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Modular Ontology Design: A State-of-Art of Diseases Ontology Modeling and Possible Issue

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ABSTRACT

The evolution of ontological engineering leaded authors to use some techniques of software engineering to design ontologies. Are obtained from these techniques the monolithic or modularized Ontologies. When is difficult to reuse some concepts of monolithic ontologies, modularized Ontologies facilitate ontology management, understandability and reuse. This paper aims to survey on ontology modularization techniques and their contribution in biomedical ontologies design. Modularization reposed on appropriated techniques and some challenges related to ontology reused, scalable querying, collaborative authoring, and distributed reasoning. For most of disease ontologies, more especially ontologies which reused IDO, these challenges are not considered, and most of them are implemented with OWL language and the novel mode to construct ontology's purpose is to facilitate reuse and interoperability of ontologies ensured by modularization.

1. INTRODUCTION

Several domains manipulate knowledge and the deal of organizations is to share this knowledge among machines and persons. It is needed to use tool to represent this knowledge. Among growing tools, authors used the core component of Semantic Web named ontology. Ontology is the set of concepts used to describe and to represent a certain domain. A formal definition is given by Gruber as *"An ontology is an explicit specification of a conceptualization"* [1] and completed by Borst as *"An ontology is an explicit and a formal specification of a shared conceptualization"* [2]. The second definition brings out the consensual aspect of ontology. There are three basic levels on ontologies: Top-level or Upper-level ontologies, Core-domain ontologies and Domain ontologies [3]. The Top-level ontologies describe abstract structure knowledge of various domains. Their organization is based on philosophic and fundamental cogitations. They are pillar for core-domain and domain ontologies. Among them, we have BFO (Basic Formal Ontology), GFO (General Formal Ontology), OBO (Open Biological Ontology), DOLCE (Descriptive Ontology for Linguistic and Cognitive Engineering), SUMO (Suggest Upper Merged Ontology) … The Core-domain ontologies or generic ontologies describe generic concepts for a certain domain and the relationships amen these concepts. Examples of core-domain ontologies are IDO (Infectious Diseases Ontology), LKIF-Core Ontology, NIF-Core Ontology. The Domain ontologies describe all concepts for a target domain. They are considered as lower-level ontologies. Other categories of ontologies can be associated to lower-level ontologies: applications and task ontologies. These ontologies are monolithic or modularized. Ontology is said monolithic if the concepts are in one unique bloc. To reuse some concepts of this kind of ontology, it is an obligation to reuse global ontology. Managing global ontology is a serious challenge for ontology designers, reasoners and users. One generic strategy to deal with the problem is modularization, which aims at replacing a huge ontology by a collection of smaller components called modules that can be manipulated independently from each other and are nevertheless capable of collaborating in providing the same service as the whole initial ontology [4, 5]. The goals for ontology modularization are scalability for querying data and reasoning on ontologies, ontology evolution and maintenance. It helps to facilitate ontology management, understandability and reuse [6]. A very important task is how to obtain modules for ontology. Someone requires composition of modules and for other it is decomposition of huge ontology to modules. For the first technique, it consists to develop independently modules and to integrate coherently and uniformly these modules. The second technique concerns extracting such modules from integrated ontology for supporting a particular use-case [4, 7]. But sometimes, designers want to build ontologies derived from several ontologies and there is no issue outside importing whole ontologies. Before thinking about modularization of ontology, it is needed to define some axes concerning use-case of ontology, type of modularization, technique which should be used, properties of modules and evaluation metrics to validate modular ontology [8]. There exist various techniques for ontology modularization. Some of these techniques are described in [4-6, 9-14]. The most popular language used in ontologies design is OWL (Web Ontology Language) and its extensions. Nevertheless, for some authors, this language presents some limits concerning the designing of modular ontology such as not supporting of localized semantics, directional semantics relations, partial reuse. Generally, OWL does not handle globally semantically sound for modular ontology [15]. Jie et al. [15] proposed an Abstract Modular Ontology (AMO) which solved some problems of OWL language. To support modular ontology designed, Shimizu and Hammar [16] proposed a plugging for *Protégé* to help users while wish construct modular ontologies by integrate some Ontology Design Patterns (ODP). In biomedical domain, especially diseases sub-domain, various ontologies are constructed to model target diseases, to share knowledge concerning a certain disease. Despite protrusion of modularization, most of the disease's ontologies have not implemented modularization and some of them reused IDO by adding some particular concepts referred to this disease. Nevertheless, modularization and its challenges can be asset for medical domain, hence this paper aims to revisit some techniques of modularization, the challenges and how it can be possible to integrated machine learning techniques. For instance, hidden Markov models offers possibilities to capture knowledge by learning properties and then define some properties for clustering before partitioning them into modules. The rest of the paper is organized as follow: the Section 2 is reserved to the definition of some terms related to module, modular ontology and disease. The Section 3 describes some methods for ontology design especially for modular ontology and Section 4 is devoted to some examples of diseases ontologies. The Section 5 focuses in discussion and the last Section is devoted to the conclusion and future trends/challenges.

2. DEFINITION

A disease is a disorder of structure or function in human, animal or plant, especially one that produces specific symptoms or that affects a specific location and is not simply a direct result of physical injury [17]. Some categories of diseases occur in tropical zone - zone situated between Cancer and Capricorn tropics - and are classified according to their vector agent: bacteria, virus, parasite and microscopic fungus [18]. Some diseases are neglected [19] and others are handled. A disease is characterized by three aspects: clinic, biologic and epidemiologic [20]. The clinic aspect covers treatment, drugs, symptoms, materials used, etc. The biologic aspect covers vector agent, its transmission, living environment, etc. The epidemiologic aspect covers the impact of disease in society, the actions of communities face to disease and its expansion in these communities. To share knowledge by experts about these diseases, among various tools, authors used ontologies to model diseases.

Ontology is a set of concepts joined by relationships and based on some functions or axioms. It is used to refer to a body of knowledge describing some domains, typically a common-sense knowledge domain, using a representation vocabulary. Given a target domain, its ontology forms the heart of any system of knowledge representation for that domain [21]. The components of ontology are: concepts, relations, instances and axioms. Concepts represent a set of entities within the domain. Relations specify the interaction among concepts. Instances indicate the concrete examples of concepts within the domain and axioms denote a statement that is always true [22].

Ontology can be monolithic or modular. Monolithic ontology is a huge ontology in which all concepts are joined among themselves and have a unique semantic. Modular ontology is ontology in which concepts are divided into modules. In software engineering, for example, module is one of the terms used to denote software component that is designed to perform a given task and is intended to interact with other modules within larger software architecture [6]. In ontology domain, a module is a subset of global ontology where concepts are heavily linked and are coherent and make sense [4, 5].

3. ONTOLOGY DESIGN METHODOLOGIES

3.1 General case

Ontology engineering describes tasks to design ontology for a domain. To achieve their goal, designers used growing types of methodologies, among them: TOVE (Toronto Virtual Enterprise), Methontology, Ontolingua, NeOn Menelas, Uschold et al., etc. These methodologies are revisited in references [23-25]. Whatever methodology used, some common steps are used. These steps are: specification, acquisition, conceptualization, formalization, implementation and evaluation. The specification is the step where the type of ontology (upper-level, core, domain, task, process, application) is defined according to the objective of ontology. In acquisition step, concepts and relations are collected nearby experts or several inputs. Conceptualization step models knowledge. In formalization step, logic representation is used for reasoning. The implementation step consists to translate formal model using ontology language such as RDF/RDFS, OWL, etc. Evaluation step consists to measure the efficiency of ontology and verify if it satisfies specifications [7].

3.2 Modular ontologies case

Ontology modularization aims at providing users of ontologies with knowledge they require, reducing the scope as much as possible to what is strictly necessary [5]. Construction of modular ontologies reposes also under the methodologies cited in Section 3.1. However, this construction is guided by some challenges: ontology reuse and alignment, value set construction, secure information exchange, collaborative authoring, distributed and incremental reasoning, scalable querying, etc. [4]. Before thinking about modularization, it is needed to define some axes concerning use-case of ontology, type of modularization, technique which should be used, properties of modules and evaluation metrics to validate modular ontology [8]. There exist various techniques for ontology modularization. Among these techniques, we can cite [4]:

(1) Distributed Descriptive Logics (DDL): it is a knowledge representation formalism intended to enable reasoning between multiple ontologies connected by directional semantic mapping [26].

(2) ε-Connexion: it is a set of "connected" ontologies. An ε-Connected ontology contains information which refers to classes, properties and linked properties and their instances [27].

(3) Package-based Description Logics (P-DL): it is an extension of DDL where concept, role and nominal names can be shared by "importing" relations among modules [28].

(4) Conservative Extensions: it consists to give the meaning of a concept of one ontology by reusing a small fragment of "foreign ontology" [29].

(5) Graph-based ontology segmentation: it consists of partitioning ontology represented as a graph to obtain modules. There exist several tools for segmentation described in [4].

In this list of techniques, DDL, ε-Connexion, P-DL and Conservative Extensions correspond to the approaches that focus on the composition of existing ontologies (modules). And Graph-based ontology segmentation corresponds to the approaches for modularizing ontologies in terms of ontologies partitioning and ontology modules extraction [5]. Using the last technique, some semantics can be decrease according partitions properties. Furthermore, authors proposed framework to support ontology modularization [8, 16, 29-33].

4. ONTOLOGIES FOR SOME DISEASES

In ref. [4], some challenges are outlined for biomedical domain. Indeed, the modularization has been applied to design ontologies of some diseases but it is insufficient because most of them reuse IDO [34] which is not modular.

Before 2010, some works existed related to share knowledge among medical communities [35] like OntoNeuroLog build in 2007 by Temal et al. [36] to assume integration of heterogeneous, brute and treated date, interoperability treatment tools. They reused DOLCE, I&DA and COPS. But concrete diseases representation by ontologies took their place around 2010 with the conception of IDO [34] by Cowell and Smith. IDO is a core ontology based on OBO which can be reuse by authors to extend to a particular disease domain because it ensures interoperability between the domain-specific extensions. This is the reason why some authors reused IDO to build diseases ontologies. In 2010, Topalis et al. [37] extended IDO to propose IDOMAL, an extension of IDO for Malaria and revisited in 2013 [38]. This ontology reused BFO and RO and covers aspects of malaria disease. In 2011, Lin et al. [39] extended IDO to propose IDOBRU, an extension of IDO for Brucellosis. The main goal of this ontology is to model various aspects of brucellosis. It imported several ontologies (BFO, RO, CHEBI, GO, IAO, NCBI, OBI, OGMS, PrO, VO) to cover all brucellosis aspects. In 2008, Pierre and Bernard-Alex [19] extended IDO to propose IDOSCHISTO, an extension of IDO for Schistosomiasis. It imported various ontologies (BFO, IDSDO, TRANS, RO, DOID, PCO, NCBI, OPL, OMRSE, ENVO, EXO) to cover all aspects of Schistosomiasis disease. Use-case did in Richard Tall Town in Senegal helped to evaluate and validate ontology. In 2015, Mitraka et al. [40] extended IDO to propose IDODEN, an extension of IDO for Dengue fever. IDODEN is modeled on IDOMAL and reused (IDOMAL, MIRO, ENVO, CL, SYMP, SO, UBERON, CARO, EFO, FB-BT, FLU, GRO, HP, NPO). In 2017, Khalil and Alfonse [41] proposed an ontology modeled hepatitis C virus infection. This ontology has validated by hepatitis domain experts. In 2018, Cardoso et al. [42] proposed ONTOPARON, ontology for modeling of SLA supported. This ontology reused NCCO and ONTOPAD. In the same year, Harjito et al. [43] proposed an automatic approach for bilingual tuberculosis ontology based on ontology design patterns (ODP). To achieve their goal, their used Text2Onto tool to collect terms under some corpus. Some resources like Tuberculosis Glossaries and ODP are used. In 2019, Béré et al. [44] extended IDO to propose IDOMEN, an extension of IDO for Meningitis. This ontology followed the spirit of IDOSCHISTO to cover the same aspects but concerning Meningitis. In 2020, with the accession of Coronavirus, in the one hand, He et al. [45] extended IDO to propose CIDO, a community-based ontology for coronavirus disease knowledge and data integration, sharing, analysis and reused some ontological resources (CHEBI, NDF-RT, DrON). And the other hand, Babcock et al. [46] extended IDO to IDO-COVID-19, an extension of IDO to COVID-19 derived from VIDO (IDO Virus) and CIDO (Coronavirus IDO). It reused some ontological resources (BFO, GO, RO, IDO, OGMS, CIDO). Among these ontologies, OntoNeuroLog, IDOSCHISTO, HCVO, TubO, ONTOPARON, CIDO and IDO-COVID-19 are modularized and not for the others. All them are build using OWL language. This list of diseases ontologies is not exhaustive and some of them are available on https://bioportal.bioontology.org and https://github.com. Ontologies described in this section are summarized in Table 1.

Table 1. Summarize table for some diseases ontologies

Ontologies	Ref.	Mod.	Ontologies reused
OntoNeuroLog	[36]	Yes	DOLCE, I&DA, COPS
IDO	[34]		OBO
IDOMAL	[37, 38]		BFO, IDO
IDOBRU	[39]		BFO, RO, IDO, CHEBI, GO, IAO, NCBI, OBI, OGMS, PrO, VO
IDOSCHISTO	[20]	Yes	BFO, IDO, IDSDO, TRANS, RO, DOID, PCO, NCBI, OPL, OMRSE, ENVO, EXO
IDODEN	[40]		IDOMAL, MIRO, ENVO, CL, SYMP, SO, UBERON, CARO, EFO, FB-BT, FLU, GRO, HP, NPO
HCVO	[41]	Yes	/
ONTOPARON	[42]	Yes	NCCO, ONTOPAD
TubO	[43]	Yes	ODP, Glossaries
IDOMEN	[44]		IDO
CIDO	[45]	Yes	CHEBI, NDF-RT, DrON
IDO-COVID-19	[46]	Yes	BFO, GO, RO, IDO, OGMS, CIDO

5. DISCUSSION AND POSSIBLE ISSUE

The contribution of modularization is not to be demonstrated in ontologies design since it insures scalability, evolution, maintenance, interoperability, management, understandability and reuse of ontologies [4-6, 10-12, 15]. Modularization can be done with bottom-top technique (build modules and join them to form ontology) or top-bottom technique (divided whole ontology into modules with appropriated techniques). For the first technique, the semantic of final ontology derives from local semantics of modules. For the second technique, semantic of ontology before modularization cannot be correctly conserved in modularized ontology. In [9], author used this second technique and show that the limit of this technique is the decreasing of semantic. For a particular domain, ontologies shared nearly semantic and build new modular ontology in this domain require to reuse all resources used by others ontologies. Hence, the main problem is to have same or near semantic before and after modularization.

Machine learning tools (hidden Markov models, support vector machine, Naive-Bayes, neural networks ...) offer possibilities to capture knowledge by learning properties. To conserve semantic of ontologies, these techniques can be used to learn ontology properties (semantic links) and then define some properties (for clustering) before partitioning them into modules. For instance, as far as ontologies are graphs, hidden Markov models (HMM) can be used to fix the modularization problem. We can consider classes as HMM states and relations as HMM symbols.

With a minimal of concepts derived from learning model a core ontology situated between core ontologies (IDO as example in the biomedical domain) and domain ontologies (diseases ontologies as example in the biomedical domain) could give a litheness to authors for constructing ontologies. Several reasons can maintain it: (1) authors will not have obligation to reuse all core ontology concepts but just the import concepts related to target domain in the desired core ontology, (2) with modular structure of this core ontology, authors can reuse a part (module) without import whole ontology and (3) the litheness will confer lightweight ontologies which can be easily portable.

6. CONCLUSIONS

In this paper, we overfeed the notion of ontology and the approaches to design it. We focused in modular ontologies design and techniques which could be applied by designers and challenges of modularization. We print out that the ontology modularization can be done either by bottom-top technique or by top-bottom technique. When in the first approach, the semantic of final ontology derives from local semantics of modules, in second, ontology semantic before modularization cannot be correctly conserved in modularized ontology. In addition, the both approaches are not usable when the modularization aims to capitalize (learn) from the existing same domain ontologies such as the disease domain.

Since the novel mode to construct ontologies purpose is to facilitate reuse and interoperability of ontologies ensured by modularization, previous works based on techniques cited in this paper and according to the usage of ontologies, we discussed about the possibility to integrated machine learning techniques to learn ontology properties. However, the deal will be how to conserve semantics insured by ontology. The Future work aims to propose a machine learning on-based framework to design the modular Ontologies.

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