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TOWARDS INTEGRATION OF ONTOLOGIES IN HEALTHCARE

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ABSTRACT: *Digital health is facing many challenges. Nowadays the use of ontologies in health care has increased and is covering wide range domains in healthcare. Using ontologies may improve the semantic interoperability and also offer the possibility to gain knowledge from them. It is significantly important not only to implement ontologies in healthcare but to integrate them in order to benefit from different ontologies. In this paper, we provide a comprehensive overview of the importance, advantages and challenges in integrating ontologies through a semantic mapping scenario between two ontologies. Integration of ontologies may support the decision-making process of healthcare providers by deriving relationships between different sets of conditions, findings, signs or symptoms.*

KEYWORDS: ontology, integration, mapping, healthcare.

INTRODUCTION

Ontology is a representational artifact, comprising a taxonomy as proper part, whose representations are intended to designate some combination of universals, defined classes, and certain relations between them [1]. Using the technologies of web semantic may provide access to additional information and facilitate the research [2]. Ontologies play a significant role in knowledge administration and integration of knowledge in healthcare [3]. From a healthcare perspective, the authors in [4] have founded that the ontologies can be used to maximise:

- meaning that can be inferred from coded data;
- different granularities of data;
- the ability to cope with temporal change in definitions, clinical practice and fluctuation;
- structural (system studies, e.g. encounters, health professionals, governance and privacy) [4].

Nowadays, there are different ontologies in the same domain that use different classes to express the same concept or class. In order to benefit from both of the ontologies it is necessary the integration of the ontologies. Ontology integration is the inclusion in one ontology of another ontology and assertions expressing the glue between these ontologies, usually as bridge axioms. The integrated ontology is assumed to reflect the knowledge of both initial ontologies [5].

So, it is very important to reuse and integrate them in order to offer additional information in this domain. For example, integrating Radiology Gamuts Ontology (RGO) with other ontologies provides opportunity in integrating radiologic knowledge with other domains [7]. RGO ontology is an ontology that defines causal relations like may_cause and the inverse relationship may_be_caused_by to express the links between diagnoses and imaging observations [6]. According to the authors in [7], the RGO ontology has more than 2000 imaging findings in the body system, more than 12000 conditions that cause findings, etc. Also, from the metrics of the RGO Ontology using Protégé can be founded that there are more than: 18000 classes, 16000 declaration axioms, 6000 logical axioms and 181000 axioms.

The aim of this paper is to provide a comprehensive overview of the importance and challenges in the mapping process in order to integrate ontologies. This research paper is structured as follows: The next sections will cover (1) the related works in the process of ontologies integration and the challenges that should be handled by the integrated ontology, (2) the case study in mapping two ontologies and finally (3) the conclusions and future work.

RELATED WORK

Understanding the relationships among multiple classes from different ontologies will be more useful for evidence-based medicine or personalized treatment than general ontologies or bigger ontologies [8]. While using ontologies improves the semantic interoperability, the integrating of two or more ontologies can offer knowledge integration between the ontologies.

Currently the RGO ontology is integrated with the Orphanet Rare Disease Ontology (ORDO) in order to support translational rare-disease research by linking knowledge of genetics and imaging phenotypes [9]. Also the authors in [10] integrate ontologies of human diseases, phenotypes with radiological diagnoses in order to highlight the possibility of categorizing the RGO concepts within the hierarchies of disease and phenotypic abnormalities. The RGO ontology is integrated also with ICD-10-CM, RadLex, and SNOMED CT [6].

The authors in [11] designed onGrid [12] and LDPMap [13] in order to generate a similarity matrix between two sets of heterogeneous biomedical ontologies, and then to identify the optimal solution for integrating the two ontologies.
According to research work in [14], ontology integration should handle the following problems:

- Identify alignment between related entities which are semantically correlated;
- Find the places where ontologies overlap and integrate ontologies;
- Prune integrated ontology through detecting ontology redundancy;
- Check the consistency of the integrated ontology [14].

Additionally, the authors in [15] present analysis and classification of ontology mapping evaluation approaches, methods and tools and according to them it is very important to offer easy to use GUI for mapping or evaluation systems.

CASE STUDY IN MAPPING ONTOLOGIES

Although ontologies enable knowledge source interoperability, different applications may use different ontologies to represent the same domain, so this issue can be solved through the establishment of semantic mapping between multiple ontologies [22]. In this paper we experimented in mapping concepts/classes of the Clinical Signs and Symptoms Ontology (CSSO) ontology with Radiology Gamuts Ontology (RGO). These ontologies were accessed using BioPortal website [16]. CCSO is an ontology that describe clinical signs and symptoms. It has 330 classes that represents development symptoms, nervous system symptoms, skin symptoms, head and neck symptoms, etc. [16]. The metrics of the CSSO using Protégé are highlighted in the figure 1.

Figure 1. Metrics of the CSSO Ontology

If we use the Merge Function in Protégé to Merge Clinical Signs and Symptoms Ontology with Radiology Gamuts Ontology in a new ontology, we can observe the following metrics.

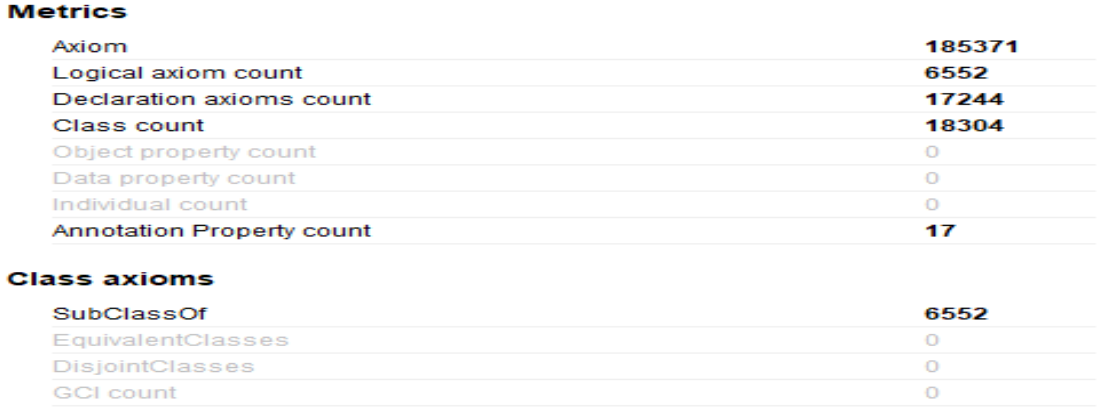

Figure 2. Metrics of the CSSO and GAMUTS Ontology

From the Metrics it can be found that the Merge Function in Protégé concatenates the two ontologies and did not merge them. This happened because of the different IRI

(Internationalized Resource Identifier) used in the two ontologies. Based on the [10], [17], [18], [19] and [20], by using the NCBO Annotator some of the terms of the CCSO ontology will be mapped directly to the RGO terms like *Hyperhidrosis*, *Thrombocytopenia*, *Neoplasm*, *Tremor*, *Asthenia*, *Hyperplasia*, *Dysphagia* etc. The data that have been annotated can be formatted in JSON or XML for transmitting or storing data. Afterwards, the data can be converted to RDF from XML by using XSLT. Also, it is very important to match the terms/classes in healthcare using synonyms and abbreviated forms of the terms/classes because the same term/class is used in different forms even they represent the same concept. By using the synonyms of the terms in the CSSO it is possible to find the exact mappings in the RGO. These terms can be linked with each other using sameAs relationship. For example, *"Bleeding"* in CCSO has synonym "*Hemorrhage*" so it can be mapped to "*Hemorrhage*" in RGO using the relationship same As.
From the integration of the two ontologies it is possible to gain knowledge from subtypes findings. For example: "*Arthropathy"* (CCSO) is mapped with "*Arthropathy"* (RGO) and according to the RGO, it has subtypes *"Arthritis*" and *"Cheiroarthropathy"*. The terms can also be mapped using other relationships like is_A relationship for example, "*Polyp*" (CCSO) is_A "*Beging neoplasm*" (RGO).

The process of integrating ontologies with RGO will provide additional knowledge to the overall integrated ontology. For instance, by mapping the CCSO ontology with the RGO ontology, we can get knowledge about the relationships may_cause and may_be_caused_by. So "*Hemorrhage"* may be caused by "*Cerebellar lesion*", "*Choroidal* mass" etc. and may_ be_caused by "*Splenic infarction"* or "*Trauma"*.
In the process of integrating and mapping the terms between ontologies it is important to highlight that the doctor's expertise plays a crucial role in the process of mapping the terms/concepts and defining their relationships. Examples of such terms could be the relation of the "*Mouth Symptom*" term in CSSO to "*Cancer of floor of mouth*", "*Carcinoma of mouth*", etc. terms in RGO. In addition, beside of automated or semi-automated tools and methods, the healthcare experts should evaluate and check the consistency of the integrated ontology.

Meanwhile it is very important to map the terms with SNOMED CT or ICD 11 in order to enable semantic interoperability in health care. According to the [21] SNOMED should be used for: specific concepts and value sets, including diseases, symptoms, signs, etc., representation of constraints on use of terminology, in simple semantic relationships, in constraints on combination of concepts and in post-coordinated expressions at various levels of nesting.

CONCLUSIONS AND FUTURE WORK

In this paper, we provide a comprehensive overview of the importance, advantages and challenges in integrating ontologies through a semantic mapping scenario between RGO Ontology and CCSO Ontology. Integrating one ontology with one or more ontologies will add value and additional knowledge to the existing ontology. By using the NCBO Annotator it can be found that more than 20% of the terms of CCSO ontology will be mapped directly to the RGO terms, while other terms can be mapping

through semantic meaning, synonyms, doctors experience etc. Mapping CSSO terms with the NGO terms will offer the possibility to give the radiologic knowledge according to Clinical Signs and Symptoms Ontology. According to the signs or symptoms of the patients it is possible to know the relations may_cause and its inverse may_caused_by through the mapping process between the ontologies. As in [10] even though some terms have been matched in an automated manner, it is very important to review the concepts by an expert in health care in order to be sure that the mapping are semantically correct and to find other mappings in the ontologies that will be integrated. Future work will focus on semantic interoperability, methods and tools in mapping, evaluating etc. and on the overall process of integration of the ontologies.

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