The Foundational Model of Anatomy in the Health Multi-Terminology Portal

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Abstract. The Health Multi-Terminology Portal is a repository dedicated to French health professionals and students. It provides access to twenty four health terminologies available in French. But until recently it was still missing a terminology about anatomy. The goal of the presented work was to enrich the portal with the Foundational Model of Anatomy, the reference ontology about human anatomy. The paper describes the method used to transform the Foundational Model of Anatomy ontology into a terminology semantically interoperable with the other terminologies of the portal. The result is a rich but lightweight terminology of anatomy, useful for a wide range of applications and users, whatever in education, resources indexing, hospitals or software publishers.

Keywords: Ontology, Terminology, Controlled Vocabulary, OWL, Portal, Life Sciences, Health, Anatomy.

Introduction

Far behind English, the second most used language for health terminologies is French. The Health Multi-Terminology Portal (HMTP)[1] is a repository developed by the Rouen University Hospital to supply health professionals and students with the terminologies available in French. HMTP includes twenty four terminologies and classifications that deal with various aspects of health, among which SNOMED for clinical term [4], MeSH (version 2010) used for indexing medical information in Pubmed, ICD10[2], MedDRA[3] for adverse effects, Orphanet for rare diseases[4], several terminologies developed by the World Health Organization: WHO-ART, WHO-ICPS, WHO-ATC, WHO-ICF etc. Most of them are bilingual (English & French). Additional terminologies/ontologies are currently being integrated, in particular SNOMED CT and LOINC (Logical Observation Identifiers Names and Codes). But until recently HMTP was still missing a terminology about anatomy.

The Foundational Model of Anatomy (FMA) is the "reference ontology about human anatomy" [4]. The FMA ontology is intended to model *canonical* human anatomy that is, "the ideal or prototypical anatomy to which each individual and its parts should conform" [4]. It contains more than 85,000 classes, 140 relationships

1 http://pts.chu-rouen.fr
2 http://www.who.int/classifications/icd/en/
3 http://www.meddramsso.com
4 www.orpha.net

connecting the classes and over 120,000 terms. The FMA describes anatomical entities, most of which are anatomical structures composed of many parts interconnected in complex ways. It specifies the anatomical structures by their relationships with other FMA entities, indicating their regions, constituents, innervations, blood vessels, boundaries etc. For example, a Heart is composed of two regions – its left and right side -, has several constitutional parts – *Wall of Heart, Interatrial, Interventricular*, and *Atrioventricular septum, Mitral Valve*, etc. -, is innerved by the *Deep cardiac plexus, Right and Left coronary nerve plexus*, etc. The FMA is a very large and perhaps one of the most complex ontology in the biomedical sciences. Consequently it may turn out to be quite difficult for users to use and to browse it.

The objective of the work is to add to HMTP a terminology of anatomy issued from the FMA. The aim is twofolds: i) to have a terminology of anatomy interoperable with the other French terminologies in HMTP ii) to offer a rich content about anatomy but nevertheless more simple than the FMA ontology and light enough so as to enable a fast and easy access to terms and resources of anatomy. The paper presents the method used to transform the Foundational Model of Anatomy ontology into a terminology.

1. Method

Semantic interoperability relies on the Unifying Model of Vocabulary (UMV2) of HMTP (§1.1). HMTP terminologies are implemented as OWL ontologies (§1.2). Going from the FMA ontology to a terminology of HMTP is based on *reification* (§1.3).

1.1. Unifying Model of Vocabulary

UMV2 is a common model for all terms, whatever their terminology. It can be viewed as a meta-model or an upper ontology designed to support broad semantic interoperability between terminologies that fulfill it. The basic concept of UMV2 is `Descriptor`, which is quite similar to skos:Concept. `Descriptor` has several attributes, e.g.; `label` refers to the preferred term used to name a descriptor in natural language, `synonym` refers to its synonyms in different languages, `notation` to its identifier. Another key concept is `Association`, used to model a relation between two descriptors as a class. It is quite similar to UML association classes. The concept `Group`, close to UML aggregation, is used to model a set of descriptors, for instance a terminology, e.g.; ICD10 or a group of terminologies, e.g.; SMQ, *Standardised MedDRA Queries*). In turn, the components and sub-components of ICD10, *Chapter, Category*, etc. are modeled as groups. For example, Chapter XI that list all the "Diseases of the digestive system", further divided into the categories from K00 to K93 (K00-K93), are all modeled as groups. UMV2 includes other general classes such as BT-NT for a hierarchical association, or RT-RT for "See Also" associations. UMV2 offers general properties that can be used to relate HMTP descriptors. The properties `BT` and `NT` of a `BT-NT` asssociation are used to assert that two descriptors have a hierarchical link. `BT` states the more general term ('broader') and `NT` the more specific ('narrower') of the association. The property `RT` states that two descriptors have *some* relationship. UMV2 also provides 'mapping' properties imported from SKOS used to align HMTP descriptors: `skos:closeMatch`, `skos:exactMatch`, `skos:broadMatch`, `skos:narrowMatch` and `skos:relatedMatch`.

Each terminology T of HMTP is built as an enrichment (UMV1-T) of UMV2. Each enrichment, e.g.; UMV1-FMA, UMV1-ICD10, defines its own specializations of Descriptor and of Association, e.g.; UMV1-FMA defines the descriptor FMAEntity and the associations FMAinnervation FMAdrainageveineux (venous drainage), while UMV1-ICD10 the ICD10categorie descriptor and the ICD10Exclusion association.

1.2. OWL representation

HMTP terminologies are implemented as OWL ontologies. The UMV2 concepts Descriptor, Association, Group and their UMV1 specializations are unary relations represented as OWL *classes*, e.g.; FMAdelimitation (Table 1 #1). The Descriptor attributes, label, synonym, etc., are represented as *Data Properties*. UMV2 general properties, such as BT, NT, RT, skos:exactMatch and the specialized properties of each UMV1-T ontology are binary relations represented as *Object Properties* (#9 #12). The terms of each terminology are represented as *individuals* of the relevant descriptor subclass. For example, the terms Heart, Lung, Surface of Heart, Right atrium etc. of the FMA terminology are individuals of the class FMAEntite, e.g.; the individual FMA_7088 (#13) for Heart (#15). A relation between two terms is represented by an OWL individual of the concerned Association joined with two property assertions. For example, the individual FMA_ bounded_by~FMA_7088-FMA_7167 of the FMAdelimitation subclass of Association (#16) asserts that the term Heart and Surface of heart are related via a relation with source Heart (#17) and target Surface of Heart(#18). The individual FMA_BTNT~55209 of the BT-NT class (#19) asserts that the terms Heart and Organ_With_Cavitated_Organ_Parts have a hierarchical relation, the broader term (#20) being Organ_With_Cavitated_Organ_Parts (FMA_55673) and the narrower (#21) being Heart (FMA_7088).

1.3. From FMA ontology to a terminology

Going from the FMA-OWL ontology to a terminology (FMA-TERM) compliant with the UMV2 metamodel relies on a *reification* process transforming FMA-OWL *classes* into FMA-TERM individuals representing terms, *Object properties* into Association subclasses. More precisely, each FMA-OWL *class* is mapped to an individual of the FMAEntity subclass, which IRI is generated from its FMAID in FMA-OWL, e.g; the individual FMA_7088 forHeart, (#13 Table 1)with the IRI http://www.chu-rouen.fr/smts#FMA_7088 created from its FMAID (7088). SubClassOf(*fma:A fma:B*) axioms are transformed into BT-NT individual assertions, SubClassOf*(fma:A* ObjectSomeValuesFrom*(fma:R fma:B))* axiom into an individual and two object property assertions, FMA-OWL *preferred-name* annotations into labels, other *annotations* into *datatypes*. *Object properties R*, e.g; bounded_by, venous_drainage, innervation etc., are transformed into Association subclasses (named FMA*R*) and two object properties with SubClassOf*(A* ObjectAllValuesFrom*(R B))* and cardinality restrictions

axioms specified in UMV1 FMA. For example, bounded_by is mapped to FMAdelimitation (#1) subclass of Association (#4) and two object properties delimite and estDelimitéPar (#9-#12) are created with value restrictions (#5-6-7-8).
The SubClassOf(fma:Organ_With_Cavitated_Organ_Parts fma:Heart) axiom is transformed into the individual FMA_BTNT~55209 (#19) and property assertions (#20-21), while SubClassOf(*fma:A* ObjectSomeValuesFrom(*fma:R fma:B*)) into a class assertion (#16) and two object property assertions (#17-18). The transformation of FMA-OWL into FMA-TERM is achieved by a Java parser with the SAX API; the resulting file is created thanks to Jena API.

Table 1 FMA-TERM terminology in OWL 2 functional syntax: Heart example

```
----------------- Classes ----------------
1. Declaration(Class(:FMAdelimitation))
2. AnnotationAssertion(rdfs:label :FMAdelimitation "Délimitation"@fr)
3. AnnotationAssertion(rdfs:label :FMAdelimitation "Bounded by /
   bounds"@en)
4. SubClassOf(:FMAdelimitation publishing:Association>
5. SubClassOf(:FMAdelimitation ObjectAllValuesFrom(:FMAdelimite
   :FMAentity))
6. SubClassOf(:FMAdelimitation ObjectAllValuesFrom(:FMAestDelimitePar
   :FMAentity))
7. SubClassOf(:FMAdelimitation ObjectMinCardinality(1 :FMAdelimite))
8. SubClassOf(:FMAdelimitation ObjectMinCardinality(1
   :FMAestDelimitePar))
----------------- Object Properties ----------------
9. Declaration(ObjectProperty(:FMAdelimite))
10. ObjectPropertyDomain(:FMAdelimite :FMAentity)
11. ObjectPropertyRange(:FMAdelimite :FMAentity)
12. Declaration(ObjectProperty(:FMAestDelimitePar)), etc.
----------------- Individuals ----------------
13. ClassAssertion(:FMAEntity :FMA_7088))
14. AnnotationAssertion(rdfs:label :FMA_7088 "Coeur"@fr)
15. AnnotationAssertion(rdfs:label :FMA_7088 "Heart"@en)
16. ClassAssertion(:FMAdelimitation :FMA_bounded_by~FMA_7088-FMA_7167)
17. ObjectPropertyAssertion(:FMAdelimite :FMA_bounded_by~FMA_7088-
    FMA_7167 :FMA_7088)
18. ObjectPropertyAssertion(:FMAestDelimitePar :FMA_bounded_by~FMA_7088-
    FMA_7167 :FMA_7167)
19. ClassAssertion(publishing:BT-NT :FMA_BTNT~55209)
20. ObjectPropertyAssertion(publishing:NT :FMA_7088)
21. ObjectPropertyAssertion(publishing:BT :FMA_55673)
```

2. Results

FMA-TERM terminology is a lightweight $\mathcal{ALN}$(D) ontology with 39 classes, 41 object properties, 19 data properties, 136 subclass axioms and 218419 individuals. It includes 81042 FMA descriptors with 81020 unique English terms corresponding to the FMA entities, 52040 unique English synonyms, 4436 unique French terms and 139 French synonyms. Twenty two FMA relationships were selected to be integrated. 52447 relations connect 39787 distinct classes and 81224 hierarchical BT-NT relations mirror the FMA ontology original structure. To consult the FMA terminology of HMTP (http://cispro.chu-rouen.fr/pts_site/index.html?lang=en, click on "Connection"; login=fmauser, password=fmapass).

The FMA terminology benefits of all the HMTP services, in particular of time performance of search and display. HMTP advanced functionalities and its terminology interoperability allow finding an anatomical entity not only within the FMA but also the other terminologies, searching for terms among the preferred terms, synonyms, definitions or codes (FMAID) in all terminologies, with three search modes (truncated, stemming or exact search). It is possible to make a search in French or English (if the terminology data include the translation). A special display enables to handle poly-hierarchies. The FMA terminology also gains in content and bilingual use. 6195 French translations were manually added to the already existing 4436 French terms (i.e. +140%) and also Latin terms. The FMA terminology is a central resource for a daily use in resources indexing. As MeSH lacks precise terms of anatomy, the FMA in HMTP is a great opportunity for librarians to improve the level of indexing, hence to allow users querying more precise terms and finding relevant resources.. It is also crucial for education. Its rich content combined with a friendly and fast interface is a useful and powerful tool for students (in medicine, physiotherapy, nursing, sports etc.). For example, when they type "supinator", they obtain 37 FMA entities, and all information about "supinator". The Relations tab provides four relationships, including "constitutional part of" pointing to the entity "Posterior compartment of forearm". If they click on it, they obtain the 10 muscles located in this particular compartment.

3. Discussion

The FMA terminology of HMTP allows users to interact more easily and faster than with the FMA ontology in Protégé or OWL. First, in general ontologies are less adapted than terminologies to daily index resources and to education. Second, compared to interactions with Bioportal, the FMA terminology benefits of some advantages of HMTP. For example, field names used in Bioportal are not always very explicit (e.g. for asthma in MeSH, Bioportal proposes a "Ro" category which means "See also"). Moreover, Bioportal does not fully respect the original models of ontologies or terminologies (e.g; SNOMED CT or FMA hierarchies). Bioportal deploys only one path (e.g. asthma in MeSH has three broader terms - with four different tree numbers - but Bioportal displays only one path by default), whereas HMTP enables to display polyhierarchies. On the other side, Bioportal provides interesting features missing in HMTP such as semantic types, data sources or user notes. Bioportal implements a friendly visualization tool to display concepts and network neighborhood. While HMTP provides access to resources like PubMed, CISMeF, Bioportal offer interesting links to other portals (Adverse Event Reporting System Data, ClinicalTrials.gov, NextBio etc.). Finally, Bioportal is a collaborative tool whereas HMTP is not.

The method used to go from the FMA ontology in OWL to the FMA terminology in HMTP is general and might be applied to move other ontologies to terminologies while reconciling the two [4]. The idea behind is that ontologies and terminologies have different uses. While ontologies provide knowledge useful to support ontology reasoning, design and maintenance, health terminologies are more simple thus more suited to resources or text indexing and retrieval, and for coding systems. The idea to go from an ontology in BioPortal to a "lexicon" might be a little similar [4], but a lexicon is even more simple than a terminology.

Conclusion

We have presented a method to integrate the FMA OWL ontology into HMTP. The result is a rich but lightweight terminology of anatomy, interoperable with other French terminologies of HMTP, well adapted to a wide range of applications and users, in particular to resources indexing & retrieval and to education. A future perspective is to improve the UMV2 model to be more consistent with standard languages, e.g. SKOS, OWL 2, and to develop a general tool based on a similar method to move Life Sciences ontologies to terminologies, while keeping a close link between them.

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