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Teaching Medicine with a Terminology/Ontology Portal

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Abstract. The Health Terminology/Ontology Portal (HeTOP) was developed to provide easy access to health terminologies and ontologie. The repository is not only dedicated to professionals but is also a valuable teaching tool. Currently, it provides access to thirty two health terminologies and ontologies available mainly in French or in English, but also in German, Italian, Chinese, etc. HeTOP can be used by both humans and computers via Web services. To integrate new resources into HeTOP, three steps are necessary: (1) designing a meta-model into which each terminology (or ontology) can be integrated, (2) developing a process to include terminologies into HeTOP, (3) building and integrating existing and new inter & intra-terminology semantic harmonization into HeTOP. Currently, 600 unique machines use the MeSH version of HeTOP every day and restricted terminologies/ontologies are used for teaching purposes in several medical schools in France. The multilingual version of HeTOP is available (URL: http://hetop.eu/) and provides free access to ICD10 and FMA in ten languages. Conclusion: HeTOP is a rich tool, useful for a wide range of applications and users, especially in education and resource indexing but also in information retrieval or performing audits in terminology management.

Keywords. Controlled Vocabulary; Internet; Database; Information Storage and Retrieval; Internet; Terminology as subject.

Introduction

The Internet is currently a major source of both scientific and health information as well as knowledge. If English is the language for more general subjects, other languages are available in the health information domain for physicians, medical students and lay people alike. Nonetheless, people are now more internationally mobile, health information should also cross borders becoming multilingual and based on several health terminologies and ontologies. Some institutions have already shown that health information is available on line in several languages: e.g. MEDLINEplus, thus providing the general public with content in English and Spanish for example. Moreover, the European Medicine Agency provides drug information for both health professionals and non-professionals in each official European language.

Health and Law are the main fields of knowledge which several terminologies and ontologies (T/O) co-exist. Over 150 terminologies and classifications in English are

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included in the Unified Medical Language System (UMLS) meta-thesaurus developed by the US National Library of Medicine in 1986.

T/O are not only increasingly more complex with rich semantic relations, but also more diversified and comprehensive. Rich semantic relations are defined as relations that provide the end-user with an added-value besides the classic broader-narrower (BT-NT) relation (e.g. the relation "is located" in the Foundational Model of Anatomy (FMA) or "genes linked" in the Orphanet thesaurus for rare diseases [1]). These knowledge resources are mainly used to index (or annotate) or perform complex tasks such as ontology reasoning, etc. To perform such tasks, terminology portals have already proven to be very useful, in particular BioPortal [2]. Since 2006, the CISMeF team has been developing a terminology portal which originally focused on French T/O [3] (URL: http://pts.chu-rouen.fr) but which is currently available in a cross-lingual version (URL: www.hetop.eu).

The objective of this paper is to demonstrate the interest of a terminology portal in teaching anatomy and rare diseases to medical students. These two fields of medicine were chosen because one ontology already exists in anatomy (FMA) and two for rare diseases (Orphanet & Human Phenotype Ontology (HPO) [4]); these three T/O have rich semantic relations.

1. Methods

Dealing with these kinds of T/O is no easy task due to structure, size, nature and specificity. First, we had to create a meta-model in order to integrate all T/O and one global generic system. Then, we had to study each T/O to integrate it without degrading it. Finally, we had to develop a web application dedicated to both health professionals and students, with an adapted graphic interface and an efficient search engine. A metamodel was designed according to several previous research projects (see Acknowledgments). This metamodel was validated as we managed to integrate not only any terminology into it [3] but also ontologies [5] such as FMA and HPO. Any new T/O to be integrated is a challenge because of its structure and functions: an expertise is necessary to understand the model and format of the data. A parser is then developed to allow integration into the metamodel and relational database. To ensure interoperability between T/O, Natural Language Processing tools have been developed and validated [6]. These tools allow the creation of several kinds of mapping between all concepts of the different T/O. It is therefore possible to navigate between T/O.

The HeTOP was designed as a web site, based on the relational database described above. This tool was evaluated by two consecutive groups of second year medical students in September 2010 and September 2011 respectively. A simple questionnaire was completed at the end of the lecture on the terminology portal, which lasted around one hour: this questionnaire used a semi qualitative scale from 0 to 100. Each medical student was asked two questions: one on the ergonomics of the portal and the other on its teaching interest.

2. Results

A total of 32 terminologies were included in HeTOP, with 980,000 concepts, 2,300,000 synonyms, 222,800 definitions and 4,000,000 relations. In the current cross-lingual

version, it is possible to navigate between 23 languages (mostly European; some not using the Latin alphabet e.g. Greek, Russian or Bulgarian; some non-European languages were also introduced into HeTOP e.g. Arabic, Hebrew, Japanese or Mandarin).

HeTOP has been a tool for teaching rare diseases to Rouen medical students since 2010. In the Orphanet ontology, for example, the Marfan syndrome is linked to two genes: FBN1 and TGFBR2. Students may want to know which other rare diseases are linked to this gene. In two clicks, the student will obtain the answer (n=6). Similarly, the Marfan syndrome is linked to 65 clinical signs (e.g. coloboma of iris). Students may want to know which other rare diseases are linked to these clinical signs. In two clicks, the student will have the answer (n=71). In fact, thanks to the multi-T/O of the portal, students will also have access to the 51 clinical signs of the HPO ontology. Such a wealth of information is most valuable to the ontologist but may be counter productive to students providing potential conflict of knowledge: some clinical signs are present in Orphanet and not in HPO and vice versa.

In addition, since 2010, HeTOP has also been a teaching tool on the anatomy course for the same Rouen medical students using the FMA ontology. For example, those students requiring information on the “supinator”. They have access to several tabs in the HeTOP portal: (a) tab "Description" provides information on the concept “supinator”, including various synonyms, translations (e.g. “Muscle supinateur” in French), UMLS Concept Unique Identifier (CUI); (b) tab "Hierarchies" provides the hierarchies for each concept for each T/O. During the teaching course, medical students are told how important it is to acquire medical knowledge from T/O hierarchies (e.g. learning various siblings of a rare disease and one or several ascendances of this rare disease); (c) tab "Relations" is the most important part of this work, providing rich semantic relations. In the example of the “supinator”, it provides the following relations: Nerve supply(ies), Segmental supply, Constitutional part(s), Member of. For the latest relation, “Supinator” is Member of : (a)"Musculature of forearm", and (b) "Set of deep muscles of posterior compartment of forearm". Students may learn these two relations, want to go a step further and know all the muscles of the forearm. The answer (n=18), see Figure 1 is two clicks later.

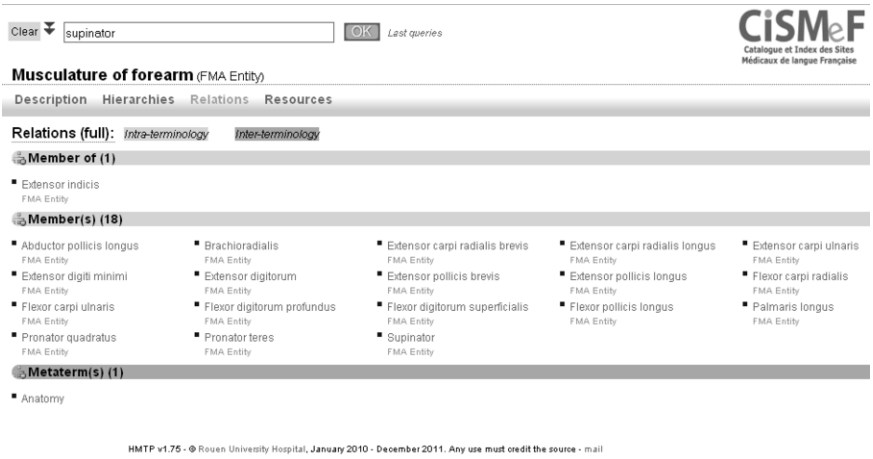

Figure 1. Screenshot of HeTOP: FMA example on musculature of forearm

The results of the two qualitative evaluation surveys performed over the previous two years on two successive cohorts of Rouen Medical School students (second year) are as follows. In September 2010, 25 medical students filled out an evaluation form (each student attending the teaching course) and in September 2011, 11 medical students completed the evaluation. The results of the questionnaire are displayed in Table 1. The mean results for the first question "how do you grade portal interest in teaching" increased from 79.9% to 87.5% between 2010 and 2011. The mean results for the second question "how do you grade the portal ergonomics" remained relatively stable at 57.2% and to 55.5% respectively.

Table 1. Results of the 2010 and 2011 evaluations of HeTOP.

	Mean (%) ± Std deviation 2010	Mean (%) ± Std deviation 2011	Mean (%) Overall
Interest in teaching	79.9 ± 12.9	87.5 ± 10.1	83.7
Design	57.2 ± 16.5	55.5 ± 19.2	56.35

3. Discussion

To the best of our knowledge, this paper describes the first experiment in teaching certain aspects of medicine (rare diseases and anatomy) using a terminology portal. This was achievable thanks to the rich semantic relations existing in HPO and Orphanet as regards rare diseases and FMA for anatomy's. This survey was conducted over the previous two years at Rouen Medical School. It has now become part of the curriculum of medical informatics for 2nd and 3rd year medical students at Rouen Medical School. It was extended to medical informatics master's degree students in Paris in September 2011. As the CISMeF catalogue (URL: www.cismef.org) is taught in half of French Medical Schools, we were then able to extrapolate the same results for HeTOP over the next ten years.

This experiment could be applied to our European counterparts as the cross-lingual HeTOP provides Orphanet in five languages (English, French, Italian, Spanish and Portuguese). The CISMeF team has already translated 68% of HPO into French. FMA is however only fully available in English. It has been partially translated into several languages (e.g. Latin, French, German, Italian) by the University of Washington (USA). The CISMeF team continued this translation task in French, currently providing a new translation for 10,000 out of 70,000 terms [7].

Furthermore, this experiment could be extended to other fields of medicine with pre-existing T/O with rich semantic relations: in particular, SNOMED CT and NCIT (US National Cancer Institute Thesaurus) for cancer.

The HeTOP presented here has the main functionalities of any terminology server, excepting the extensive management of terminologies (e.g. adding a new hierarchy). To the best of our knowledge, the HeTOP is the first of its kind in French. The main added value of HeTOP when compared to any UMLS browser [8] is the possibility it offers to access the main health terminologies in French or multi-lingual terminologies and classification from WHO, not yet included in the UMLS (e.g. WHO-ATC for drugs or WHO-ICPS for patient safety).

Other portals propose searching and navigating T/O such as NCBO Bioportal and the EBI Ontology Lookup Service [9]. These tools are also very friendly but do not

allow users to navigate through terms or cross-search synonyms in different languages. Nonetheless, the bilingual and cross-lingual version of HeTOP allow a contextual link to BioPortal for the 12 T/O included in UMLS. The goal is to enhance our results thus providing the best possible service to users: this link was tested with success by a French physician requiring an extended view of LOINC using both HeTOP & BioPortal to create an interface terminology in biology.

Whereas assessment of HeTOP has demonstrated that its content was most appreciated by students, these studies show a need for improvement in its design. Indeed, this kind of portal is complex and necessitates further research on new user access. In addition, a wider study on the portal quality and its use would be of value.

From its beginnings as an educational aid, to the present, HeTOP is now mainly dedicated to medical librarians to index resources in a multi-terminology mode. HeTOP is also very useful not only for translators, terminologists and ontologists, but also healthcare professionals, in particular physicians using HeTOP for coding and using services on demand (e.g. info buttons). This portal is also a valuable tool for performing audits on terminologies. Finally, HeTOP is a good way to show research work results (mappings, automatic translations, new T/O, etc.).

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