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Description Logics as information resource: as example of its application in cardiology

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Abstract

This paper regards the study of terminologies, Ontology, and its inference in Description Logics. With no intent of broadening its comprehension, we depict description logics and its role as a mediator in Information and Computer Sciences and pointing at some of the most important resources that have currently caught the medical field's attention. Based on previous writings, the main causes of death nowadays in developed and developing countries are related to cardiovascular diseases. Data from the World Health Organization (WHO) state that about 140,000 people die every year in Brazil due to heart disease. In addition, most of these deaths, including those caused by sudden illness, could be avoided by the pre-monitoring and the pre-diagnosis of cardiac arrhythmias and myocardial ischemia using ECG, since the electrocardiogram is one of the tools that offer fast and reliable data sets for cardiac diagnosis. This study has as a tool the analysis for the treatment and organization of digital information for its recovery, use, and reuse in a monitoring and automatic pre-diagnosis system to assist the doctor in quickly and accurately detecting cardiac arrhythmias. Therefore, the idea, in the methodological light of Information Sciences - creating, processing, preserving, and (re) using digital information - is to develop a system to generate, store, transfer, and interpret electrocardiogram information for cardiac arrhythmia diagnosis. The system should be used in health centers, mobile ICUs, hospitals and universities to assist health professionals when working on medical reports of patients with pathological abnormalities. As a result, it is expected that a system containing database of electrocardiograms, with pathologies related to clinical cases of cardiac diseases, will be introduced and approved as an assisting tool for health professionals to use throughout the process of managing information. They will prepare their patient's clinical report to pass on information, following the principle of ubiquity, which when deployed in public health systems will provide faster assistance and accuracy in the identification of cardiac abnormalities, and patients will have specialized service. Thus, we introduce a structure of description logics and its approach in the cardiology medical environment, and we compare the use of description logics in the pathology environment by using a practical model of description logics use in terms of diseases related to the circulatory system of the human body.

Keywords

Terminologies; Ontologies; Description Logics; Medicine; Cardiology; Database

Introduction

This present study aims at depicting a description logics analysis as information resource in cardiology, by means of a domain structure and ontological knowledge based on the International Classification of Diseases (ICD), approaching Diagnostic Related Groups (DRG). With Description Logics, knowledge representation takes place through functional approach, ie, precise specifications are provided of functionalities to be given by knowledge base and of the inferences to be made. Thus we emphasize the fact that creating a domain ontology in cardiology based on some ontological reasoning is useful and necessary, to provide support for ontological decision making, based on the structure of detailed descriptive logics, allowing health professionals to make decisions based on a medical ontology of cardiology; thus we state that a description logic consists of a descriptive language that is used to define how concepts and roles are formed, through a system that specifies data about the concepts and roles, and a system that can specify the properties of elements and their ways of inferring on a knowledge base.

The insertion of description logics analysis in the healthcare environment can be made in health centers, hospitals, and universities, aiming to help professionals when working with reports of patients that have heart abnormalities and pathologies. Pre-monitoring and automatic pre-diagnosis aim to assist the physician in a faster and more accurate detection of cardiac arrhythmias. Through Information Sciences and its learning within an interdisciplinary field, which includes all aspects of information generation, via measurement and observation, through capture, analysis, representation, organization, evaluation, storage, processing, presentation, protection and retention. An image database that contains several types of electrocardiograms will be provided with pathologies related to clinical cases of cardiac diseases, and, thus, it will become a fundamental tool for health professionals in the task of making accurate reports about their patients. That will allow health professionals to make decisions through Information Sciences to determine the laws and principles that belong to the analysis, development, and assessment of data, information, and system of knowledge.

Ontology and its inference in description logics

For Information Science, and other interdisciplinary fields, the responsibility to process knowledge is one of its concerns; and the consistency of the terminology used to find and classify information is one of its purposes. Thus the importance of using ontologies to describe and relate entities in a domain of knowledge. The term ontology has been discussed and applied in various scientific fields, Information Science, Computer Studies, Medicine, among others. Originating in philosophy, with the meaning of "category", it is represented for being used to sort something. Several definitions for ontologies can be found in publications, several types, propositions for its application in different areas of knowledge, and propositions for constructing ontologies, methodologies, tools, and languages (ALMEIDA et al., 2003).

Ontology would be an explicit specification of a conceptualization, in other words, the explicit definition of concepts and their relations, properties, and constraints formally expressed. It allows you to specify an abstract, simplified view of a subject that needs to be represented (GRUBER, 1993).

Using ontologies makes the developer able to perform high level reuse, which is a constant problem in the Computer and Information Science; it also allows applications of domain of knowledge to be shared, singularly used in the vocabulary applied to heterogeneous software platforms, in Information Science use of the ontology term, applies to the evolution of publications on the subject over the years, the frequency of keywords associated with the papers of studies related to ontology. As Figure 1 shows a classification of types of ontologies

according to their level of dependence on a particular task or point of view, it distinguishes top-level, domain, application, and task ontologies (GUARINO, 1998).

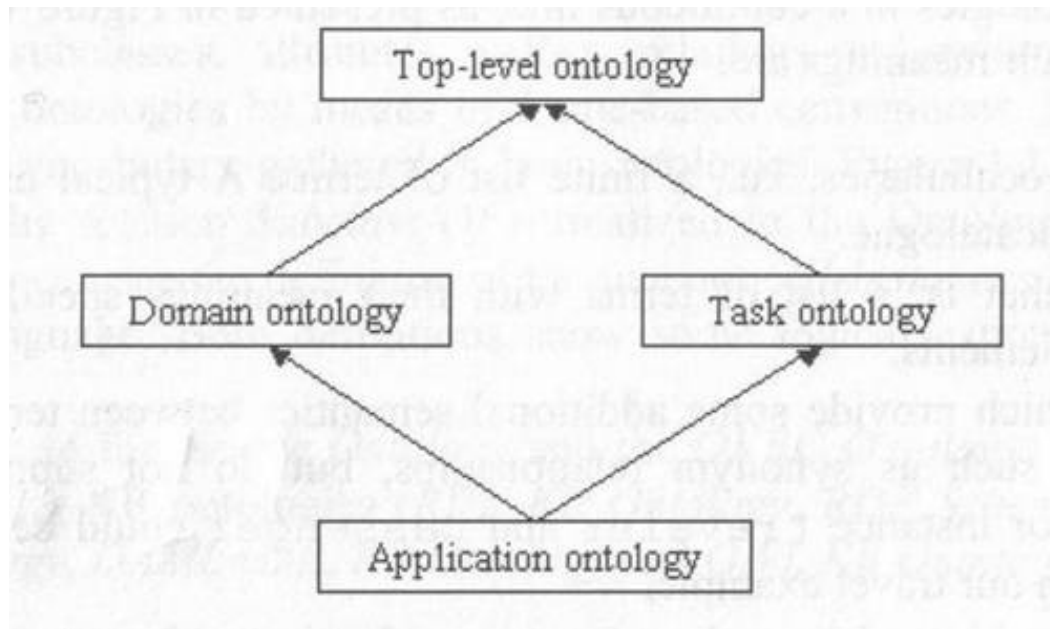


FIGURE 1 - Categorization Source: (Guarino, 1998).

The treatises of Aristotle study categories that can classify entities and introduce the term "differentia" for properties that distinguish different species of the same genus. The technique of inheritance is the process of merging "differentias", defining categories by genus and differentiation. Aristotelian contribution to the study of Ontology is the basis for the norms of inheritance and the foundation of description logics (GUARINO, 1998).

Description logics and its representation

Description Logics *DL* is a family of formalisms based in first-order logic for knowledge representation; they are considered to be the most important formalism for the representation of knowledge, unifying and providing logic base for traditional systems in this area (frames, semantic networks, object-oriented representations, semantic data models and systems of types). Description Logics is used to design systems that have a language to define a KB (*Knowledge Base*), and tools to make inferences on the basis set (NARDI *et al.*, 2003) .

With Description Logics knowledge representation takes place through functional approach, ie, precise specifications are provided of functionalities to be given by knowledge base and of the inferences to be made; see figure 2 (NARDI *et al.*, 2003).

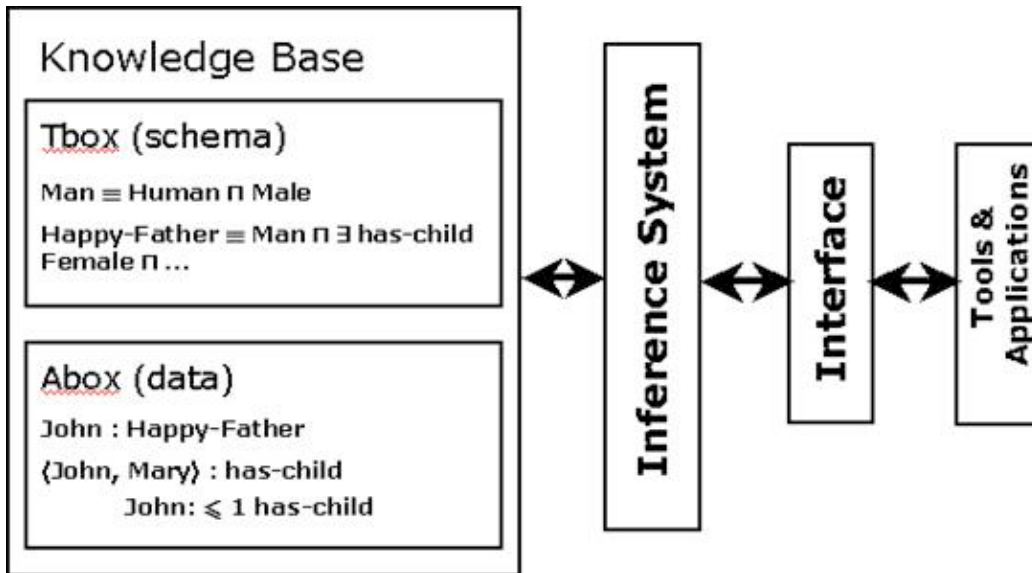


Figure 2 - Architecture of a DL system Source: (Horrocks, 2002).

In practice, the functional description of the system is made using an interface called "Tell&Ask", which specifies operations that allow the construction of the knowledge base (Tell operations) and operations that allow information from the base of operations Ask. Within the knowledge base one can make a clear distinction between "intensional knowledge" (general knowledge about the problem domain or universe of discourse) and "extensional knowledge" expertise in a particular problem. Analogously, the architecture of a DL system comprises two components a "TBox" and an "Abox"; it presents a typical architecture of a system based on Description Logics (NARDI *et al.*, 2003).

Figure 3 shows that the TBox contains intensional knowledge in the form of terminology or taxonomy, it is built by statements that describe general concepts and their properties (HORROCKS, 2002).

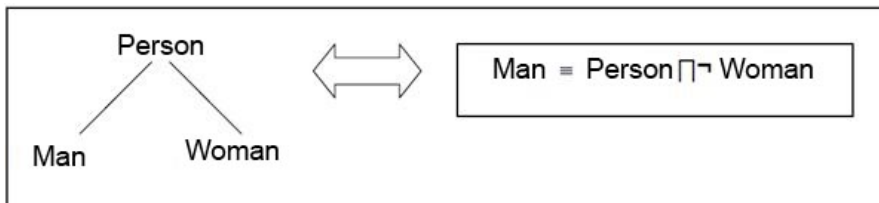


Figure 3 - Example of TBox intensional knowledge Source: (Horrocks, 2002).

The ABOX contains extensional knowledge, also called "assertional" knowledge that is specific to the individuals of the domain, as shown in figure4 (HORROCKS, 2002).

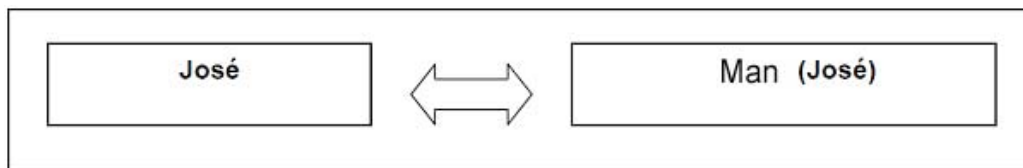


Figure 4 - Example of ABox extensional knowledge Source: (Horrocks, 2002).

The Description Logics describes the area in terms of concepts (classes), roles (properties, relationships) and individuals. In terms of logic, the names of concepts are equivalent to unary predicates - in general, formulas with one free variable. The names of roles are equivalent to binary predicates, in general, formulas with two free variables. The names of individuals and or elements are equivalent to constants. The set of operators is restricted so that the language is decidable and with low complexity. Through these systems, not only can knowledge base be validated in relation to its correctness, but also implicit knowledge can be made explicit from the knowledge expressed in the base. These two aspects can be reduced to the following tasks of inference (RUSSELL *et al.*, 1995).

Description logics and its approach in the medical environment of cardiology

The terminology in medicine, especially in cardiology, is characterized by a large amount of calls, which are best described as linguistic artifacts that link the various senses or meanings of linguistic entities. Terminologies in cardiology are often built with well-defined purposes such as for retrieving documents, pointing to resources, recording statistics of mortality and morbidity, or billing for health services. Terminologies in cardiology do not use formal well-defined descriptions, they define the terms through human language expressions and express associations between terms through informal relations, close to those of human language, formed from Greek and Latin roots, prefixes, and suffixes; aiming to simplify language, find accuracy in the meaning of words, and scientific exchange between nations of different languages of culture. A great amount of data used to solve complex tasks requires increasingly sophisticated techniques for smart management of information and knowledge, increasing interoperability of content in large repositories supported by different types of automated reasoning (REZENDE *et al.*, 1998).

The emergence of a growing set of semantic reference systems, often characterized as vocabularies, thesauri, terminologies, and ontologies, is practical result of these efforts. The current progress of knowledge management in cardiology has essentially had two reasons for so: the establishment of vocabularies and index classification systems, such as the International Classification of Diseases, and the Index Medicus, developed for interests of public health and epidemiology; and the decision support systems and medical specialists, in the 1970s, inspired by the computer industry in the subarea of Artificial Intelligence to assist medical decisions (RUBIN *et al.*, 2008).

Therefore, the standardization of terminology in medicine has its long history. It was established in 1880 as the International Classification of Diseases (ICD), based on the *London Bills of Mortality*, to identify an approximate number of 200 causes of death, and it would generate codes to every single diagnosed disease at the time. As shown in Figure 5, the ICD is also used as basis for the *Diagnostic Related Groups - DRG* used in billing and medical care (WHO, 1995).

In the field of Bioinformatics, it represents the application of formal ontology to the representation of biological entities (SCHULZ *et al.*, 2007).

| | |
|--------------|---|
| H52 | Disorders of refraction and accommodation |
| H52.0 | Hypermetropia |
| H52.1 | Myopia <i>Excludes: degenerative myopia (H442)</i> |
| H52.2 | Astigmatism |
| H52.3 | Anisometropia and aniseikonia |
| H52.4 | Presbyopia |
| H52.5 | Disorders of accommodation Internal ophthalmoplegia (complete)(total) Paresis } Spasm } of accommodation |
| H52.6 | Other disorders of refraction |
| H52.7 | Disorder of refraction, unspecified |

Figure 5 - Extracted from the International Classification of Diseases, 10th version Source: (WHO, 1995).

Ontology and its principles for medicine

Following the principles is the *Open Biomedical Ontologies (OBO) foundry*; so far, it consists of 60 ontologies that, without changing much in terms of granularity, canonicity, and stage of development, each one intends to represent a well-defined object of study (ROSSE *et al.*, 2003).

Among the most frequently consulted OBO ontologies, there is the Fundamental Model of Anatomy (FMA) (FMA, 2001).

The FMA deals with the anatomical structure of the mammalian body (especially human). However, despite the fact that the domain of electrophysiology of the human heart is significantly important to Biomedicine, there is still no electrophysiology ontology in OBO, neither in the literature of biomedical ontology (SMITH *et al.*, 2007).

For the study of cardiac diseases, the ECG in cardiology is the most frequently used test to measure the activity of the heart, using digital signals (GESELOWITZ, 1989).

In recent years, both the storage and transmission of ECG recordings were the subject of standardization initiatives; see figure 6. However, the focus of these standards is typically as data and information to be represented in computer systems and transmission of messages with the purpose of aiding in the diagnosis (SMITH *et al.*, 2007).

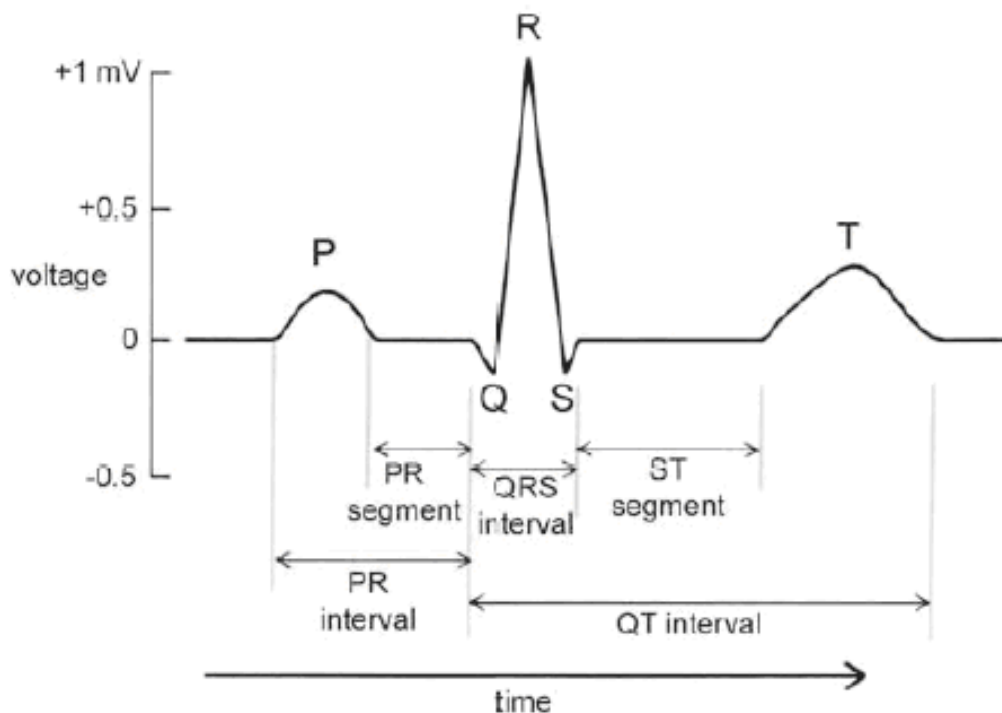


Figure 6 - A typical cycle of the heartbeat on the ECG waveform Source: (Laske; Iaizzo 2005).

Functional synthesis of the heart and its pathologies as elements for description logic

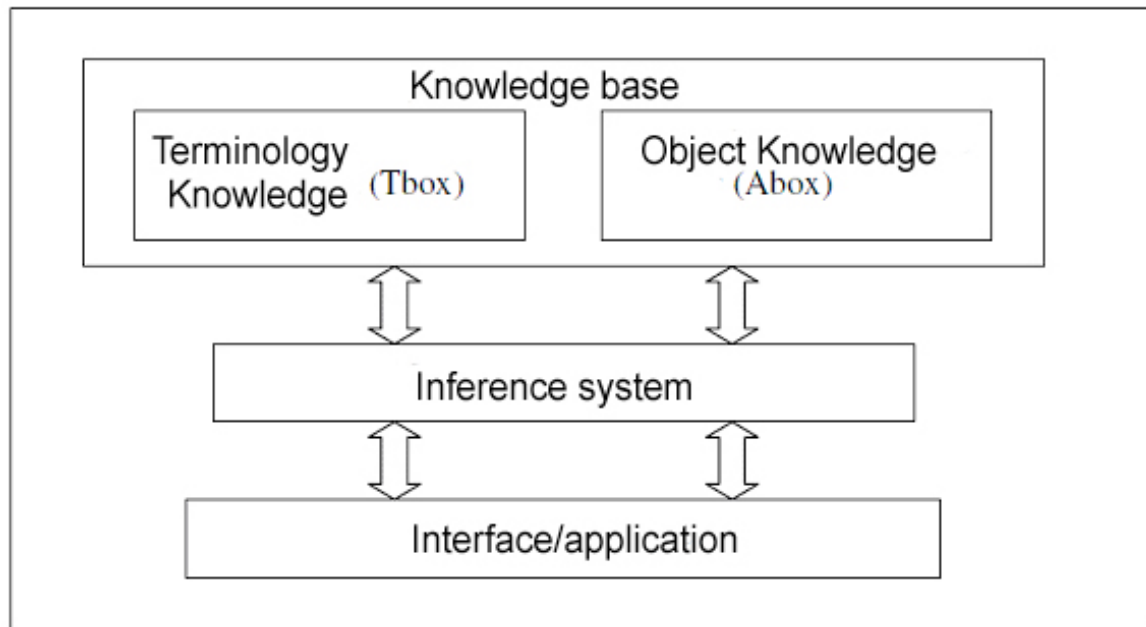
Heart as a pump, within the concept, is constituted by two separate pumps: the right heart that pumps blood through the lungs, and left heart that pumps blood through peripheral organs. Each of these separate hearts is a pulsatile two-chamber pump composed of an atrium and a ventricle. The cardiovascular system consists of a closed system of vessels, veins, and capillaries, heart, and a vascular network of distribution. Periodically stimulated, the heart muscles contract pushing the blood through the vessels to all parts of the body, thus, before several physiologic factors a patient may develop. Heart Diseases, which are factors that may condition patients to be exposed to increased risk of developing heart and blood vessel diseases. There are several risk factors for cardiovascular disease, which can be categorized into immutable and mutable (GUYTON, 1988).

As an example of these abnormalities, there is the Cardiopadia, which regard both structure and cardiovascular function, already present at birth, even if they are identified much later, may or may not develop the Heart Valve Disease, which is another factor that consists of a malfunction of the four heart valves. This defect may be in the opening or closing of valve. Therefore, stenosis, which is when the opening is smaller than normal or incomplete, and it develops valve insufficiency when its closing is incomplete or when there is perforation of the valve, these are the most frequent types of valvulopathy. The coronary arteries are in the early emergent branches of the aorta just above the aortic valve, and its onset can be observed in both coronary artery ostia, located in the aortic sinuses, or right and left Valsalva sinus. Having only one ostium or even more than two can occur, however rarely. It has been reported in literature up to five independent ostia. The development of coronary artery disease (CAD), a type of heart disease, is caused by the gradual blockage of the left and right

coronary arteries. The heart as a functional pump, gathers, oxygen and nutrients from the blood flowing through these arteries (ROBBINS *et al.*, 2000).

Description logics applied in cardiology

Note the fact that creating a domain ontology in cardiology based on some ontological reasoning is useful and even necessary. It should provide support for ontological decision making, but it should also allow us to make decisions as openly as possible in the resulting domain ontology. Based on a cardiology medical ontology, we may state that description logic consists of descriptive language, which is used to define how concepts and roles are formed, using a system to specify details of the concepts and roles (TBox), and a system to specify the properties of elements (ABox); Figure 7 shows the applied options of inferring on the knowledge base (GUARINO *et al.*, 2002).



(Knowledge base; Terminology Knowledge, Object Knowledge; Inference system, Interface/application)

Figure 7 - Structure of a system in Description Logic Source: (Calvanese, 2003).

Knowledge base consists of two parts, one that deals with terminology, which has a set of axioms and statements that describe the structure of the domain (TBox), also called intensional knowledge, and another which deals with statements about elements, or objects (ABox), ie, the ABox contains extensional knowledge about the domain of interest. The basic model of the statement in a TBox is the definition of concepts, and this basis consists of a set of inclusion assertions among the concepts. In the statement below there is the expression of the 'Valvular Heart Disease' concept, which is translated as an element belonging to the 'Heart' concept and to the 'Heart Disease' concept (NARDI *et al.*, 2003).

Valvular Heart Disease = Heart \cap Heart Disease

According to this other statement, in order for it to be Coronary Artery, the element must belong to the 'Heart' concept instead of belonging to the 'Valvular Heart Disease' concept.

Coronary Artery = Heart \cap \neg Valvular Heart Disease

For ABox one can make statements in two ways, where the construction of extensional knowledge takes place when concepts and roles make assertive statements about elements. The statement of concepts: Valvular Heart Disease (Stenosis), which means that the element 'Stenosis' belongs to the Valvular Heart Disease concept. The statement of roles: heart_diseases (stenosis, Right_Coronary_Artery), which means that the element 'stenosis' relates to the element 'Right Coronary Artery' through the role of 'heart_diseases'. There are different description logics, each one is appropriate to some particular situation. Examples are shown in table 8 where the components of the basic description language called AL, from which other description languages are extensions (CALVANESE *et al.*, 2003). Letters C and D represent description of concepts, letter R represents roles and letter A the atomic concepts.

| | | |
|--------|-------------------|---------------------------|
| C,D := | A | Conceito atômico |
| | T | Conceito universal |
| | \perp | Conceito botton |
| | $\neg A$ | Negação atômica |
| | C II D | Intersecção |
| | $\forall R.C$ | Restrição de valor |
| | $\exists R.T$ | Quantificação existencial |
| | $\exists R.\perp$ | limitada |

(Atomic Concept, Universal Concept, Botton Concept, Atomic Denial, Intersection, Restriction of Value, Existential Quantification, limited)

Table 8 - Grammar of description language AL Source: (Moreira, 2002).

The description logic systems not only store terminologies and assertions, but also enable services that perform inference about knowledge representation. The following sections will show typical services of inference in TBox and ABox, as well as a description of the Tableaux method of inference (VIEIRA *et al.*, 2005).

Unsatisfiability of the element

The satisfaction of the concepts checks if a concept description can never have instances because of inconsistencies or contradictions in the model. An example of unsatisfiability is the Cardiovascular System concept (VIEIRA *et al.*, 2005).

Valvular Heart Disease \equiv Heart \sqcap Heart Disease
 Coronary artery \equiv Heart \sqcap \neg Valvular Heart Disease
 Cardiovascular System \equiv Coronary artery \sqcap Valvular Heart Disease

Checking classification

Checking classification between two descriptions of concepts, C and D; C contains D when the set of objects that are instances of D are also a subset of objects that are instances of C. A practical example of this is the concept of HEART VALVE DISEASES, which is composed of instances of the 'Valvular Heart Disease' concept and has a relationship, through the 'heart_diseases' properties, with the heart concept (VIEIRA *et al.*, 2005).

Valvular Heart Disease \equiv Heart \sqcap Heart Disease

Heart valve diseases \equiv Valvular Heart Disease \sqcap \exists heart_diseases_.Heart

Equivalence among instances

Claiming that the concepts C and D are equivalent, $C \equiv D$, is the same as saying that they have the same instances. One example is the concept of Right Coronary Artery and Electrical Conduction Disorders of the Heart (Vieira *et al.*, 2005, p. 127-167)

Valvular Heart Disease \equiv Heart \sqcap Heart Disease

Right Coronary artery \equiv Heart \sqcap \neg Valvular Heart Disease

Electrical Conduction Disorders of the Heart \equiv Heart \sqcap \neg Heart Disease

Checking consistency and inference in ABox

The ABox model is consistent if there is an instance that makes both the

Abox and TBox true. This can be exemplified when you create an instance. The 'Valvular heart disease' concept called 'Stenosis'. As the

'Valvular Heart Disease' is formed by the concepts of Heart and Heart Diseases, Stenosis will also be part of these concepts. Valvular heart disease being a satisfiable concept, ABox is consistent (VIEIRA *et al.*, 2005).

The TBox model:

Valvular Heart Disease \equiv Heart \sqcap Heart disease

The ABox model:

Valvular Heart Disease (Stenosis) Consistency check:

Valvular Heart Disease (Stenosis) \equiv Heart (Stenosis) \sqcap Heart disease

(Stenosis)

Checking proceedings in elements

Here is checked whether a particular element is an instance of a specific concept. An example of such inference is to say that the Stenosis element is part of the Valve Diseases of the Heart concept, however what is explicit there is that Stenosis is a Valvular Heart Disease and it has a direct relationship, through the property of heart diseases, with Right Coronary Artery (VIEIRA *et al.*, 2005).

TheTBox model:

Valvular Heart Disease \equiv Heart \sqcap Heart Disease

Heart valve diseases \equiv Valvular Heart Disease \sqcap \exists heart_

diseases_.Heart

The ABox model:

Valvular Heart Disease (Stenosis)

Coronary Artery (Right Coronary Artery)

heart_diseases (Stenosis, Right Coronary Artery)

From this information we can say that the Stenosis element belongs to the Heart Valve Diseases concept.

Heart valve diseases (Stenosis) \equiv Valvular Heart Disease (Stenosis) $\sqcap \exists$ heart_diseases.Heart

Return of element

It finds the most specific concept on which of the elements is an instance, applying this service in the 'stenosis' element as to have as return the lowest concept on hierarchy of concept to which the element belongs, as shown in the models below (VIEIRA *et al.*, 2005).

The ABox model:

Valvular Heart Disease (Stenosis)

Heart valve diseases (Stenosis)

Return of element:

Stenosis \rightarrow Valvular Heart Disease

Performing identification on knowledge base

This service identifies, in the knowledge base, elements that are instances of a given concept (VIEIRA *et al.*, 2005).

The ABox model:

Valvular heart disease (valvular insufficiency) Valvular heart disease (Stenosis)

Coronary Artery (Left Coronary Artery) Result:

Valvular heart disease \rightarrow Valvular Insufficiency, Stenosis

Coronary Artery \rightarrow Left Coronary Artery

Conclusion

Our study has provided an analysis of description logics as information resource in cardiology; it allows an element of domain representation within the medical reality to the pathologies of the heart. With a well-structured and well-defined description logic structure, knowledge base, terminologies, and

its object inference precisely defined, it describes a framework acknowledge by the International Classification of Diseases (ICD), with an approach to the Diagnostic Related Groups (DRG). Since such ontology base is fundamental and necessary to provide the support

for the system of inference, which can become an application interface for decision making within the environment of Cardiology, as an important knowledge base of disease identification and classification with regards to the circulatory system, and, in this case, the heart has been the choice for the study. Nowadays, one of the main causes of death in developed and developing countries is cardiovascular disease. Data from the World Health Organization show that about 140,000 people die each year in Brazil due to heart disease. Most of these deaths, including those from sudden illness could be avoided if there was a pre-monitoring and a pre-diagnosis of such arrhythmias and myocardial ischemia using ECG. Electrocardiogram is one of the fastest and most reliable tools for diagnosing the heart. However, when a patient is monitored continuously for 24 hours, one can get a significant amount of data, reaching over 100 000 heartbeats to be analyzed by the physician. Therefore, this study aims to analyze the description logics to be used in health centers, hospitals, and universities, with the purpose of assisting health professionals in conducting follow-up reports of patients who have heart pathology abnormalities. Pre-monitoring and automatic pre-diagnosis aim to assist the physician in a faster and more accurate detection of cardiac arrhythmias. Therefore, it may be concluded that a knowledge base with the aid of description logic allows health professionals in an assisted or educational environment to identify in the knowledge base elements that are instances of a given concept.

Collaborators

The authors Paulo Augusto Loncarovich Gomes and Karina da Paz Loncarovich participated likewise on the elaboration of this article.

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