Chapter 26
Detecting Human Diseases Relatedness:
A Spreading Activation Approach Over Ontologies

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ABSTRACT

Due to the ubiquitous availability of the information on the web, there is a great need for a standardized representation of this information. Therefore, developing an efficient algorithm for retrieving information from knowledge graphs is a key challenge for many semantic web applications. This article presents spreading activation over ontology (SAOO) approach in order to detect the relatedness between two human diseases by applying spreading activation algorithm based on bidirectional search technique. The proposed approach detects two diseases relatedness by considering semantic domain knowledge. The methodology of the proposed work is divided into two phases: Semantic Matching and Diseases Relatedness Detection. In semantic matching, diseases within the user-submitted query are semantically identified in the ontology graph. In diseases relatedness detection, the relatedness between the two diseases is detected by using bidirectional-based spreading activation on the ontology graph. The classification of these diseases is provided as well.

1. INTRODUCTION

Usually, computers were used to perform tasks with none sort of intelligence or semantics, thus there is a necessity to make computers as intelligent as human (Antoniou & Harmelen, 2004). The defect of non-semantic applications is painted by the statement "lack of semantics" particularly when the talk is concerning information retrieval. Numerous semantic-based applications require an efficient algorithm for querying large ontologies. The semantic web gives the meaning of knowledge, making software systems can perform intelligent tasks instead of users using a semantic content called ontology (Berners-

Lee, Hendler & Lassila, 2001). Ontology is a repository in which information is organized and used in semantic-based applications (Shadbolt, Berners-Lee & Hall, 2006). Therefore, ontologies are widely used as a semantic knowledge base in different areas of computer science such as information retrieval (Oberle, 2014), text classification (Fathalla, Hassan & El-Sayed, 2012), scholarly communication (Fathalla, Vahdati & Auer, 2017) and bioinformatics (Fathalla & Kannot, 2017).

The use of ontologies in the field of health informatics has become a mainstream activity within bioinformatics due to the vast growth of healthcare system. In bioinformatics, ontology is used for the representing and organizing medical vocabularies.

In neurophysiology, communications between neurons are demonstrated by a way of activation, which propagates from one neuron to another via connections called synapses to transmit information using signals (Ali, 2009). This phenomenon observed in the nervous systems of living organisms and called spreading activation. This phenomenon was later exploited in Artificial Intelligence as a method for searching associative networks, neural networks, or semantic networks (Schumacher, et al., 2008). Spreading activation in a generic form is a set of methods suitable for mining multidimensional networks with oriented weighted links. Collins and Loftus (1975) discussed in their research that the Spreading Activation (SA) runs on semantic networks (Anderson et al., 1983) and is used for information retrieval process (Burns et al., 1981). The spreading activation algorithm is suitable to work with incomplete data and with large datasets. It runs on a graph structure that comprises a set of nodes connected by edges. The concepts are nodes which have an activation value and the relations between them are represented by edges between these nodes. An activation value is assigned to each node in the graph and then the algorithm spreads to the nodes with the highest activation value. The algorithm runs in a set of iterations and terminates when a stopping condition is reached. The output of the algorithm is the list of nodes that have been fired according to their activation values. For each iteration or cycle, there are three substantial actions:

- The list of nodes is expanded by adding adjacent nodes of the latest activated nodes.
- The activation value of each node in the list is recomputed based on the activation value of the node itself and the weight of links which exist between other nodes.
- The list is filtered by excluding the nodes with activation values less than a given threshold.

This paper intended to demonstrate how to identify whether two human diseases are related to each other or not. If so, what are the set of diseases connecting them. For instance, is there a relatedness between "Vasculogenic impotence" and "Transvestism"? And if so, what is the path (set of diseases) of the relatedness?

Finding a relatedness between diseases is important because physicians can treat patients not only based on the symptoms they suffer from but treat the real cause of this disease which may be related to another disease that causes these symptoms, i.e., physicians can treat the cause disease, not the symptoms.

To the best of our knowledge, it has not yet been established whether biomedical ontologies could be used to detect relations between human diseases. After reviewing the literature, we believe that this approach is a first such attempt of detecting the relations between diseases using ontologies combined with Descriptive Logic (DL) rules. We have added several Descriptive Logic (DL) rules to the ontology to be able to infer the semantic relations between diseases which are not explicitly provided. In addition, we have also developed the semantic matching and relatedness detector algorithm for identifying user-submitted diseases in the ontology graph and detecting the relatedness.

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