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## Ontology Maintenance System for Rheumatoid Disease

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### Abstract

The constructed medical ontologies need to be updated in order to reflect the changes occurred on the medicine such as the clinical findings, treatments and their side effects. Various researchers defined the ontology maintenance as the process of updating the ontology or the evolution of the ontology. Other researches consider the ontology maintenance as a composed process that involves the ontology evolution and pruning. This paper presents a Rheumatoid ontology maintenance system that incorporates both of the ontology evolution and the ontology pruning. The evolution is executed in a way that ensures the ontology consistency and its relevance to the domain of interest.

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### 1. Introduction

The large medical datasets cannot be easily interpreted; they need an intelligent technique to obtain the inferred knowledge. Ontology is used in the medical domain for the semantic knowledge representation that enables the intelligent agents and intelligent computer systems to understand the data and discover new knowledge. Ontologies can be used in Decision Support Systems (DSS) 1,2,3,4, medical data integration 5,6,7 and knowledge management 8, 9 . Shaban-Nejad and Haarslev 10 have defined the notion of ontology maintenance as the changes that are occurred on the ontology. Luczak-Rösch11 assumes that the ontology maintenance concerns about the evolution of ontologies that are used in an application. DoingHarris et al 12 consider the ontology maintenance as the process of

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expansion of current ontology. They assume that the ontology maintenance is the variant of ontology evolution notion. Maedche and Volz 13 have defined the ontology maintenance as a composed process that involves two sub processes; the ontology evolution and the ontology pruning.

For the ontology evolution, Haase and Stojanovic 14 have defined it as the adaptation and the update of the ontology in a consistent manner. Plessers et al 15 have defined the ontology evolution as the adaptation of the ontology according to the changes that are appeared in the domain of interest taking into account the ontology consistency. The changes are classified into basic and complex changes. The basic changes involve the addition and deletion of ontology elements. The complex changes involve merging, splitting and moving the ontology classes. Zablith et al 16 proposed five tasks for the ontology evolution which are; detecting the need for changes, suggesting changes, validating changes, assessing impact and managing changes. Several ontology evolution systems were proposed in the biomedical and medical domains. Messaoud et al 17 presented a SemCado system that relies on the use of Causal Bayesian Network (CBN) to evolve the ontology. Dos Reis et al 18 proposed DyKOSMap, which focuses on the evolution of mapping between two new Knowledge Organization Systems (KOS) based on the evolution occurred on existing KOS. The KOS involve ontologies, semantic networks and Thesaurus.

Sari et al 19 presented a system to evolve a sub ontology. It relies on the comparison between the current sub ontology and the change log file of the whole evolved ontology. Doing-Harris et al 12 presented the Semi-Automated ontology Maintenance (SEAM). The evolution of ontology relies on the recommendation of extracted terms from biomedical and clinical texts. For the ontology pruning, Kim et al 20 have defined the ontology pruning as the deletion of irrelevant concepts from the ontology. Swartout et al 21 presented a pruning approach that relies on the experts who determine the relevant terms to the SENSUS ontology. The pruning process is implemented in a manual way. Maedche and Volz 13 proposed a pruning system that relies on the use of generic and specific documents to identify the relevant concepts. The system enables the user to accept or reject the proposed concept. K.D. Pandya and C. Pandya 22 proposed a pruning system that focuses on the deletion of object properties based on a weight, this weight is determined according to the characteristics of the property such as domain, range, complement, inverse, functional...etc.

The objective of this article is the presentation of a new ontology maintenance system that is composed of two subsystems; the ontology evolution and the ontology pruning. This paper is organized as follows; section 2 presents the proposed ontology maintenance system architecture, section 3 provides the evaluation of the proposed system and section 4 contains the conclusion and future work.

## 2. The Proposed Ontology Maintenance System

The proposed system is based on the notion of the ontology maintenance that is defined by Maedche and Volz 13. As figure 1 demonstrates, the system consists of two subsystems; the ontology evolution subsystem and the ontology pruning subsystem. Both of the ontology evolution subsystem and the ontology pruning subsystem rely on the use of accredited medical sources such as the Unified Medical Language System (UMLS) 23, RxTerms 24, a treatment database and a medical corpus.

### 2.1. The ontology evolution subsystem

The ontology evolution subsystem enables the user (health care personnel, patient, physician...etc.) to implement both of basic and complex changes on the medical ontology. It takes the ontology, medical terms, basic and complex changes inserted by the user as inputs. The evolution subsystem ensures that these entries can't cause inconsistency in the ontology. The output of this subsystem is an evolved ontology. If the ontology evolution subsystem detects the existence of the term or its synonyms in the treatment database, it will recheck again the ontology to make sure that the medical term synonym(s) didn't exist in the ontology. If the user inserts a medical term that presents a commercial treatment name that is used only in Egypt, the ontology evolution subsystem will check the treatment database to get the scientific name of this treatment and it will search the ontology to make sure the inexistence of the scientific treatment name. If the ontology didn't contain the treatment, the ontology evolution subsystem will recommend the scientific name to the user who will make a decision either to add it in the ontology as an individual attached to a specific class/subclass or creating a new class and attach the scientific treatment name as individual. In

case that the ontology evolution subsystem didn't find the treatment name in the treatment database, it will check its existence in the UMLS and the RxTerms to get the scientific name of the treatment. If the medical term is a clinical finding, the ontology evolution subsystem will search the UMLS to get its normalized term taking into consideration the manipulation of the composed terms. Moreover, the evolution subsystem enables the user to execute other basic changes such as creating/deleting a class, an individual, an object property, a data property and an object property assertion.

For the complex changes, the ontology evolution subsystem enables the user to execute the merge, move and split operations on the ontology. Concerning the merge operation, the ontology evolution subsystem asks the user to insert the names of two classes to be merged, then the subsystem will check their existence in the ontology and asks the user for which class the two classes will be merged, if this class does not exist in the ontology, the subsystem will create it. Concerning the move operation, the subsystem enables the user to change the ancestor of a specific class (change the class hierarchy). For the split operation, the subsystem enables the user to split a specific class into two or more new classes. During the move and the split operations, the subsystem takes into consideration the subclasses, data properties, object properties and individuals that belong to classes under operation.

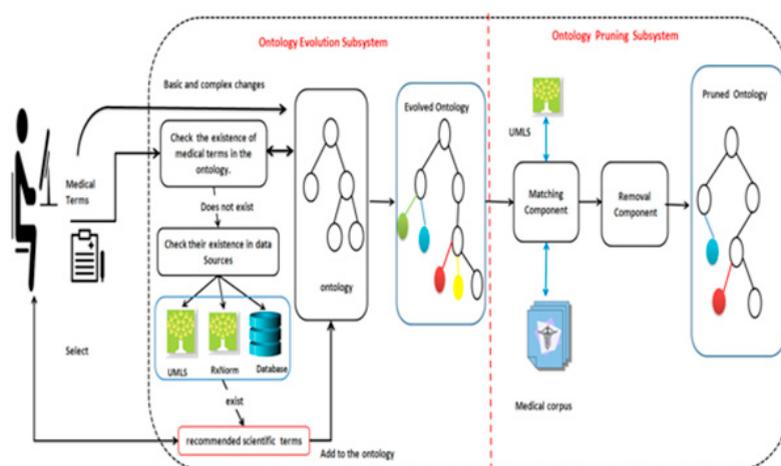


Fig..1. - Ontology Maintenance System Architecture

## 2.2. The ontology pruning subsystem

The ontology pruning subsystem involves two phases; the matching and the removal. In the matching phase, the pruning subsystem takes the evolved ontology as input and gets the ontology elements (classes, subclasses and individuals) to match them with the UMLS and the medical corpus. The matching is executed between the ontology elements and the UMLS concepts that belong to the immunosuppressant, antirheumatic and autoimmune drugs (since the UMLS contains more than 9000000 medical concepts and the Rheumatoid disease belongs to the autoimmune diseases that share a common group of treatments called the immunosuppressant). The unmatched ontology elements are checked with the medical corpus which produces a new list of unmatched terms that are to be removed from the ontology in the removal phase.

The medical corpus is a collection of medical articles created by several experts in the domain of the Rheumatoid Arthritis. It consists of the following corpora:

American college of Rheumatology guideline for the treatment of Rheumatoid Arthritis 25 published by PubMed articles 26.

Rheumatoid Arthritis: early diagnosis and treatment outcomes 27.

Juvenile Rheumatoid Arthritis 28.

Rheumatoid Arthritis medications 29.

Types of Rheumatoid 30.

### 3. The System Evaluation

The evaluation of the developed ontology maintenance system is performed through three steps; evaluating the ontology evolution subsystem according to ontology evolution life cycle proposed by Zablith<sup>16</sup>, comparing the ontology evolution subsystem with the other medical ontology evolution systems and evaluating the ontology pruning subsystem. Table 1 demonstrates the applied ontology evolution tasks in the proposed system and other approaches. Most of the ontology evolution approaches focus on detecting the need for evolution, suggesting changes and validating changes tasks. Only the sub ontology evolution approach executes the assessing impact task. Both of the DyKosMap and the proposed system perform the managing changes task. The mining relation reversals approach focuses on the discovery of inconsistencies that can be occurred on evolved ontologies, therefore it doesn't execute most of the ontology evolution tasks.

For the proposed system, the task of detecting the need for evolution relies on the user and data sources (the UMLS, the Rxterms and the treatment database). The user will determine the need to update the ontology. For the suggesting changes task, the candidate changes are provided by the structured data sources which provide the updated commercial names and the scientific names of a treatment. The validating changes task relies on the data sources to ensure that the medical term inserted by the user is relevant to the domain of interest before adding it to the ontology. It is implemented through the use of a set of rules that prevent the changes that may cause inconsistency, e.g. the system will prevent the user to add a class, an individual, an object property or a data property that is already exist in the ontology. Furthermore, if the user decides to apply merge/split operations, the system executes a set of rules that ensures the ontology consistency after performing the required operation (the system takes into account the subclasses and individuals of the class(es) affected).

Table 1- The Evaluation of Evolution Approaches according to the Ontology Evolution Life Cycle presented by Zablith

Criterion/ Ontology Evolution Approaches	Detecting the need for the evolution	Suggesting Changes	Validating Changes	Assessing Impact	Managing changes
SemCado	Yes	Yes	Yes	No	No
DyKosMap	Yes	Yes	Yes	No	Yes
Sub ontology evolution	No	Yes	Yes	Yes	No
Mining relation reversals	Yes	No	No	No	No
SEAM	Yes	Yes	Yes	No	No
The Proposed System	Yes	Yes	Yes	No	Yes

For the assessing impact task, the proposed system can be integrated with a health care application in order to assess the impact of ontology changes on this application. For the managing changes task, the system applies the changes to the ontology and keeps these changes in a log file. Table 2 shows the comparison between the developed ontology evolution subsystem and other medical ontology evolution systems. This comparison is based on the following points:

- Input.
- Consistency verification 31 Applying a set of rules or constraints to prevent the changes that can violate the ontology consistency.
- Change implementation 32 Allowing the user to accept or decline the proposed changes.
- Kind of changes It involves the execution of basic and/or complex changes.
- Output.

Table2- The Comparison between The Proposed System and Other Ontology Evolution Systems according to Input, Consistency Verification, Change Implementation, Kind of Changes, and Output.

Criterion/ Approach Name	Input	Consistency Verification	Change Implementation	Kind of Changes	Output
SemCado	Observational dataset and single domain ontology	Yes	Yes	Basic	Causal Bayesian network with suggested new causal relations and enriched ontology
DyKosMap	New published versions on KOS and current mapping	Yes	No	Complex	Up to date mapping between evolved KOS
Sub Ontology Evolution	Health ontology and its change log	Yes	Yes	Basic and Complex	Creation of evolved sub ontologies for every health applications
Mining Relation Reversals	For transitive closure, ontology concept and isa relations. For inversal relations, transitive closures' concept pairs of two versions	Yes	No	Detects the reversal relations	Transitive closure and inversal relations between two ontology versions
SEAM	Clinical and biomedical text	Yes	No	Detects the changes in the domain	Recommended terms, synonyms and relationships
The Proposed ontology evolution Subsystem	Medical ontology and Medical term/ Complex changes	Yes	Yes	Basic and Complex	Recommended standardized terms to be added in the ontology. Evolved ontology

The proposed system uses a set of rules for each kind of change to ensure the consistency of the evolved ontology. The proposed system enables the user to implement both of the basic and complex changes. For the ontology pruning subsystem, the evaluation relies on the testing of two ontologies that belong to the Rheumatoid disease which are; a Rheumatoid ontology developed by the authors and the RAO ontology which is provided by the Bioportal 33. The constructed Rheumatoid ontology consists of 18 classes, 95 individuals and 8 object properties while the RAO ontology consists of 280 classes, 0 individuals and 0 object properties. After the application of the pruning subsystem, the constructed Rheumatoid ontology contains 16 classes, 71 individuals and 5 object properties and the pruned RAO contains 278 classes.

#### 4. Conclusions

What distinguishes the proposed ontology maintenance system from the other systems is that it executes both of the ontology evolution and the ontology pruning. In the ontology evolution, the basic and complex changes can be implemented taking into consideration the consistency of the evolved ontology. The pruning process ensures the relevance of ontology concepts and individuals to the domain of interest. In future work, the ontology maintenance system can be used in a DSS system for patient management.

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