e-Health – For Continuity of Care
C. Lovis et al. (Eds.)

doi:10.3233/978-1-61499-432-9-1023

Describing Localized Diseases in Medical Ontology: An FMA-based Algorithm

Jean CHARLET[a,b,1], Laurent MAZUEL[b,c], Gunnar DECLERCK[b,d], Patrick MIROUX[e], and Pierre GAYET[f]

[a] *Assistance Publique – Hôpitaux de Paris, Paris, France;*
[b] *INSERM, U1142, LIMICS, Paris, France, Sorbonne Universités, UPMC Univ Paris 06, UMR_S 1142, LIMICS, F-75006, Paris, France, Université Paris 13, Sorbonne Paris Cité, LIMICS, (UMR_S 1142), Bobigny, France;*
[c] *ANTIDOT, Lambesc, France;*
[d] *Costech, Université de Technologie de Compiègne, Compiègne, France;*
[e] *Dpt Urgences CHU d'Angers, Angers, France;*
[f] *Centre hospitalier de Compiègne, Compiègne, France.*

Abstract. ONTOLURGENCES is a termino-ontological resource developed to index and retrieve information in electronic Emergency Medical Record. In this project, we improved the ontology coverage to accommodate both anatomical and pathophysiological concepts in emergency medicine. This work lead to the automatic addition of 3,470 concepts and their underlying semantic formalization. In our method, we reuse and select the anatomical concepts relevant to emergency from FMA: To capture the anatomical specific concepts, (i) we involved Emergency practitioners and identified the key concepts from this domain; (ii) we applied an automatic algorithm to define the semantic relationships and integrated the result in the existing ontology.

Keywords. Ontology, Anatomy modeling, Physiopathology Modeling, FMA,

Introduction

In the LERUDI project, we have developed a terminological and Ontological Resource (TOR) relevant to emergency health care: ONTOLURGENCES. This model provides (i) all concepts applicable to this specific domain, (ii) the concept relationships, and (iii) the corresponding concept name in the Electronic Health Record (EHR). ONTOLURGENCES allows the annotation and indexing of the EHR document, and also the information retrieval (IR) in indexed files [1].

In this article, we aim at accommodating both pathophysiological and anatomical points of view in the TOR. These two points of view play a key role in the characterization of medical pathologies: by definition, a disease is a biological process anatomically localized by its cause or its observable events. The work of building a medical ontology must integrate and explicitly model them. However, significant challenges had to be tackled in order to achieve successfully the modeling. Practically, if the key concepts of anatomy (the human body and its division into finer elements)

[1] Corresponding Author.

and pathophysiology are accessible, their accurate modeling becomes quickly complex and error prone. In addition, ensuring logical as well as semantic consistency when modeling both points of view added to this complexity.

To overcome these difficulties, we chose to reuse a reference resource, the Foundational Model of Anatomy (FMA) to model systematically the anatomical concepts in ONTOLURGENCES. Then, we followed the following method: (i) we analyze as accurately as possible the pathophysiological approach/point of view in emergency medicine and develop accordingly the pathophysiological axis of the ontology, (ii) we construct the localized diseases axis reusing the FMA diseases.

In Section 1, we describe both resources involved in our project, ONTOLURGENCES and FMA; in Section 2, we develop the proposed methodology; in Section 3, we provide the first results of this project and discuss them briefly; and finally, in Section 4, we conclude and propose some perspectives.

1. Used Resources

The used resources are the ontology of emergency, ONTOLURGENCES, and the OWL version of the FMA.

OntolUrgences v3.03 was completed in July 20132, and is the test version of our/this work. It includes: 10,191 classes, 60 Object Properties, 1 Data property, and 11,591 logical axioms including 11,339 subclass axioms, and 89 equivalent class axioms. OntolUrgences is built with core-ontology OntoMénélas 4[3] [2].

FMA (Foundational Model of Anatomy) is a “reference ontology about human anatomy” [3][4]. Its goal is to modelize canonical human anatomy, i.e. “ the ideal or canonical anatomy to which each individual and its parts should conform” [4]. It contains more than 85,000 classes, 140 relationships connecting the classes, and more than 120,000 terms. We used the FMA v3.2.1 in OWL Full, and available on downloading site[4]. Its metrics are the following : 84,454 classes, 237,382 individuals, 132 Object Properties, 167 Data property, and 1,719,576 axioms including 87,803 subclass axioms. The large number of axioms and instances are related to the OWL full nature of this version of the FMA where each class is both son and individual of the superclass.

2. Method

Our Method is as follows: firstly, we organize all the concepts of diseases covered by the ontology in a subsumption tree representing the pathophysiological perspective as understood by emergency physicians – e.g. CardiacAneurysm is-a aneurysm. Then, the algorithm of the building branch of localized diseases occurs in two steps:

1. Building of the branch of localized diseases with defined concepts;
2. Building of the branch of anatomy necessary for the expression of axioms – e.g. the heart itself.

---

[2] http://purl.oclc.org/NET/spim/ontologies/public/OntolUrgences/
[3] http://purl.oclc.org/NET/spim/ontologies/public/OntoMenelas/
[4] http://sig.biostr.washington.edu/projects/fma/release/v3.2.1/alt_formats.html

In the following paragraphs of this section, we provide details on some notations and application conditions of our algorithm, and we develop the 2 steps.

2.1. Notation and vocabulary

- O is the work ontology, and O_{fma}, the OWL modelization of the FMA;
- We note the namespace of the ontology O onto: and that of the FMA fma:. So, onto:Cancer and fma:Heart are respectively valid URIs in the ontology O and in the ontology O_{fma};
- In the rest of the paper, the terms 'class' and 'concept' are interchangeable;
- We also use the set notation for axioms (statements in functional writing OWL2 [5]) when we describe ontology objects. For example : *Declaration(Class(fma:Heart))* $\in O_{fma}$

2.2. Initial hypotheses about the ontology

The main motivation is the automatic enrichment of our medical ontology based on the FMA anatomical location. Thus, the initial step is the annotation of each concept of the ontology by an FMA identifier: medical experts have added annotation onto:pourFMA to each disease of ontology O to locate.

2.2.1. Creation of diseases hierarchy

The general idea is to build a concept "DiseaseOf" for each location copied from the FMA. For example, if we copy fma:Heart, we will create a concept onto:DiseaseOfHeart. However, the anatomical hierarchy FMA does not translate directly into a hierarchy of diseases. You must use the whole-part hierarchy to be medically relevant. For example, if a patient experiences chest pain, he has a disease of an organ of the thorax (heart / lung). Following the anatomical subsumption, we could infer that lung disease is a disease of an organ. If, in absolute terms, the subsumption is not false, it has no medical relevance, therefore no interest. Conversely, with a whole-part relationship, we infer that the lung disease is a disease of lower respiratory tract or a chest disease and will be medically relevant. For this, we use whole-part relationships of the FMA. They are 4 [5]: (i) fma:part_of, (ii) fma:regional_part_of, (iii) fma:systemic_part_of, (iv) fma:constitutional_part_of. In the following, we use the generic name "part_of". The algorithm is as the following:

- Given a concept X, if there is a relationship "part_of" from this starting concept to a concept Y, then we create the concept DiseaseOfY as father of the concept DiseaseOfX. For example, if we have the axiom *ObjectPropertyAssertion(fma:part_of fma:Lung fma:Thorax)*, then we create axiom *SubClassOf(onto:DiseaseOfLung onto:DiseaseOfThorax)*;
- To ensure the final connectivity of the graph, if the concept has no relation "part_of" then we follow the classical subsumption relationship.

This new hierarchy is connected under the concept onto:Disease.

In addition, it is possible to transform these concepts in defined concepts such as restrictions are relations to the FMA. For example, onto:DiseaseOfLung is semantically

---

[5] http ://www.w3.org/TR/owl2-syntax/

defined as "a localized disease on one and only one instance of the lung." For this, we use the relationship onto:localizedOn defined between disease and location. Given a concept fma:Lung and a concept fma:Thorax such as *fma:Lung part_of fma:Thorax* then the concept onto:DiseaseOfLung is a concept defined by the following axiom:

EquivalentClass(onto:DiseaseOfLung
ObjectIntersectionOf(onto:DiseaseOfThorax
ObjectSomeValuesFrom(onto:localizedOn fma:Lung)
ObjectAllValuesFrom(onto:localizedOn fma:Lung)))

2.2.2. Copying the FMA

For each concept with an annotation onto:pourFMA, we copy in *O* the set of annotations axioms of the classes of FMA and all its relative superclasses to fma:Physical_anatomical_entity. Once this concept is reached, the hierarchy must be stored in the original ontology. In our case, it's stored under onto:StructureAnatomique, but the exact concept is of course dependent on the ontology in which the FMA is merged. Note that we retain the original FMA URI to keep all the benefits of Linked Data URI.

3. Results

The builded ontology has 12,396 Classes, 60 Object Properties, 1 Data property and 17072 axioms, among them, 13332 subclass axioms and 3559 equivalent classes axioms. These numbers are interesting to compare with those of ontology in its previous version, particularly in the axioms of equivalent classes, 89 versus 3559 that reflect the creation of new defined concepts (see Figure 1).

One of the advantages of our algorithm is that it is incremental: it is possible to apply the algorithm on an result ontology itself. In other words, it's not necessary to keep a "non-algorithm" version in order to apply it several times. Each concept is recovered and the axioms and restrictions are created if necessary. Moreover, the trees derived from the FMA are reduced to keep only the concepts used.

4. Discussion and conclusion

Evaluation of this work was done in-house and was first used to develop the presented algorithm. The model complexity of the FMA and understanding of what it could give has led many cycles of development. The first result is that we actually have at our disposal an algorithm that avoids us a long, tedious and error-prone task.

The next step is to work on improvements of the algorithm in order to propose an organization of the localized disease hierarchy "more medical": this goes by putting forward in the algorithm the systemic part_of which is close to the specialty description of medicine. We work on this topic in order to minimize manual final intervention on the ontology. The final goal is, if possible, to eliminate any manual intervention on the localized disease hierarchy.

The long term step of the evaluation could be to take the new version of OntolUrgences in LERUDI project and make a comparison of indexing some patients computerized records with the old version versus the new. But this would require that the French terms are all associated with ontology concepts, especially those of the FMA. This is currently not the case. We are considering for this to use translations made by CISMeF on EHTOP[6] portal and to propose them to the FMA consortium to enrich her ontology with French terms.

A long term perspective is the application of this algorithm on an ontology of another medical field.

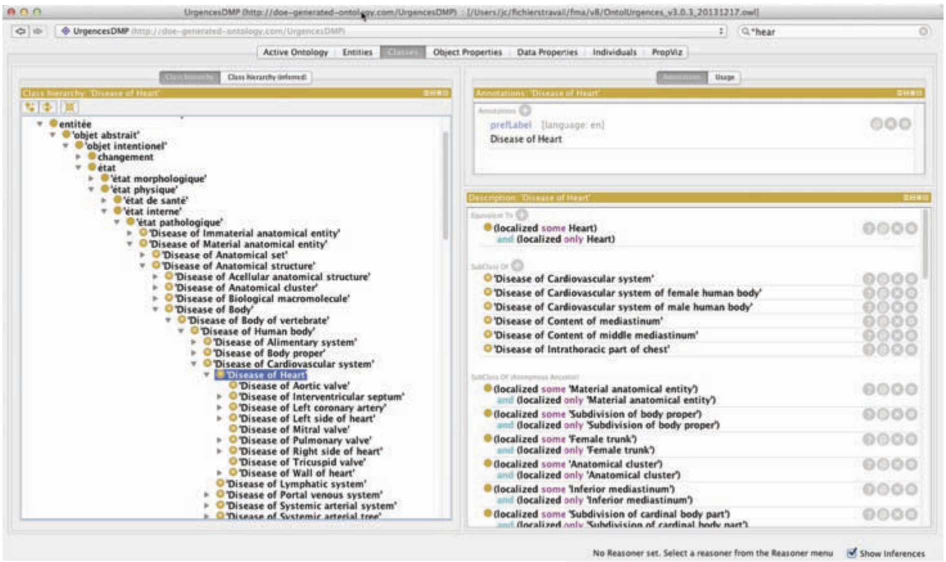

Figure 1. Screenshot of ontology in Protégé view at the "Disease of Heart" concept. Seen above the tree diseases back to the concepts of the core ontology OntoMénélas. At the window descriptions, we see the description of the concept and defined much of locations inherited from super classes. Given the number of possible inferences in the ontology, location reasoning is precalculated.

References

[1] Charlet J., Declerck G., Dhombres F., Gayet P., Miroux P. & Vandenbussche P.-Y. Building a Medical Ontology to support Information Retrieval: Terminological and metamodelization issues. In: Proc. of the10th International Conference on Terminology and Artificial Intelligence, Paris, France, 2013, p171-8.

[2] Bouaud J., Bachimont B., Charlet J. & Zweigenbaum P. (1995). Methodological principles for structuring an “ontology”. In Proceedings of the IJCAI’95 Workshop on “Basic Ontological Issues in Knowledge Sharing”, Montréal, Canada.

[3] Rosse C. & Mejino J. L. V. Jr. (2003). A reference ontology for biomedical informatics : the Foundational Model of Anatomy. Journal of biomedical informatics, 36(6), 478–500.

[4] Rosse C. & Mejino J. L. V. Jr. (2008). The Foundational Model of Anatomy Ontology. In A. Burger, D. Davidson & R. Baldock, Eds., Anatomy Ontologies for Bioinformatics : Principles and Practice, chapter 4, p. 59–118. New York : Springer.

[5] Mejino J. V., Agoncillo A. V., Rickard K. L. & Rosse C. (2003). Representing complexity in part-whole relationships within the Foundational Model of Anatomy. AMIA ... Annual Symposium proceedings / AMIA Symposium. AMIA Symposium, p. 450–4.

[6] http://www.ehtop.eu/