNOMED CT, concepts include epistemic information (i.e. what is known about a situation) such as Confirmed, mixed with contextual information such as the temporal reference (e.g. past), and the reference to a clinical concept, which may or may not be instantiated in the particular case (e.g. cancer or allergy). But not only exists this overlapping in the terminology side, but also in the information model one.

For instance in the openEHR or HL7 information models we can find entities such as OBSERVATION or EVALUATION, that have some implicit clinical meaning. An example of information models including both epistemic and ontological information is shown below (see Figure 1) with an excerpt from the openEHR diagnosis archetype [20]:

The information model element “ELEMENT[at0002.1]” and the information model cluster “CLUSTER[at0011]” should together specify, possibly by means of reference to classes in a terminology, the focus condition of the diagnosis. On the other hand, the element “ELEMENT[at0.32]” specifies the epistemic status of the diagnosis, i.e. whether the diagnosis is provisional or a working diagnosis. Thus, as in the case of terminologies mixing representation of clinical entities with contextual and epistemic information described above, also in information modelling the boundary between representation of clinical and information entities is unclear.

The problem how to combine both, information models and terminologies, in an interoperable way has already been addressed in other works. The HL7 community, as a result of the Terminfo Project [21], provided a guide for the use of SNOMED CT in the HL7 V3 Clinical Statement pattern. Also in [22], the difficulty to bind clinical information models to terminologies was highlighted and a tool that included a system for binding openEHR archetypes to SNOMED CT was presented [23]. The UK National Health System (NHS) has also developed a guide on terminology binding in the context of the Logical Record Architecture (LRA) project [24] for use in an EN13606 based logical model.

As participants of the SemanticHealthNet project, we consider the accumulated ontology engineering and upper-ontology creation experience as a useful resource to demarcate the border between the information represented by terminologies and by information entities and therefore we propose the following ontological framework, consisting of information entities, domain and top-level ontologies (see Figure 2).

In order to relate the domain ontology with the ontological representation of information entities, the top-level ontology BioTopLite [17] will help constrain the way in which information and domain ontologies combine. As domain ontology parts of SNOMED CT will be used. Its concepts will be represented as subclasses of the top-level ontology categories. Finally, the information entities ontology will describe the contents of documents such as the contextual and epistemic features of clinical information.
The ontological framework proposed will make use of the Semantic Web standard OWL DL, based on description logics, which will provide syntactic and semantic standardization and makes the resulting artifacts computable by description logics reasoners such as HermiT.

Moreover, the use of an ontological approach will make possible to clearly distinguish clinical entities (SNOMED CT concepts that represent invariant features of the medical reality) from information entities. Figure 3 illustrates such distinction and by using the relation isAbout, the latter ones can be formalized in terms of the former ones.

Figure 3: Information entities vs Clinical entities

Opposed to clinical entities (which instantiate clinical SNOMED CT concepts), Document and Document Item denote information entities that come out of certain clinical actions, such as observations, investigations, or evaluations. A Document will contain the information provided in a specific episode of care, we call healthcare situation. In contradistinction Document Items will be part of some Document and will reflect different clinical situations of the patient. Clinical situations are defined, following the current harmonization process between IHTSDO and WHO, as phases of a patient’s life during which he/she is bearer of some pathological conditions.

\[
\text{ClinicalSituation} \equiv \text{processualPartOf some (BiologicalLife and (hasParticipant some (HumanOrganism and (locusOf some ClinicalCondition)))})
\]

If we apply the previous definition for representing the fact that a patient has pain in the chest, the following OWL expression will be defined:

```
EVALUATION[at0009.1] matches { - Diagnosis
data matches {
  ITEM_TREE[at0001] matches { - structure
    items cardinality matches {1..*; ordered} matches {
      ELEMENT[at0002.1] matches { - Diagnosis
        value matches {
          DV_CODED_TEXT matches {
            defining_code matches {[ac0.1]}
            - Any term that ‘is_a’ diagnosis
          }
        }
      }
      ELEMENT[at0.32] occurrences matches {0..1} matches {
        - Status
        value matches {
          DV_CODED_TEXT matches {
            defining_code matches {[local::
            at0.33, - provisional
            at0.34] - working
          }
        }
      }
    }
  }
}

CLUSTER[at0011] occurrences matches {0..*} matches {
  - Location
  items cardinality matches {1..2; ordered} matches {
    ELEMENT[at0012] occurrences matches {0..1} matches {
      - Body Site
      value matches {
        DV_CODED_TEXT matches {
          defining_code matches {[ac0000]}
          - Any term that describes a body site
        }
      }
    }
  }
}
```

Figure 1: Excerpt of the openEHR diagnosis archetype

Figure 2: Common Ontological Framework
ClinicalSituationWithChestPain equivalentTo
processualPartOf some (BiologicalLife and
(hasParticipant some (HumanOrganism and
(locusOf some (set_Pain and
hasLocus some set_Chest)))))

This is just an ontological clarification of what is implicitly meant by ClinicalSituationWithChestPain. It is still subject to discussion whether SNOMED CT finding concepts are principally to be interpreted as denoting situations (under the above definition). In this case, the whole expression would conflate into the SNOMED CT concept chest pain. In our example the pre-coordinated concept SNOMED CT chest pain has been used but this could have been expressed also by using a post-coordinated expression. The equivalence of both expressions would be identified by classifying the proposed ontology.

ClinicalSituationWithChestPain equivalentTo
processualPartOf some (BiologicalLife and
(hasParticipant some (HumanOrganism and
(locusOf some (set_Chest and
hasLocus some set_Chest)))))

A crucial aspect of terminology binding in our model is that DL expressions such as the above exemplified are NOT primarily used as classes of which instances would then constitute the “real” things such as a concrete situation which entails a concrete chest pain. This approach would collide with the need to represent the clinical discourse in all its nuances: the doctor records the patient’s alleged chest pain. He may believe it with a certain likelihood, but cannot discard that the patient feigned the pain. Or the doctor treats the patient just on the basis of a hypothesis, such as in the meningitis example discussed above. The following expression shows how discourse or document items about a clinical concept can be represented, without committing to the material existence of any instance of this concept:

DocumentItemAboutSituationWithChestPain equivalentTo
DocumentItem and
(isAbout only ClinicalSituationWithChestPain)

The operator ‘only’ expresses that if the document item is about anything, then it can only be about an instance of the class ClinicalSituationWithChestPain. Thus, the existence of a document on chest pain does not necessarily entail the existence of chest pain in the patient.

3 Results

In this section we will show how the same clinical information is represented heterogeneously depending on the EHR standards and terminologies used. More specifically we will show its representation according to openEHR, HL7 CDA [25] and SNOMED CT. Attention will be paid to the SNOMED CT bindings provided and the information embedded in the information model structures. We will compare these representations and then we will provide their definition in OWL by using the ontological framework presented in Section 2. Finally we will discuss some of the problems we found.

The example chosen is the following clinical information statement, “The patient is not diabetic”, extracted from the history section of a summary report. According to openEHR, this statement could be represented by using the archetype openEHR-EHR-EVALUATION.exclusion-problem_diagnosis.v1. Following, in Figure 4, an extract of its encoding is shown.

By means of a template the value of the “ELEMENT[at.09]” can be fixed to the default coded text value 73211009 |Diabetes mellitus/| being a reference to the SNOMED CT code 73211009 with the preferred term “Diabetes mellitus” for the sake of human readability.

Here we can observe how information entities may include some semantic as it happens with the openEHR ELEMENT entity that has the meaning no previous history of embedded. If the same statement is represented by using the HL7 CDA standard, the encoding depicted in Figure 5 will be obtained.

It can be observed how the fact that the patient does not have diabetes mellitus is defined inside a problem section template, as an observation, a kind of HL7 ACT entity, whose value is the following SNOMED CT post-coordinated expression:

373572006 |clinical finding absent| : {
246090004 |associated finding —
73211009 |diabetes mellitus|
}

It is noteworthy that the treatment of negation is fundamentally different from openEHR. Whereas in openEHR the whole archetype embodies the negative meaning, HL7 does not allow, for safety reasons, any negation semantics within the information model. This requires that the target terminology caters for negation, which explains the need of a post-coordinated SNOMED CT expression as demonstrated.

SNOMED CT, due to its hierarchy of “context-dependent categories” provides concepts for expressing information entities such as document sections, thus blending information entities and domain-ontology aspects (see Figure 6):

Our examples demonstrate that for the same piece of information there are different possible representations: in openEHR the negation is expressed by the archetype proper and the clinical meaning is provided by the binding to one simple SNOMED CT code; in HL7 CDA it is achieved via the binding to a complex SNOMED CT expression that embodies the negative meaning. The latter is also the case with the SNOMED only encoding, where the SNOMED CT expression from the second example is wrapped into a contextual information entity, using the concept 422625006 |history of present illness section| from SNOMED CT.

How can we make sure that the three encodings have
Figure 4: OpenEHR: “The patient is not diabetic”

Figure 5: HL7 CDA: “The patient is not diabetic”

Figure 6: SNOMED CT: “The patient is not diabetic”
equivalent meanings? SemanticHealthNet will examine the role of an ontological framework for semantic annotations that can be generated out of the diverse information model / ontology combinations, and from which semantic equivalence should be computed automatically, using OWL-DL as a language that allows a logic-based rendering of the information, which then can be submitted to description logics classifiers for testing semantic equivalence. In this framework the fact that a person does not have diabetes mellitus in their clinical history will be expressed by means of a complex semantic annotation of the information. This piece of information that states the absence of diabetes in the history is identified as the instance (or annotation) of an OWL class, here called (DocumentItemAboutSituationWithoutDiabetes), which is, ontologically, a specialization of the class InformationEntity. By means of the isAbout relation it is related to a logical expression that defines the clinical situation under scrutiny (ClinicalSituationOfPersonWithoutDiabetes):

ClinicalSituationOfPersonWithoutDiabetes equivalentTo
  processualPartOf some (BiologicalLife and
  (hasParticipant some (HumanOrganism and
  (not (locusOf some set_Diabetes)))))

DocumentItemAboutSituationWithoutDiabetes
  (abstractPartOf some Document) and
  (isAbout some
   ClinicalSituationOfPersonWithoutDiabetes)

In order to obtain this ontological representation, some patterns and mapping rules have previously to be defined at the level of the archetype or clinical models. For instance that the SNOMED CT concepts finding with explicit context instead of findings must be used when binding HL7 documents and archetypes to SNOMED CT. Clinical finding present, clinical finding absent, clinical finding suspected or clinical finding unknown are subtypes of the concept finding with explicit context and they could be easily transformed to our ontological representation. For instance, this would be the representation for the case clinical finding absent:

DocumentItem and isAbout some (Situation
  and not associatedFinding some ClinicalCondition)

In the above representation the relation associatedFinding has been used for simplification reasons but it is defined as:

associatedFinding –
  processualPartOf o hasParticipant o locusOf

As a result, all meaning-bearing elements in clinical documents would then be annotated by logical expressions in OWL-DL, which refer to one or more SNOMED CT concepts. The equivalence of such expressions could then be verified by DL reasoners.

However, there are some issues that have to be better studied as the representation of the context. The statement that the patient has no diabetes mellitus does not say anything about its precise context. Questions such as “does it refer to the result of some examination that has just been done?” or “is it part of his/her past history?”, have to be answered. In the representations above provided, the context has been expressed also in different ways. In openEHR the context was embedded in the information entities (No previous history of) as well as in HL7 CDA where it was included in the problems section. In SNOMED CT a concept from the hierarchy record artifact was used (history of present illness section). Therefore, the context of the finding has to be clearly identified and in order to do it it is necessary to establish some patterns that will depend on the EHR standard used.

4 Conclusions

Clinical information representation and interoperability between health systems have been a study subject for several decades in the medical informatics community. After several years of standards development, the partially independent development of information model standards and terminologies created the new problem of unambiguously harmonizing both representations and that is one of the biggest barriers of semantic interoperability.

The border between the kinds of content that should be represented by the EHR standards information models and by clinical terminologies is blurred. For instance, in SNOMED CT the status of a diagnosis can appear as a pre-coordinated term (e.g. confirmed cancer) or can be represented in the information model with a specific status field. In order to constrain the ways in which terminologies and information models can be combined we have presented a common ontological framework based on the use of upper level, domain and information ontologies. Our experience is that formally rich upper level ontologies can guide and standardize the process of developing new ontologies. As domain ontology we have used SNOMED CT due to its increasing use as reference terminology. Selected SNOMED CT concepts together with information entities are introduced as extensions of BioTopLite upper level categories.

The approach we propose will annotate clinical documents with logical expressions in OWL-DL defined according to our ontology. These expressions will refer to one or more SNOMED CT concepts and the equivalence of different expressions will be then be verified by DL reasoners.

This work is part of the SemanticHealthNet project, which aims at improving the exchange of clinical data by developing new approaches to clinical information modeling. Our approach constitutes a first step towards interoperable representations of health records by using a shared logical framework based on ontologies. We think that such framework is required for the correct exchange of clinical information from heterogeneous sources, providing
clear and unambiguous meaning of medical terms. However, the SemanticHealthNet project has barely begun, and there are still many open questions. Current healthcare standards are huge enterprises that contain hundreds of different artifacts, with sometimes very particular and context dependent interpretations. The feasibility of creating rules for every possible EHR standard is not guaranteed. Also, this approach is using the automatic classification feature of OWL-DL, which is supported by current reasoners. Healthcare applications depend on near real time response for users, and complex logical inferences may be impractical for real use. We are exploring, in parallel, solutions to these shortcomings that will be presented over the development of the project. We are confident that a pragmatical and multi-disciplinary approach, aiming to find the best of both worlds, is the way forward to solve semantic interoperability issues.

Acknowledgements

This work has been funded by the SemanticHealthNet Network of Excellence within the EU Seventh Framework Program, Call:FP7-ICT-2011-7, agreement no.: 288408.http://www.semantichealthnet.eu/.

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