Chapter 7
Automated Diagnosis through Ontologies and Logical Descriptions: The ADONIS Approach

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ABSTRACT

The combination of the burgeoning interest in efficient and reliable Health Systems and the advent of the Information Age represent both a challenge and an opportunity for new paradigms and cutting-edge technologies reaching a certain degree of maturity. Hence, the use of Semantic Technologies for Automated Diagnosis could leverage the potential of current solutions by providing inference-based knowledge and support on decision-making. This paper presents the ADONIS approach, which harnesses the use of ontologies and the underlying logical mechanisms to automate diagnosis and provide significant quality results in its evaluation on real-world data scenarios.

1. INTRODUCTION

The rapid growth of research and development using Artificial Intelligence in biological and medical information systems has drawn worldwide attention on the management of medical knowledge (Liu et al., 2009). Since many artificial intelligence approaches have dealt with the diagnosis problem and its application in complex environments such as medical domains (Fuentes-Lorenzo et al., 2009), semantic technologies can provide a strong cutting-edge baseline for

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knowledge-oriented medical diagnosis systems. The semantic technologies (Berners-Lee et al., 2001), which have been developed and improved alongside the advancement of the Semantic Web, can be exploited to reveal machine-readable latent relationships within specific diagnostic-related information in the medical discipline, where the homogeneity of terminology is particularly problematic (Fuentes-Lorenzo et al., 2009). The specification of the domain knowledge (in our case, the medical domain) by the use of ontologies (Fensel, 2002) provides a knowledge-based system with the opportunity of adding the semantics of the domain, stating an explicit conceptual description of the domain (Fuentes-Lorenzo et al., 2009).

However, the efficiency and soundness of Semantic descriptions must be backed up by their underlying logic. The lattice of logic languages and formalisms is not a trivial issue and hence, an ontology must be perfectly defined and explained to serve as a basis for real-world medical applications. For this reason, an accurate and checked ontology should be defined in order to create a base for the medical diagnosis systems. Also, the description of the diseases, symptoms, laboratory tests and other clinical parameters should be done with rigor and checked by medical doctors. This description is the problem presented in the most of actual software for clinical diagnosis where not all the possibilities are taken into account, because in some cases this software are not able to make the correct inference of the disease.

This paper presents ADONIS, an architecture that includes a well-structured ontology for automated diagnosis and a three-fold formalization based on Description Logic that will allow to future medical systems that implements these techniques to perform real and more accurate diagnosis that traditional and actual systems.

The remainder of the paper is organized as follows. Section 2 outlines related research in the area. In Section 3, the architecture for the ADONIS approach is presented, which demonstrates technological support and potential solutions for problems in current medical diagnosis based in semantics and logical descriptions. An evaluation using calculation of precision and recall rates of the system is also presented in section 4. Conclusions and future work are discussed in Section 5.

2. STATE OF THE ART

The aim of this section is to provide a detailed state-of-the-art of the technologies present in this work and a description of the application domain.

2.1. Ontologies and Semantic Web

An ontology can be defined as "a formal and explicit specification of a shared conceptualisation" (Studer et al., 1998). Ontologies provide a formal, structured knowledge representation, with the advantage of being reusable and shareable. Ontologies also provide a common vocabulary for a domain and define –with different levels of formality- the meaning of the terms and the relations between them. Ontologies based on description logics (Gruber, 1995) or related formalisms provide the added benefit of enabling certain kinds of reasoning over the terms, relations and axioms that describe the domain. Knowledge in ontologies is mainly formalized using five kinds of components: classes, relations, functions, axioms and instances (Gruber, 1993). Classes in the ontology are usually organized into taxonomies. In computer science scenario, instances are equivalent to individuals in ontology terminology (Gómez-Perez, 2001).

According to Sicilia et al. (2009), ontologies are currently being used in the medical domain as an integration of heterogeneous sources (Dogac et al., 2006; Orgun & Vu, 2006) and as a tool to engineer formal knowledge descriptions from existing and diverse medical terminologies (Lee et al., 2006). A recent review on the use of ontologies in Medical domain can be found in Simonet