Building a Medical Ontology to support Information Retrieval: Terminological and metamodelization issues

Jean Charlet
AP-HP, Paris, France;
INSERM U872, Paris, France;
Jean.Charlet@upmc.fr

Gunnar Declerck
INSERM U872, Paris, France;

Ferdinand Dhombres
INSERM U872, Paris, France;
Université Pierre et Marie Curie, Paris, France;
Hôpital Armand Trousseau, AP-HP, Paris, France;

Patrick Miroux
Dpt Urgences CHU
Angers, France;

Pierre Gayet
Centre hospitalier de
Compiègne, France;

Pierre-Yves Vandenbussche
INSERM U872, Paris, France;
MONDECA, Paris, France.

Abstract

ONTOLURGENCES is a termino-ontological resource (TOR) developed for retrieving information in electronic Emergency Medical Record. This ressource describes the meaning of the relevant concepts of the field and the different labels of those concepts when they occur in documents. In this paper we show that: (i) the sustainability of such a resource requires a precise articulation between terms and concepts, and (ii) such a requirement can be met via the implementation of standardized procedures based on a meta-model architecture allowing the modeling of all necessary KOS and other knowledge structures.

1 Introduction

The use of terminological systems for the creation of ontologies raises several major issues (García-Silva et al., 2008). Obviously, ontologies and terminologies play a similar normative role. They aim at establishing a common vocabulary and make use of shared representations and concepts to allow the documents interoperability and facilitate knowledge building. However, ontologies and terminologies have clearly a different formal approach on Semantics. Ontologies are *concepts* architectures and are not organized lists of *terms*. Unlike terms, the concepts are characterized by *formal definitions*. The formal aspect enables the computerized treatment of the information. To use an ontology to normalize a document is, in that sense, to encode it by bringing a characteristic allowing the automated treatment of the information.

However, the creation of ontologies involves sometimes the use of terminologies, or even more radically, the use of corpus of text. If the ontology is to be integrated within an automated information treatment system, as for example the information retrieval (IR), the concepts should match with the terms appearing on the documents to enable the information treatment. The ontology should ensure the *coverage of the terminological domain*. The conceptual representation would otherwise be unusable.

The Lerudi (emergency services) Project intends to develop an Information System (IS) offering an overview of the Electronic Health Record (EHR) to the health professionals. Additionally, it aims at facilitating the quick reading of the EHR to allow quick medical decisions under tight time constraints. The field experimentation of that project is the reading of hospital files by an emergency regulating physician. Practically, Lerudi is IR system based on a Termino-Ontological Resource (TOR)[1] named ONTOLURGENCES. The TOR (a) plays the field model role by listing the relevant concepts; and (b) ensures the link between the concepts and their name in the EHR documents. This double function should not only enable an easy annotation and indexation of the patient files, but also facilitate the retrieval of information from the indexed records.

The ONTOLURGENCES development included 6 phases: (i) the building of the TOR ontological skeleton based on a corpus analysis method; (ii) the use of existing terminological and ontological

[1] A TOR is an ontology in which terms are linked to concepts in a systematic and exhaustive way. Several methods exist to link terms and concepts depending on the representation target (Reymonet et al., 2007).

resources to manually complete the TOR concepts system; (iii) the automatic enhancement and (iv) semi-manual TOR enhancement at the terms level; (v) the TOR enhancement of concepts in relation to the medicines; and finally, (vi) the implementation of validation and quality control procedures.

The first 2 phases of the TOR correspond to the usual ontology construction method, widely tried in our team and did not raise major issues. However, the 3 following phases that were specific to the TOR development were much more problematic. Specifically, the TOR terminological enhancement required external resources: *Knowledge Organization System* (KOS). These external resources are only useable in an architecture supporting a complex modeling of the target TOR. In particular, the architecture should accommodate the terms, the concepts and their interrelation, and simultaneously, the KOS used for the enhancement. The last stage corresponding to the quality control is also specific to this project and was necessary considering the various participants involved in the TOR construction.

Trough the detailed description of the process guiding this TOR construction and validation within a large team, we aim at showing that: (i) the sustainability of such a resource requires a concise articulation between terms and concepts; and (ii) such a requirement can be met via the implementation of standardized procedures based on a meta-model architecture allowing the modeling of all necessary KOS and other knowledge structures.

The rest of the paper is organized as follows: Section 2 briefly presents the advantages of using ontologies for retrieving information. The first two steps of the TOR construction and its specificities are presented in Section 3. Section 4 provides an overview of the UniMoKR model that enables the implementation of the TOR terminological and conceptual enhancement procedures; and its uses. Section 5 describes the TOR different enhancement phases[2]. The validation and quality control are detailed in the section 6. Finally, the paper concludes with a summary and a discussion in Section 7.

[2]Due to the lack of space, the step of conceptual enhancement of drugs branch of the ontology is not described in this paper.

2 Why using an ontology for information retrieval?

To begin with, we should ask ourselves what the point in using an ontology for an IR is. Specifically, the main advantage of an ontology is to allow an automated reasoning based on the conceptual structure and semantic relations between notions. Consequently, in addition to subsumption relations (*is-a formal relation*), we modeled the semantic relations between signs, diseases and medical specialties. These relationships enable an interface (*i.e.* a cloud of words) to display the medical specialties that characterize a given EHR.

An ontology for IR has also *de facto*, as any ontology, a structure that depends on the task (Charlet et al., 1996; van Heijst et al., 1997). This structure is not a quality in itself for the IR, but it nevertheless has two advantages: (i) a well-structured ontology is easier to maintain than a poorly structured ontology, (ii) a well-structured ontology enables valid reasonings. This second point is obviously expected from any ontology, but it is clear that it is not always satisfied. Another important property that has to possess an ontology for IR is the coverage of the terms relevant to express the notions of the target-domain. The following two examples will illustrate these points:

Example of the importance of the formal structure of the TOR. Considering the important following question that asks an emergency physician about a patient: “Has my patient already been infested by an enterobacteria in the past?”. Consider that the patient’s record contains a document annotated with the concept of “Salmonella”. For the system to conclude that Salmonella is an enterobacteria, it is necessary that the TOR specifies that the concept “Salmonella” has a transitive relation of specialization with the concept of “enterobacteria”. In this way, the answer to the question of the emergency physician will be positive, *even if the patient record is not directly annotated with the more general concept of enterobacteria*.

Example of the importance of the terminological coverage of the TOR. The annotation of the noun phrases “paracetamol”, “Dafalgan” and “paraml.” requires that the TOR has a unique concept representing these three syntagms

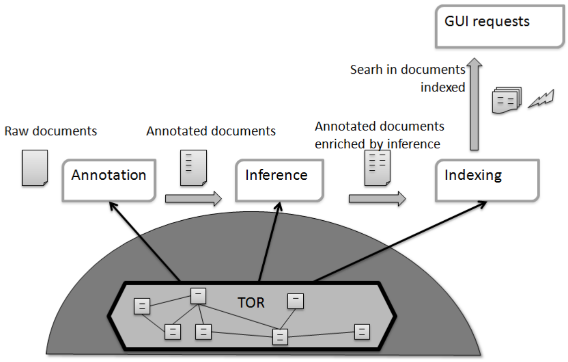

Figure 1: Uses of the TOR in the Lerudi project. The TOR supports the processes of annotation, indexing and inference.

and the availability of the terms related to the chemical molecule (paracetamol) and the proprietary drug or its brand name (Dafalgan).

It appears clearly that, the quality of the information displayed to the final user of the IR system crucially depends on the quality and richness of the TOR. The processes of annotation, indexing and inference rely on the formal structure and the terminological completeness (*i.e.* its capacity to cover the terms of the domain). The figure 1 below illustrates the different uses of the TOR in the Lerudi project.

3 Terminological and ontological resources used for designing ONTOLURGENCES

3.1 Domain of ONTOLURGENCES

ONTOLURGENCES has been built in several steps and by using different resources, and its target knowledge field has been clarified gradually. From the very beginning of the project, we realized that the knowledge field that had been originally set for the TOR (that is: the repertoire of concepts that had to be present in the TOR) had to evolve. We had left with the idea of building an ontology representing only the specific concepts used by the emergency physicians. But it turned out that, from the perspective of information retrieval in EHRs, such restriction a priori of the target knowledge field was a mistake. Indeed, the information system aims to allow the emergency physician to quickly find medically relevant concepts in EHRs. But these concepts can not be reduced to concepts specific to the medical emergency field, they can instead meet *any medical specialty*.

In the paragraphs below, we present the main phases of the development of ONTOLURGENCES and the terminological and ontological resources we have used. We do not discuss the problem of the organization of these stages and cycles of development. For this question, we may refer to (Dhombres et al., 2010).Suffice it to say that during the development process of ONTOLURGENCES, we followed the ARCHONTE method developed by B. Bachimont Bachimont et al. (2002).

3.2 The processing of textual data

In the ARCHONTE method, the domain ontology is built on the analysis of documents generated during the activity to be modeled. In our case, we have encountered great difficulties in accessing a corpus that could perform this function. The emergency services being not computerized, and the paper documents shorter and less numerous than in other services, it was difficult to find documents in sufficient numbers to make up the corpus in question.

Consequently, we used two other kinds of documents: the acts of the *Urgences* conference of the discipline and the *Guides to Good Practice*. Besides the difficulty we had to preprocess the corpus, the main problem was the coverage capacity of the corpus compared to the target. Indeed, the corpus of the conference proceedings, that was fully processed, has shown its limits in terms of scope. Conference papers are in many cases concerned with the "rare bird", that is with questions that are not representative of the problems that emergency physicians are confronted daily. A specific work has shown this clearly by comparing the terms most frequently detected in the corpus with the actual incidence of the emergency diseases (Gayet et al., 2010).

This issue of availability of the corpus should not be underestimated: in the areas where we can base the construction of the ontology on a corpus analyzed by tools of natural language processing (NLP), resorting to existing terminologies operates in the validation process of the work having been done. In the case of interest here, they occur much earlier in the development process.

3.3 Reusing the specialty thesaurus

For the PMSI[3] coding, the emergency physicians make use of an CIM-10 extract which contains about 1,000 terms. These terms covering an important part of the terminological repertoire used by emergency physicians for coding, it appeared necessary to incorporate them in the ontology. Consequently, a concept was created and defined for each of them.

One of the major limitation of the project is the fact that the CIM-10 terms are suitable for coding, but some of them are difficult to manage in an ontology because they encompass several heterogeneous concepts. For example, one can find terms such as "subject waiting to be admitted elsewhere, in a suitable establishment" or "Symptoms and signs involving cognitive functions and consciousness, other and unspecified". The concepts associated with such terms, because they articulate in a complex way a multitude of heterogeneous concepts, are difficult to model.

3.4 Reusing the CCAM

The french CCAM classification (commune classification of medical acts) has the benefit of having been designed by teams familiar with ontologies. Which means a priori that each concept of this classification has been validated by a formal representation (Rodrigues et al., 1999). The reuse of the CCAM thus enabled us to incorporate a classification made up in accordance with consistent principles to our TOR.

The problems rather came from the way the CCAM is organized and designations used for the acts, which are built for specified accounting policies and not at all suitable for their expression in medical documents - our target. Much work has thus consisted in renaming the terms associated with concepts (*cf.* § 3.6).

3.5 Reusing the SNOMED V3.5

The creation of the branch of diseases concepts is always a major part in the constitution of medical ontologies. As the necessary corpus for the design of such a branch were not available or did not cover the whole area, we decided to complete the work by integrating in ONTOLURGENCES the diagnoses branch of the SNOMED V3.5[4]. This procedure was mainly carried out by physicians and required more than 100 hours of work: The SNOMED V3.5 was notoriously too specific - what could be expected - but appeared also very badly organized - which was quite surprising. From the 25,000 diseases present in the The SNOMED V3.5, 6 500 have been preserved.

3.6 Additional methodological comments

To complete the description of the construction of ONTOLURGENCES, a few points of clarification are further needed:

1. ONTOLURGENCES was developed with the OWL2 description logic (DL) language and with the Protégé ontology editor;
2. The SKOS[5] language was used for the formalization of the terms. The SKOS language is representation language for knowledge organization systems such as thesauri, taxonomies, or any other type of controlled or structured vocabularies. This standard provides some primitives dedicated to the terminology with for each language, a preferred term `skos:prefLabel`, synonyms `skos:altLabel` and a definition `skos:definition`. Those primitives belonging to a standard commonly used are suitable for the representation of names and synonyms of the concepts of the ontology and can be perfectly mobilized within an ontology described in OWL.
3. The resources used in the construction of ontology are diverse. As far as possible, we memorize the origin of the concepts with an annotation that specifies the identifier of the concept in the original resource, *SnomedId*

[3] The french Information System Medicalization Program (PMSI) is intended to introduce concepts of analytic accounting in the administrative management of hospitals: diagnosis and procedures performed in a health facility are coded and recorded, reported to a patient and to the various costs in the structure.

[4] The SNOMED V3.5 is a multiaxial classification whose development has been initialized by Canadian anatomopathologists. Its aim is to represent the whole domain of medicine and related notions of society. It contains 105,000 concepts. SNOMED V3.5 exists in French and was chosen as the *reference terminology* by the French government (Rosenbloom et al., 2006). An ontology, the SNOMED-CT, has been derived from this classification by successive reorganizations and integrations of other terminologies. SNOMED-CT is not entirely available in French.

[5] The *Simple Knowledge Organization System* (SKOS) is developed within the W3C since 2003.

for SNOMED V3.5 or *FmaID* pour the FMA (*Foundational Model of Anatomy*).

4. The concepts of the ontology can be distinguished among those used for IR and the others. The latter are either high-level structuring concepts – *e.g.* IntentionalObject – or medical concepts too general to be discriminating – *e.g.* PhysicalExamination. This feature is described via a boolean annotation – terminologicalConcept – which specifies if the concept has a "terminological" character (it is potentially useful for IR) or not.

4 Meta-modelize to support enhancements

The ONTOLURGENCES ontology provides a conceptualization of the emergency field with terms to designate its concepts. This conceptualization can benefit from (i) the terms present in the KOS of Health to increase the detection of concepts in documents processed and from (ii) specific concepts about drug molecules in the ATC classification. To develop this new resource, you must be able to represent the KOS and ontology at the same level of description. Indeed, these resources are available in different formats and languages.

4.1 The UniMoKR metamodel

The diversity that exists in the nature, representation, and organization of the knowledge can be explained by different pasts, objectives, and uses. However, these KOS always intend to grasp information, to share it, and to support the human and computised processing. Thus, it is possible to extract a common model core from this obvious heterogeneity (i.e. a model common to all knowledge structuring). In the field of knowledge organization system representation, some norms and standards are in place and facilitate the interoperability (Miles, 2006; Clarke, 2008). Although SKOS and BS 8723 allow terminologies representation, none of them adress the issue of concepts group in a satisfactory manner[6]. We reuse in this project, the UniMoKR model designed in our previous work (Vandenbussche and Charlet, 2009)[7]. This model uses and extends modeling elements from SKOS, BS 8723 and is already used by research and commercial projects (Joubert et al., 2011; Vandenbussche et al., 2013).

The Termino-Conceptual part of UniMoKR model describes the relation between a *Concept* and its related *Preferred Term* and *Simple non preferred Terms* (aka synonyms) in each language. The Group Part enables not only the representation of a whole terminology, but also the representation of a terminology subset. It allows two different ways to characterize membership: by intension (concepts have to meet the restriction requirement to be part of a group; all concepts answering this request are implicitly members of the group) and by extension (concepts have to explicitly refer to this group via the relationship *inGroup*) Our modeling reified the SKOS original alignement relations and allows alignments representation generated by various sources as well as the representation of the associated metadata information. Finally, meta-classes intend to guaranty the UniMoKR model extensibility and to facilitate its re-use and adaptation: some artifacts particular to some terminologies are not taken into account in UniMoKR; however, they need to be represented to avoid the loss of information.

5 Linguistic enhancement of the ontology

As mentioned above, for the Lerudi information system to be operational in situation, it is necessary that the TOR ONTOLURGENCES covers almost all linguistic forms under which medical concepts relevant for emergency decisions appear in the EHR the system will have to deal with. Ultimately, the system must also be able to accommodate the "shortcuts" and "imperfections" of the language in which patient records are written, which for instance make use of abbreviations or may simply contain spelling errors.

The overall Lerudi system works as follows: the text of the various documents comprised in the EHR is processed by an algorithm that seeks to establish a correspondence (if necessary, by integrating NLP methods) between the phrases (treated as mere strings) and the system of concepts of the TOR. If a string has been matched with a concept, the concept will be used to index the document.

Now, medical records are usually written in natural language (or at least in this semi-standardized language suitable for concrete medical activities),

[6]For instance SKOS and BS 8723 models can not cope with SNOMED CT value sets or any concept groups defined in intension (`http://schemas.bs8723.org/`).

[7]The model is accessible at `http://bit.ly/15azC7k`

for the semantic interpretation process to reach a satisfactory level (or an optimal one: the optimum being set by the performance attained by an emergency physicist), it is often necessary to have available all lexical variations (synonyms, short forms, etc.) that may present the textual form of the concept. If a form encountered in the EHR has not been specified in the ontology, the record will not be indexed with the corresponding concept. The medical term will not be displayed by the interface. The emergency physicist will then have to put up with an incomplete or incorrect information.

To overcome this problem, two terminological enhancement processes of the TOR have been performed: (i) an automatic enhancement of the TOR by the adding of terms extracted from various KOS; (ii) a semi-automatic enhancement of the TOR by the adding of noun phrases extracted from the EHRs.

5.1 Enhancement of the TOR through the alignment with KOS

A first version of the enhanced TOR is realized through the aligment of the emergency domain ontology with few KOS relevant for the field, including CIM-10, SNOMED 3.5, MedDRA, ATC. By providing a controlled vocabulary, the KOS support the functions of analysis (annotation) of the EHRs. But, du to the difficulty to validate alignments, we decide to keep just the alignment to SNOMED V3.5. The alignment was performed with the alignment software ONAGUI (Mazuel and Charlet, 2010)[8] and by manually validating all the automatic alignments made.

Finally, during an export phase, the TOR, now optimized for annotation and indexation, is made available in the SKOS format. Once the concepts of the ontology enhanced with lexical forms from the KOS, the representation model of the TOR is converted to SKOS. This operation of conversion is performed with the model transformation method described at the section 4.1.

5.2 Enhancement of the TOR through the analysis of noun phrases

To improve the terminological completeness of ONTOLURGENCES TOR, a complementary semi-automatic enhancement procedure was introduced. This procedure incorporates the principles of the *bottom-up* methodology used by the designers of domain ontologies. It includes the following steps: (i) we first analyze with NLP tools the content of the documents produced by the operating health professionals (*i.e.* the EHRs), in order to extract (this time by mobilizing statistical methods) the noun phrases likely to be among the most structuring of the considered field of knowledge, that is the terms that are specific and essential to the field; (ii) Once these terms are identified, health professionals (emergency physicians): A) perform a filtering operation to retain only the terms actually belonging to the medical field and likely to be clinically relevant during the process of IR in EHRs and B) validate the relevance of the identified synonymous terms; (iii) these terms are then: A) added as synonyms (`skos:altLabel` tag) when they meet medical concepts already present in ONTOLURGENCES TOR, or B) converted into new concepts, when they refer to notions that do not yet have a conceptual representation in the TOR (in that specific case these terms correspond to the so-called *candidate-terms* of the *bottom-up* methodology. This conceptual conversion step requires to produce a formal definition of the concept being considered, which means firstly positioning the concept in the existing ontological hierarchy.

[8] `http://sourceforge.net/projects/onagui/`

6 validation Processes

6.1 Why using validation procedures?

After one year of work, it appeared that the implementation of control procedures was necessary to maintain the quality of ONTOLURGENCES TOR, and that these procedures had to be replayed regularly. Indeed, (i) many stakeholders, physicians as well as modelers, are working together on the ontology, and despite all our efforts, we have not always been able to correctly apply the guidelines for the maintenance of quality and the homogeneity of the TOR. In addition, (ii) many instructions are binding and a person may apply them one day and forget them another.

In a first step, these procedures do not address the structure of the ontology. The main reason is that at this level of development of the TOR and given the skills of the team, the problems we encountered were first terminological problems. But it is clear that problems of structuration, also present, call for future treatments (*cf.* 7). Our procedures are based on patterns, or anti-patterns when managing mistakes to be avoided. This work falls under the current research area concerned with the control of the quality of ontologies, as can be read on more structural points in (Roussey et al., 2010) or (Rector et al., 2004).

6.2 Which meta-model?

The quality control procedures were designed to ensure that the TOR meets the criteria of a specific meta-model. As far as this part is concerned, the meta-model can be expressed by the list of following rules:

- Each concept must carry an annotation ¡*terminologicalConcept*¿ in Boolean format;
- Each terminological concept (*cf.* previous rule) must have one, and only one, `skos:prefLabel` in French.
- Each terminological concept must have zero or one Skos:prefLabel in another language. The other relevant languages are: English, for the communication, and Latin, widely represented in the etymology of medical concepts;
- Due to the IR algorithms functioning, two different concepts must not have the same `skos:prefLabel` or the same `skos:altLabel` (string of identical characters);

6.3 The procedures

The procedures are implemented by uploading the ontology to a SESAME store, and via SPARQL requests. Consequently, the quality criteria are verified in the *triplestore*:

- *Each terminological class must have a prefLabel*
- *Each class must have one, and only one, prefLabel in the same language.*
- *Each prefLabel must be associated with a language.*
- *Each altLabel must be associated with a language.*
- *Each class must carry a hiddenLabel.*
- *Each class must carry only one hiddenLabel in the same language.*
- *Two classes must have the same prefLabel for the same language.*
- *Two classes must not have the same altLabel for the same language.*
- *Two classes must not have the same hiddenLabel in the same language.*
- *Two classes must not have one identical prefLabel and altLabel in the same language.*
- *Tracking of multiple parent classes.* The fact that one concept has two parent concepts is not a problem in OWL. However, this pluriparentality can be symptomatic of flawed modeling. In our methodology, the ontology is first designed based on a differential approach. The double heritage appears with the implementation of the defined concepts. Two parents can be allocated to one concept as an intermediary solution before the enhancement of the modeling. However, this double heritage, either intended or not, must be tracked and listed.
- *Additional requests.* Most of the additional requests have two primary optimization purposes – (i) to standardize the frag-URI in relation to lowercase or uppercase letters, and (ii) to standardize the labels and remove, to the possible extend, the characters (such as brackets or parentheses) that could hinder proper matching.

7 Conclusion and perspectives

Lerudi is a project that applies a specific methodology to the requirements of the medical emergency environment. The goal of this project is to build a TOR capable of retrieving information efficiently. Trough the complete description of the building process and the TOR validation in a large team, we have shown that: (i) concepts and terms must be precisely articulated within such a resource; (ii) the developed meta-modeling architecture must allow the modeling of all necessary KOS and other knowledge structures; (iii) standardized procedures based on this architecture may be implemented to enable the modeling.

Finally, the integration of the KOS in the same format and the RDF transformation service (capable of operating pre-treatments) allow to generate a termino-ontological resource with a lexicalization able to carry out the annotation, inference and indexation actions of the patients files. This project demonstrates the possibility to accommodate multiple KOS and to provide an efficient resource based on different request and transformation treatments.

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