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PAPER

An Ontological Model for Artificial Reasoning Application to Medical Diagnosis

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ABSTRACT

Representing knowledge in a language that is both understandable by humans and easily exploitable by machines remains the subject of several research studies. Domain ontologies are recognized as an efficient way to describe knowledge through concepts and relations in several domains of expertise while remaining shareable and reusable. This paper aims to propose an approach for "artificial reasoning" that we consider as a foundational pillar for "Artificial Intelligence." Our particular interest in this work is on how to design systems that can use human knowledge to process and solve the complex problem of diagnosis, given the required expertise in a specific domain of knowledge. The approach we present in this paper is based on using properties of ontologies, by representing expert knowledge through a graph reasoning model, to formalize the diagnosis process using an ontology-based model. We first describe our proposal on how to represent expert knowledge in a general way before focusing on the diagnosis problem. Finally, we apply the whole process to the specific domain of cardiology.

KEYWORDS

ontologies, healthcare, diagnosis, artificial reasoning, conceptual graphs

1 INTRODUCTION

Artificial intelligence (AI) is the branch of science and engineering that aims to reproduce or simulate human intelligence as closely as possible within smart programs, which can perform several cognitive tasks that would normally require human intelligence [1]. AI algorithms can tackle learning, perception, problem-solving, language processing, and/or logical reasoning.

There are various sub-domains of AI, such as machine learning (ML), deep learning, natural language processing, computer vision, robotics, and expert systems, which are also known as knowledge-based systems (KBS).

Knowledge-based systems are a type of AI that relies on explicit knowledge representation in a set of rules and facts that are used to reason and make decisions to

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solve complex problems. We consider logical reasoning an integral component of KBS that enables the system to evaluate and reason about the represented knowledge. The main objective of this work is to examine how to leverage domain ontologies to define the process of artificial reasoning for diagnosis problems in various fields that require human expertise.

The remainder of the paper is articulated as follows: after introducing the context of our work in Section 2, we present some of the related work in Section 3. Then, we explain how we define an ontological model of a specific domain of knowledge, namely healthcare, and how we use it to develop a reasoning model in Section 4. In Section 5 and 6, we describe the use of our model to simulate the reasoning of a practitioner during a diagnosis process before applying it to medical diagnosis in cardiology. Finally, we present our implementation framework in Section 7 before concluding the paper in Section 8.

2 CONTEXT AND BACKGROUND

There are several definitions of an ontology. In [2], it is defined as "a formal and explicit specification of a shared conceptualization," while [3] defines it as "an explicit, detailed, and shared specification of a domain understanding, facilitating communication between pairs." A more formal definition of ontologies is given by [4], where "An ontology O is a structure $O = < C, R, I, A >$ where C is the set of Concepts of the modelled world. R is the set of Relations (taxonomic or not) between the concepts of C. I are Instances of the concepts of C, and A are the Axioms and inference rules expressed in a formal language and applicable on the concepts of C."

Among several existing types of ontologies, a domain ontology [5] is specialized in a particular domain (medicine, finance, etc.). Domain ontologies can be represented as hierarchical structures of concepts that make sense in the described domain. Concepts are described by terms and related by semantic (labeled) links so that it might be possible to represent all the vocabulary of a given domain, assuming this vocabulary is finite, in a corresponding ontology.

According to [6], the vocabulary of a knowledge domain can be provided (or represented) by an ontology, with a variable degree of formalism, fixing the meaning of concepts and the relations that unite them. From a philosophical point of view, there can be no vocabulary of a domain knowledge representation without the existence of the ontology (or the conceptualizations that form the basis of knowledge). Moreover, due to ontologies' structure and properties, a domain-specific ontology clarifies the domain knowledge structure, thus enhancing the power of conceptualization of that knowledge.

3 RELATED WORK

In the last decade, ontology usage has witnessed a growing interest in AI and information systems fields. Previous works have shown that ontologies can be used for expert knowledge representation, decision support, and semantic search. For example, [7] used an ontology-based smart search system algorithm for processing custom queries in e-commerce, while [8] proposed a cybersecurity ontology for the dynamic analysis of IT systems. [9] used an ontology to represent knowledge about business models and strategies to design an intelligent decision support system. [10] also proposed an ontology-driven system for the specific generation of exercises in an auto-evaluation learning context. Finally, [11] reviewed the usage of ontologies for knowledge representation in the context of the Internet of Things.

Ontologies have been used in medical diagnosis in several previous contributions, such as in [12] and [13]. While it is noticeable that these examples aim to build ontologies or to leverage them for knowledge representation, they generally do not focus on the reasoning during the diagnosis process. Our main goal is to leverage the characteristics of ontologies to define a way to "simulate/mimic" human reasoning during the diagnosis process, the goal being to reproduce the progressive character of knowledge acquisition on a given diagnosis problem.

While the results of ML-based approaches [14] have been impressive, it is important to acknowledge that these methods do not replicate the "natural" process of problem-solving. Training a model to recognize images or generate grammatically correct sentences using vast datasets comprising tremendous amounts of examples essentially involves a comparison process. The efficiency of this approach stems more from the large volume of data considered than from the inherent efficiency of the process itself, which fundamentally differs from the way humans learn and solve problems.

In contrast, traditional rule-based systems [15] typically rely on explicit knowledge representations, such as rules and facts, which are utilized by inference engines to reason and make decisions. While the reasoning process in these systems aims to mimic human reasoning, it faces challenges when dealing with incomplete information unless it has been explicitly programmed.

In this paper, we aim to explore the field of knowledge representation by introducing artificial reasoning models to define an ontology-based approach that "simulates" human reasoning. For this purpose, we focus on KBS that can be seen as programs operating inferences from a given knowledge base, enriching its content by deductions, inductions, etc. This objective led us to examine how representing expert knowledge by ontologies could allow the definition of ontology-based artificial reasoning models. We focus on domain ontologies, in order to leverage their structure and properties, to reach this objective for a given domain of knowledge.

This ontology-based approach enables reasoning through a distinctive knowledge representation framework. By representing the knowledge through a conceptual (semantic) graph rather than facts and rules in a knowledge base, and by totally separating the reasoning mechanism (inference) from the information (knowledge), this approach ensures that knowledge is both transferable and reusable across different systems. It also deals with the complexity of human knowledge that is considered a progressive "association of concepts." Furthermore, it enables the demonstrability of the reasoning, which is a critical feature for transparency, particularly in domains requiring close scrutiny (such as cardiology).

Finally, this approach allows us to leverage ML techniques for semi-automatic enrichment of our ontology. In other words, the ML models could be used to process massive amounts of data (structured, semi-structured, and unstructured) originating from different sources, as well as to identify entities (classes), patterns, and relationships in a domain of knowledge. This makes an expert (or a group of experts) able to assess and update the domain ontology accordingly.

4 REASONING AND KNOWLEDGE REPRESENTATION

For many years, ontologies have been identified by the research community as one of the most relevant solutions not only to represent knowledge but also to represent its processing mechanisms. This is enabled by their capacity to embed inference mechanisms that are helpful to include reasoning models in the global problem of knowledge representation and processing [16].

The reasoning process commonly consists of operating inferences and drawing conclusions from available (acquired) knowledge. Consequently, emulating this human ability is crucial for KBS in their process to create new "elements" of knowledge (facts). By combining existing elements of knowledge through inference mechanisms, new facts will enrich a pre-existing knowledge base. In the case of a diagnosis process, facts are generally obtained by requesting additional information on the considered subject through answers to selected and relevant questions. We call this functionality "*fill in the gaps.*"

Our main hypothesis is that any diagnosis process can be modeled as a comparison process between two sets: the one describing the status of the object of diagnosis and the one (if available) containing all known and potential situations (in the related domain), which in fact constitutes the knowledge of the expert (or practitioner) in the considered domain. This is true, for example, for diagnosis in mechanics, since a car mechanic tries to collect a maximal amount of information that describes the current state of an engine and progressively compares this collected set to their own knowledge (which can be considered as a more global set) of the normal functioning of the engine. More specifically, the problem we aim to explore in this paper is how domain ontologies and their characteristics can be used to represent this comparison process and how this provides a way to simulate human reasoning in diagnosis processes.

5 THE REASONING PROCESS

During a diagnosis interview, an expert of a specific domain would first engage in communicating and gathering information freely provided by the person of interest to obtain a description of the current situation. Throughout this early stage of the process, the expert draws all relevant deductions from obtained knowledge without steering the interview. When the expert considers that they have collected enough information, which can constitute a critical mass of knowledge, that can lead them to some relevant conclusions. They begin to guide the interview more actively by asking specific and targeted questions in order to confirm or eliminate each potential conclusions that can, to their knowledge, match the described situation.

In medicine, for example, after collecting this critical mass of information, particularly but not only in terms of symptoms, the main focus of the practitioner becomes to identify a direction to follow in order to identify the disease. This means that in the beginning of the process (except in the very particular and uncommon cases where some very characteristic signs are clearly and visually recognizable), the practitioner has no preconceived idea of the diagnosis they will establish. At this stage, the information gathered through the questions/answers will be most likely partially useful to identify the disease. In fact, after a rather short time, the practitioner identifies one or several "potential diseases." These can be considered as “candidates” to be the final (and normally correct) diagnosis. From here on, a change happens in the practitioner’s attitude since they now have a preliminary idea of the disease described by the patient. They stop passively gathering information from the patient (which can be considered as reasoning “forward”) and start asking targeted questions (similar to “backward chaining”) in order to confirm or infirm elements that may complete the “set” of elements (of knowledge) that were collected previously. This step continues until the practitioner definitely and surely (in his opinion) reaches a coherent set of facts that corresponds to a precise diagnosis.

All this communication scheme between the practitioner and the patient is correlated to a “mental model” of the doctor, which is based on the doctor’s own knowledge, experience, and skills. Hence, if we assume that this mental model of symptomatology can be represented by an ontology of medicine (or any particular domain of expertise), we can also assume that knowledge (concepts) collected from the patient description (even with possible inconsistencies that will later be rejected by the doctor due to their irrelevance to the diagnosis) necessarily constitutes a subset of the whole knowledge of the practitioner. This further leads us to consider that the diagnosis process consists of retrieving and identifying the correct subset among all the practitioner’s knowledge, replicating the exchange process that occurs during medical examination, as represented in Figure 1.

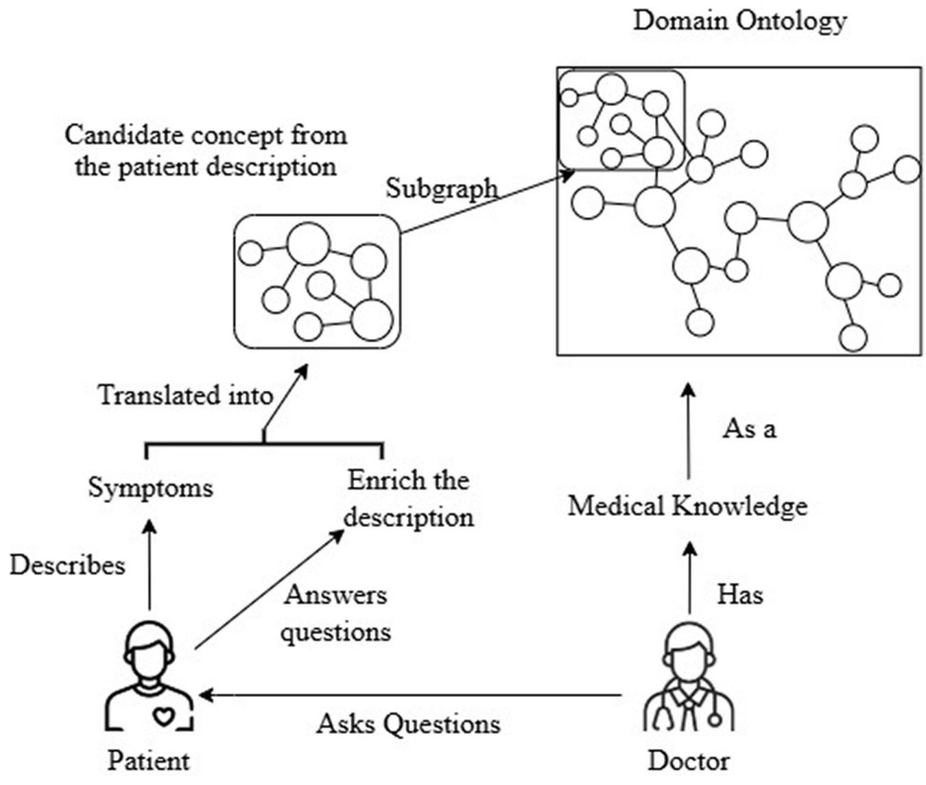

Fig. 1. Medical diagnosis representation using ontologies

6 AN ONTOLOGICAL MODEL FOR MEDICAL DIAGNOSIS

In [17], the authors proposed a meta-model that predefines the set of the necessary information to construct cohesive medical descriptions (see Figure 2).

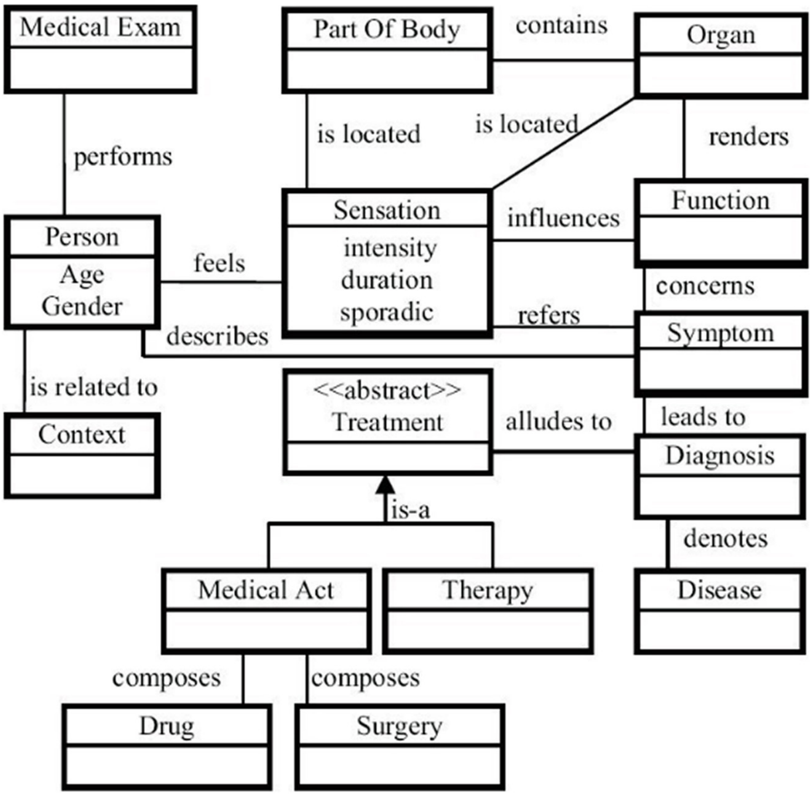

Fig. 2. A meta-model for disease description [18]

This meta-model can be seen as a "meta-ontology" that describes the foundational concepts and relationships that are necessary to validate any instance of a disease, in terms of a specific diagnosis in other words, it aims at representing the concepts that define any disease and that are expected to be mastered by a doctor. If we make an analogy with Figure 1, the following structure would represent the domain ontology of a doctor at its highest level of abstraction, where, for example, using concepts such as <symptoms>, <sensation>, <organ>, <part of body>…, any instance of a disease that is described by any patient should match this "meta-ontology."

The metamodel introduced in [18] defines a structured framework for generating consistent medical descriptions, ensuring accessibility for the care-seekers while addressing the professional standards required by healthcare providers. It is used to extend each symptomatology in terms of intensity, location, etc. As the symptoms may be common to certain conditions, their characteristics (duration, location, intensity, etc.) differ depending on which diagnosis is involved. The pattern of occurrence and likelihood of these symptoms help practitioners to distinguish between different conditions, despite the potential overlap in symptomatology, thus enabling a more accurate and precise diagnosis.

6.1 Applying the model to the expert diagnosis process

To illustrate our methodology, let us consider three well-known diseases in cardiology, each one corresponding to a specific diagnosis: "Dilated cardiomyopathy," "Angina pectoris" and "Heart Attack" (also known as myocardial infarction), as described in Table 1. Each diagnosis is associated with a distinct set of defined symptoms, which may overlap or be shared by other diseases. These symptoms include, for example, chest pain, fatigue, shortness of breath, and palpitations. Each of these symptoms may vary in presentation and intensity across different diagnoses. It is worth noting that at this stage, we only focus on the clinical description of a patient situation, and that we will tackle other information, such as results of medical exams, in a dedicated part of this work.

Table 1. Dataset for reasoning model

Symptoms	Dilated Cardiomyopathy	Angina Pectoris	Heart Attack
Paleness	X	X	X
Weakness	X	X	X
Dizziness	X	X	X
Nausea	X	X	X
Shortness of breath	X	X	X
Abdomen Swelling	X		
Lack of appetite	X		
Sudden weight gain	X		
Persistent cough	X		
Fainting	X		
Palpitations	X		
Sweating		X	
Chest pain		X	X
Crushing Feeling		X	
Vomiting		X	
Arm or shoulder pain			X

Human reasoning generally involves making inferences and drawing conclusions based on available information, experience, and intuition, refined iteratively through trial and error. This flexible process may revisit assumptions when new information challenges prior conclusions and leads to progressively reducing the set of possible diagnoses until reaching its lowest possible size. In medical diagnosis, it starts with gathering initial information to form a baseline. Then, as the expert collects enough knowledge, they narrow down potential diagnoses. The expert then guides the process with precise, targeted questions to refine the diagnosis.

To this **enrichment phase** of available knowledge corresponds a **reduction phase** of the set of possible diagnoses that match the collected knowledge. This continuous enrichment/reduction process can be organized in three levels, each one corresponding to the type of inference that can be operated:

- **Level 01:** A unique symptom or a reduced set of symptoms generally **evokes** one or more *relevant* diagnoses.
- **Level 02:** An *intermediate group of symptoms*, which significantly limits the number of possible diagnoses without necessarily leading to a single one, **suggests** the related set of diagnoses.
- **Level 03:** A *group of conclusive symptoms* **determines** a single diagnosis.

In our process, knowledge is refined by distinguishing the evolution of the collected symptoms, depending on whether they are relevant, suggestive, or conclusive for a concluding set. Figure 3 describes how we model the expert reasoning process.

At (00), the set of possible diagnosis *{SPD}* is initialized to all diagnosis nodes, and the set of described symptoms *{SDS}* is empty (this stage being the starting point of the reasoning process).

In (01), if a single symptom does not suggest a diagnosis (i.e., *Card (SPD) = 0*), it means the symptom (e.g., *S1*) needs to be linked with others such as *S4* and *S8* to trigger a diagnosis. So, we return to step (00) for more symptom readings.

(02) shows how the set of possible diagnoses *{SPD}* is reduced by a symptom *Si* in *{SDS}*. This reduction happens after asking the patient about symptoms in the associated set *{SAS}* to confirm or rule out a diagnosis. If confirmed, the diagnosis stays in *{SPD}*, and the symptom is added to the patient's description *{SDS}*. If not, the symptom is removed, and the diagnosis path is eliminated.

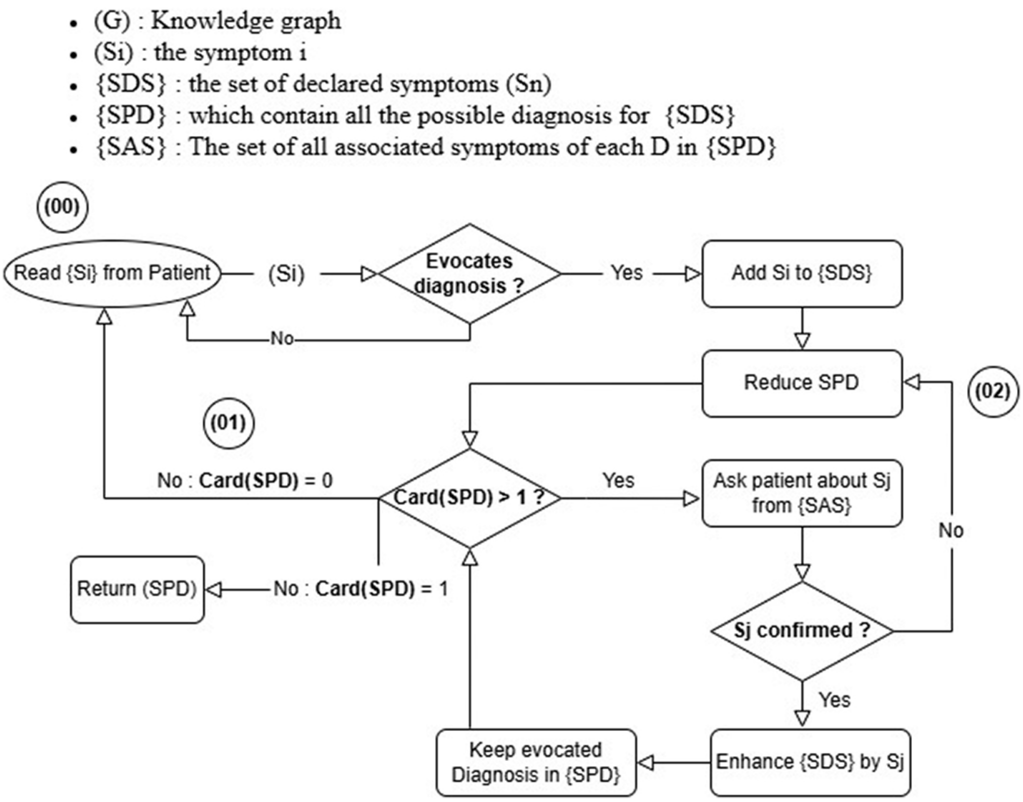

Fig. 3. A model for the expert reasoning process

For more clarity and to reduce the complexity of the model, we focus at this stage on the data corresponding to symptoms, diagnoses, organs, and body parts, without considering intensity or duration, for example. When we map all the above datasets onto the metamodel described in Figure 2, we get the instantiation represented in Figure 4.

As an example, applying this process to the dataset presented in Figure 4, which is a subgraph of a domain-ontology that contains knowledge related to cardiology, results in the following graphs, derived from the metamodel (see Figure 4) and which represent, respectively, each level of the progressive knowledge acquisition (see Figure 8), all dynamically interconnected in one knowledge graph (see Figures 5, 6, and 7).

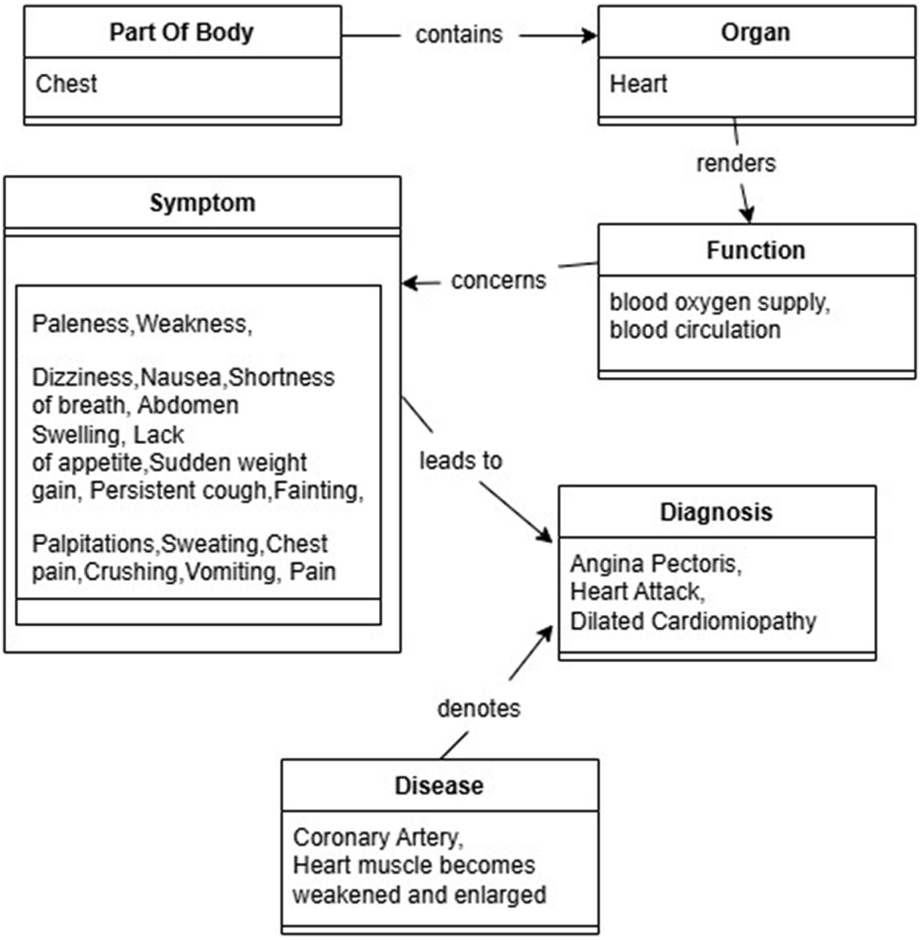

Fig. 4. Meta-model instantiation on dataset

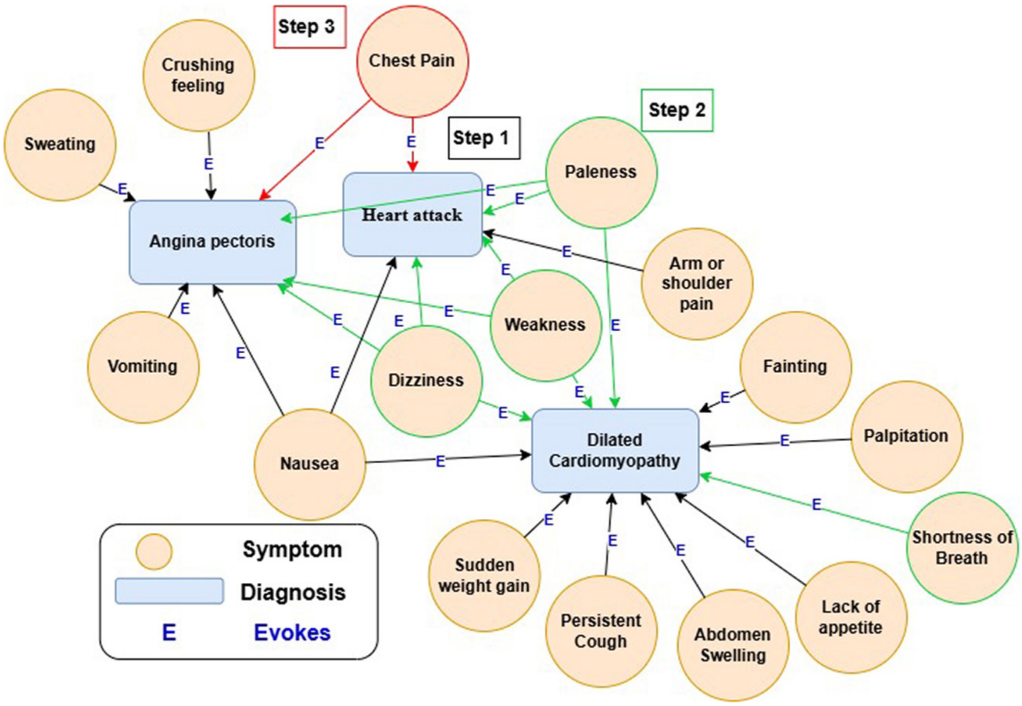

Fig. 5. Knowledge graph level 01

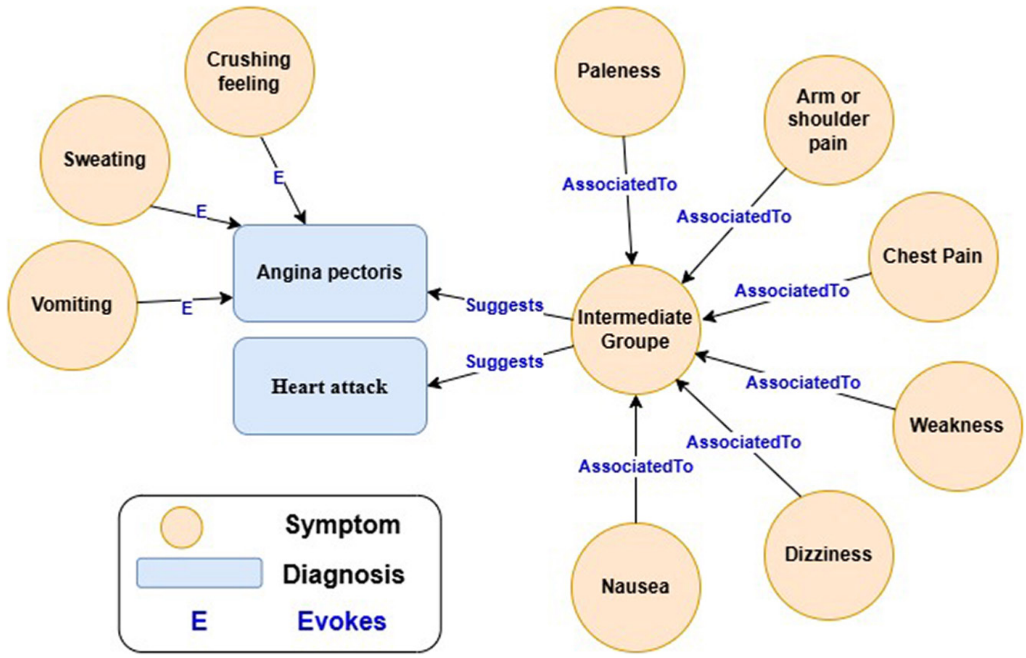

Fig. 6. Knowledge graph level 02

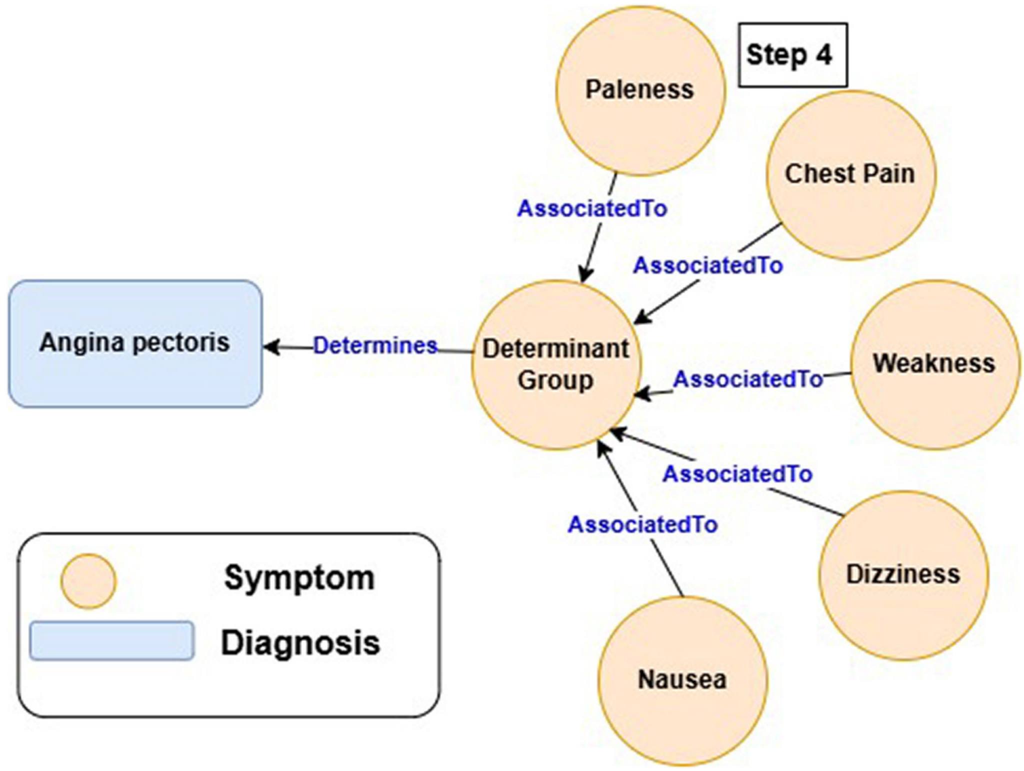

Fig. 7. Knowledge graph level 03

Level 01: SAS = {All symptoms in (G)}
SDS = {Ø}
SPD = {All diagnoses in (G)}
Level 02: ***(? Symptoms)*** *[Successive acquisition (green nodes and edges)]*
SAS = {All associated symptoms to each SPD}
SDS = {Weakness, Paleness, Dizziness, Nausea, Shortness of breath}
{SDS} ***evokes*** *SPD*
SPD = {Dilated Cardiomyopathy, Angina Pectoris, Heart Attack}
Card(SPD) = 3 > 1 → Continue
Level 03: ***(?/Chest Pain)*** *[Key symptom verified to refine the diagnosis].*
Yes/Chest Pain:
{SAS} = {SAS} - {Chest Pain}
{SDS}={SDS} U {Chest Pain}
SDS} ***suggests*** *SPD*
SPD = {Heart Attack, Angina Pectoris}
Card (SPD) = 2 > 1 → Continue
Level 04: ***(?/Arm or Shoulder Pain)***
(No/Shoulder pain):
SAS=SAS - {Arm or Shoulder Pain}
No modification on SDS
{SDS} ***determines*** *Angina Pectoris*
SPD = {Angina Pectoris}
Card (SPD) = 1 → STOP

Fig. 8. Applying the process to a real case

7 IMPLEMENTATION

Our model has been implemented using ontology editor Protégé [19] in its version 5.5.0. The meta-model instantiation with the specific knowledge corresponding to the three considered diseases (refer to Table 1) is represented in Figure 9. The OntoGraf Protégé plugin gives support for interactively navigating the relationships of our OWL2 [20] ontology. It is worth mentioning that this example is given only to illustrate our approach. Therefore, and for clarity reasons, only a subset of the mentioned concepts (namely symptoms) is represented here. Moreover, Figure 9 highlights the relationship between the provided meta-model (see Figure 2) and the proposed symptomatology (refer to Table 1).

We have developed a client-server application where the server-side is an API implemented with ASP MVC 8 [21]. We used the dotNetRDF [22] library to handle SPARQL [23] queries over our ontology. In the instantiated graph, we use SPARQL queries to search for the symptom pattern (a combination of inputs) that leads to a potential diagnosis, and we display the results of our queries using the GoJS Library [24] in a graph form on the client-side.

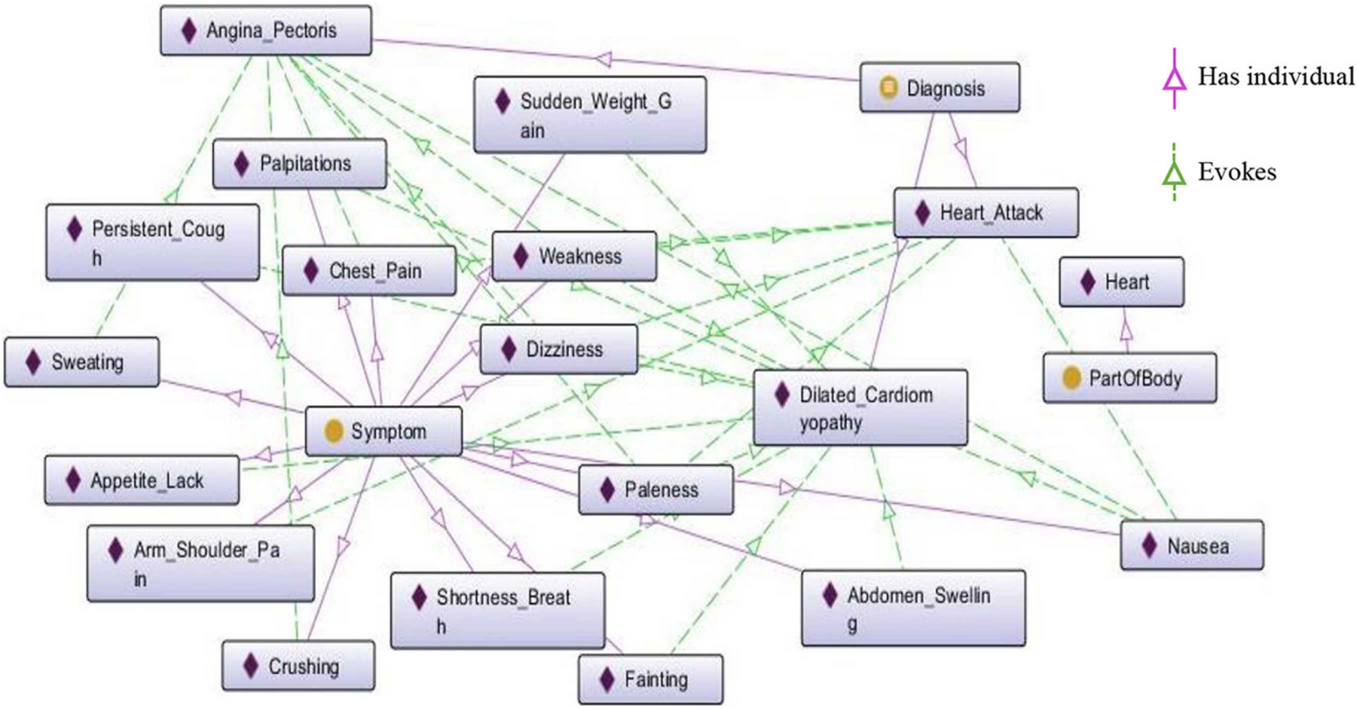

Fig. 9. A part of the OntoGraf representation of the global ontology

This implementation consists of two primary components: the Inference Module and the Knowledge Module. The Knowledge Module is responsible for storing the ontology and providing APIs access for data retrieval and updates. The inference module, utilizing embedded reasoners, provides reasoning techniques to the knowledge base in order to derive conclusions. This solution will be made accessible to users who wish to integrate it into their own systems (on publicly accessible software repositories) and will have a double benefit: (1) the users will be able to leverage our enablers in their systems (the inference module in any diagnosis domain and the knowledge module in cardiology), and (2) it will allow us to continuously gather information and feedback, thus enabling continuous improvement of both the inference and knowledge modules.

8 CONCLUSION AND FUTURE WORK

In this paper, we proposed an ontology-based approach that simulates the human reasoning process in relation to diagnosis. We then applied it to the medical field of cardiology, illustrating how we can represent knowledge to operate artificial reasoning, and how such models can be applied to reproduce human-performed diagnoses. After a review of related works, we introduced our ontological model and described how it can be used to support a communication scheme in any domain of expertise. We then described how this model can be used to formalize a diagnosis process. We applied it to the specific domain of cardiology disease diagnosis with a first set of test cases that illustrated its validity through significant and highly promising results.

As a second stage, we are currently examining the behavior of our model on a significant number of real cases, provided by our partner cardiologist, to further validate our methodology. In parallel, we are investigating the use of a Large

Language Model (LLM) [25] [26] to facilitate patient interactions. This LLM will be trained to convert patients' descriptions of symptoms in natural language into expert insights, which will then be incorporated to guide the diagnosis process, leading to more accurate conclusions. Additionally, the LLM will be integrated with authoritative sources of knowledge such as SNOMED-CT [27], enriching patient descriptions with standardized terminology. This approach will improve the ontology's functionality by training the model to answer natural language queries based on the ontology's structure and to propose potential improvements, such as adding new classes, properties, or annotations, while maintaining alignment with established clinical standards.

It is important to emphasize that our primary goal is not to create a tool for automated diagnosis but rather to develop a model that enables the representation of expert knowledge and reasoning mechanisms using ontology-based frameworks. Our ultimate goal is to combine ontologies and LLMs into an integrated solution, where the domain ontology supports artificial reasoning, while the LLM facilitates continuous enrichment of the ontology and enables natural language interaction.

9 ACKNOWLEDGMENT

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