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AsthmaONTO: An Ontology System for Diagnosing asthma disease

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ABSTRACT

Asthma is a common chronic inflammatory disease of the airways in the lungs. According to World Health Organization Asthma is often not properly diagnosed or treated, especially in low-income and lower-middle-income countries. People with untreated asthma can experience disturbed sleep, daytime fatigue, and poor concentration. Detecting the disease at the right time can help to reduce the risk it may cause. Using Semantic Web technologies, this project aims to improve asthma disease diagnosis. We created a domain ontology (AsthmaONTO) that encompasses asthma disease domain knowledge. The ontology includes terminology, relationships, and properties that can be used in the diagnosis of asthma. To diagnose asthma disease and predict the risk of asthma, a set of rules has been developed based on valid links between ontology concepts. It also offers therapy options and recommendations. A domain expert submitted a sample set of individuals with asthma disease, which was used to evaluate the proposed system. The system correctly diagnosed 48 of the 54 cases patients with asthma (ratio of correctness is 88.8%), according to the findings.

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1. Introduction

Combating human illness, as well as promoting and maintaining health, are critical challenges for the human species' survival. Spite of the progress in appropriate treatment and control purposes, medicine remains powerless to combat a lot of illnesses that claim the lives of millions of people [1،2]According to world Health Organization (WHO) documents, chronic diseases claim the lives of 12.6 million people each year [3].Asthma is a chronic pulmonary disease. which has an impact on the lungs (the breathing tubes that carry air into our lungs). Because the airways in their lungs narrow, this can make breathing difficult, resulting in coughing, whistling (wheezing), and other symptoms. Around 300 million of people all over the world asthmatic sufferer, which affects people of all ages and ethnic backgrounds [4], and has claimed the lives of 461,000 people.[5] By 2025, it is estimated that another 100 million people will have asthma [6]. As a result, asthma has a significant socioeconomic impact on both adults and children of Mortality, burden of disease, life quality, and medical costs [7]. The goal of optimal asthma management, according to some experts, should be to achieve and maintain disease control [8–9].

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All current asthma guidelines emphasize the importance of asthma control, which should be measured using a variety of criteria End-points such as diagnostic, functioning, and inflammatory outcome, and also patient-reported results, are included (PROs). One of the options suggested for dealing with a big amounts of data is ontology. Domain definitions with their related relationships are incorporated into ontology as a shared knowledge [10]. Ontology provides a shared and common structure of information between humans and machines, removing ambiguities in words and allowing domain knowledge to be reused. It was originally characterized as an explicit formal statement of the terms in the domain and their relationships [11] .Semantic web reasoning can be defined as a technique for automatically generating fresh associations based on experience expressed as a collection of resource relationships as well as some extra info in the form of a list of rules [12].

The importance of ontologies can be summed up as follows: [13]

• Ontological analysis elucidates knowledge structure.
• Knowledge cannot be represented by a vocabulary. without ontologies that underpin it.
• The ontology is a kind of theoretical model that can be utilized in a variety of uses and has a high likelihood of reusing and interoperability.
• Common vocabularies (for humans and agents).
• A common understanding as to how data is organized

It has been used in a variety of fields, including the semantic web, biology, and medicine, to name a few. Its popularity stems from its capacity to simplify problems. The current paper focuses on ontology as a computational aid for clinical problems in the medical domain. Medical ontologies are used to solve problems in the medical and biomedical domains, such as disease diagnosis and treatment. [14] understanding of the structure of data.

We discovered that an ontology-based method could be a superior option for asthma diagnosis after conducting a thorough literature review and empirical testing. An ontology-based method to asthma diagnosis is presented in this research.

2. Related Work

There are many works that have used the Semantic Web in diagnostics, using different methods and techniques

(Al-Hamadani & Alwan, 2015) [15] Describes an expert structure for detecting coronary artery disease. The system's designing is based on ontological knowledge about the patient's condition. symptoms in order to expand the information base The SWRL instructions are used to determine the appropriate medicine and the patient's required procedure This system's architecture is consists of the following modules: The fact base module is part of the knowledge base module.in addition to the rule base, inference engine, and explanation modules. The truth is retrieved from the operator as the patient's indicators utilizing the user interface to make the appropriate choice, the inference engine relies on facts and rules. The user will be informed of the final choice findings via the user interface.in addition to the inference regarding this decision from the explanation module. Several general practitioners examined this method utilizing 16 different scenarios. Validation and evaluation of the system should be tested. The recalling and accuracy ratios were computed 0.83 and 0.87 were calculated.

research fixates on identifying the coronary artery illnesses, which are a type of heart disease of cardiac problems This system's diagnosis is solely dependent on the patient's indicators. without the use of into description the results of diagnostic test (ECG, X-ray, and CT scan).

(Rawte & Roy, 2015) [16] describes an expert system based on ontology for thyroid illness diagnosis. This system used ontology to build field knowledge and guidelines to infer thyroid illness diagnoses. The ontology, reasoner, rule base, and MySQL database are all part of it. Data and SWRL rules are created for users according to the symptoms supplied via the user interface. Jena, a reasoner, then infers and makes the diagnosis based on the data and criteria. Ontology and neural networks were used to build this system. A total of 60 examples were taken. The neural network is trained using only TSH, T3, and T4 values in the blood, but the ontology-based expert system receives no training. The findings reveal that an expert system based on ontology produces better genuine outcomes with lesser complication than a neural network expert system.

In terms of employing ontology to represent knowledge, this system is in accordance with our study. It is assessed by comparing its neural network-based implementation against an ontology-based implementation. A sample set of patients with asthma illness given by a domain expert is used to test our approach.

(Alharbi, Berri, & El-Masri, 2015) [17] shows a diabetes identification and therapy suggestion system. The system that was implemented is made up of two parts: a domain ontology that was created to create a knowledge base system and standardize domain expertise that contains the essential rules for diagnosing and treating diseases. OWLDL created and developed the domain ontology, SWRL created the rules, and the JESS inference engine executed them. The user interface, the inference engine, the knowledge base, and the ontologies are all components of the system. The graphical user interface allows the user to trial the system or obtain a diagnosis for a specific situation. The inference engine is a reasoning element that usage the knowledge base's ontologies and instructions to understand a diagnosis for a specific case.

(Thirugnanam, 2013) [18] creates a disease information system using an ontology-based method This system is made up of three basic components: a knowledge base, rules, and query processing. The ontology for modeling diseases and their interactions with symptoms is part of the knowledge base. The semantic reasoner and SWRL Rules are both part of the Rules component. The query processor looks for disease relationships using SWRL rules. It returns diseases that are linked to the specified symptoms. The query processor then gives the user the results. Executing rules retrieve disease facts with indications depending on the provided principles, and the resulting axioms are subsequently represented in the ontology.

In terms of creating ontology, this technique is identical to ours. However, it differs in terms of illness types and diagnostic processes.

(Almutair & El-Masri 2014) [19] proposed a General understanding-based Ontology method for diagnostic testing based on clinical experience standards (CPG) to assist Doctors and medical personnel professionals' personnel make the optimal diagnostic decision for the patients. This framework is a generic foundation that may be customized to quickly construct a specific clinical practice guideline. The process is divided into four stages. The first stage in this Procedure is to select a related clinical practice recommendations resource to serve as the foundation for this study. The second stage, and the study topic, involved selecting 30 diseases from various human organs and visualizing them in tables built on their signs and symptoms, indicators, in addition to diagnosis techniques. The third stage is to capture and model the typical indicators and indications of these 30 illnesses, and the patient will be effectively diagnosed using the differential diagnosis that will be provided at the conclusion. Using Protégé, the fourth and last phase in this process is to convert these models were included in to knowledge base ontology architecture for Patient Analysis based on the Clinical training recommendations.

This technique may be thought of as a basic knowledge base for a variety of diseases; however, it lacks further information regarding the specifics of each disease in terms of laboratory tests and risk factors, especially for diseases that overlap symptoms and diagnostic procedures.

(Runumi Devi & Deepti Mehrotra , 2020)[20] The purpose of this work is to put forth a semantic rule-based modeling and reasoning strategy aimed at formalizing the description of dengue disease in conjunction with operational definitions (semantics) that aid clinical and diagnostic reasoning. Semantic Web Rule Language (SWRL) logical rules are used to include the operational definitions, enhancing the knowledge base's expressive power. For the purpose of linking dengue fever with an ICD code, a dengue knowledge base has been created and expanded with the International Classification of Diseases (ICD) ontology. Along with providing interoperability, the knowledge base may be used to justify diagnostic categorization that can identify dengue symptoms and foretell the likelihood that patients would contract the illness. 153 actual patient cases are successfully categorized using the SWRL rules' operational definitions.

(Kouamé & Mcheick , 2021)[21] suggest methods based on adaptive mechanisms for identifying COVID-19 symptoms To better offer COPD patients with the right care, .SuspectedCOPDcoviDOlogy, an ontological model made up of five ontologies for identifying suspicious instances, has been developed in order to accomplish this aim. These ontologies make use of alarms, service management, vital sign parameters, and symptom parameters. The COPDology put out by a prior study in the COPD field is improved by suspectedCOPDcoviDOlogy. An experimental research comparing the outcomes of an existing test for the detection of COVID-19 with those of the suggested detection system is carried out to validate the solution.Lastly, based on these findings, we draw the conclusion that a strict combination of detection rules based on vital sign and symptom data can significantly increase the dynamic detection rate of COPD patients suspected of having COVID-19, enabling prompt medical attention.

Of the above works, none of the works dealt with the diagnosis of asthma using ontology. As far as we know, ontology has not been used to diagnose asthma during previous years. Therefore, our work focuses on diagnosing asthma using ontology. Our system diagnoses asthma based on a patient's symptoms, risk factors, and results of diagnostic tests. Other works cited have focused on different types of diseases, but in this paper we focus on asthma.

3. AsthmaONTO METHODOLOGY

An Ontology is a machine-readable characterization of the concepts of a domain as well as the interrelations between the domain's definitions. As the Internet grew in popularity, so did the demand for networked knowledge to be shared and reused. Interoperability, sharing, and reuse are made easier using ontologies, which are designed to suit these objectives. They do so by refocusing systems enterprise away from technology and toward a solution that works on both a knowledge and technological level. This is a critical step that must be completed in order to fully utilize the electronic knowledge that is available. In 2015, there was a Universal Conference on Emerging Technologies in Engineering. (ICNTE-2015) The Semantic Web is the future of the Web, in which information is given unambiguous significance, allowing machines to automatically analyze and incorporate information available on the Web. OWL promotes more machine interpretability than XML by imparting meaning to Web contents.

Several approaches for constructing and building ontology-based expert systems have been offered. METHONOLOGY is one of these approaches, and it is thought to be the most thorough ontology engineering system [22], [23], [24]. METHONOLOGY, as shown in Fig 1 (derived from [25], [23]), not only specifies the stages through which the development method should proceed, but it also depicts the depth of certain of these stages. Although this system appears to have numerous discrete stages, the majority of them interact with one another and can be used at the same time.

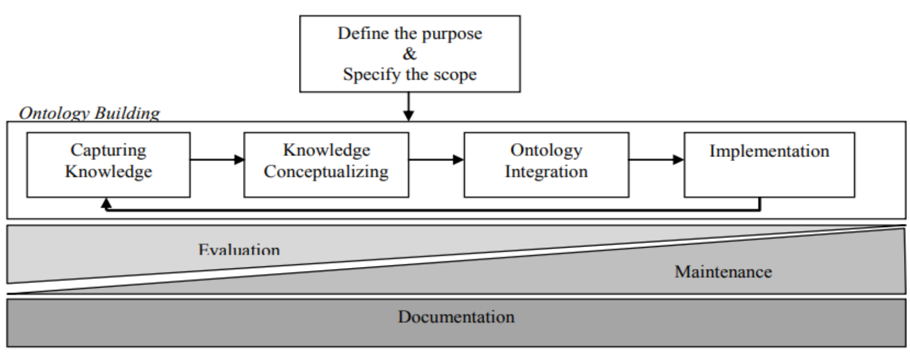


Figure 1: Ontology development life-cycle.

Fig 1 : Ontology development life-cycle

This methodology is used by (AsthmaONTO), and the steps of the development process are as follows:

1. Define the purpose and specify the scope stage: This stage should determine the purpose of building the ontology, its formality, and its scope. The purpose of (AsthmaONTO) is to be a knowledge representation of asthma disease

domain embedded in an easy to use GUI. According to [26] the degree of formality of the ontology depends on the amount of formal knowledge against the natural language knowledge present in that ontology. Since (AsthmaONTO) was expressed in a formally defined language (OWL), its degree of formality is "semiformal". The proposed system can be used by the specialist in the field or normal user in order to get an accurate diagnosis based on the reasoning process embedded in the ontology.

2. Capturing knowledge stage: The depth of this stage starts to be high in the beginning of the design process and it decreases as the process goes farther. Several techniques were used in capturing the knowledge required to build (AsthmaONTO). Nanostructured interviews with the experts in the field were useful to specify the requirement of these experts, while structured interview was used to get the detailed knowledge and to specify the concepts, object properties, data properties and the relations between them. On the other hand, formal and informal text analyses were used in this stage to identify the main structure of the ontology and to fill the concepts in this structure respectively.

3. Knowledge conceptualization stage: This stage depends on the output of the previous stages by constructing the Glossary of Terms (GT) [23] which include the concepts and the properties used in designing the ontology. These terms are useful to construct the class hierarchy and to determine the properties and the relations between these classes. First the conceptualization table should be formed. Table 1 shows sample of this table. Next, this table should be transferred into a conceptualization tree. For this purpose, we use the rich semantic tree (RST) suggested by [27]. Fig2 shows the (AsthmaONTO) RST tree and the key shapes used in it.

4. Integration stage: This stage specifies if the developed ontology can have benefit from other existing ontologies. This process exemplifies the feature of ontology reusability, when the existing ones can be integrated in to a new developing one.

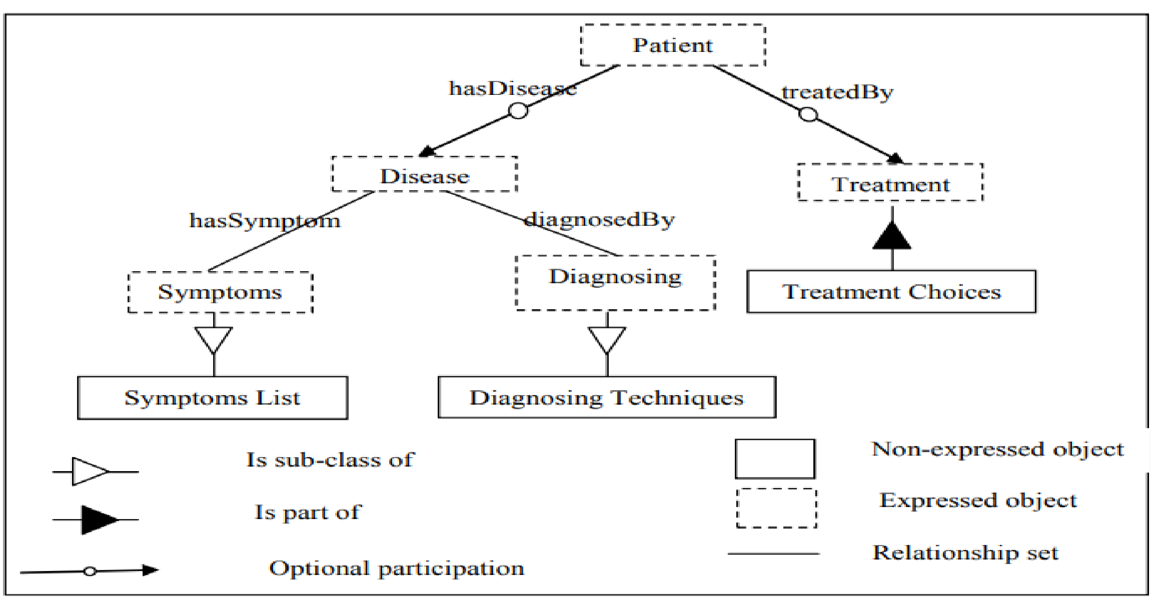


Fig 2: AsthmaONTO RST

5. Implementation Stage: This stage is responsible on implementing the ontology in one of the available ontology editors. (AsthmaONTO) used Protégé 4.0 as an editor which has the ability to check the lexical and syntax errors. Protégé is integrated with several types of reasoners which guarantee the completeness, consistency, and non-redundancy in the defined ontology. The reasoner used by the developed ontology is Pellet [28]. Fig 3 shows sample the tree structure built by using OWL Viz plug-in embedded with Protégé for (AsthmaONTO) ontology. Protégé provides the abilities to create the ideas and their data properties and entity properties in sensitive explanation logic [29]. Fig 4 show the object properties in Protégé. Fig 5 presents the Data properties in Protégé.

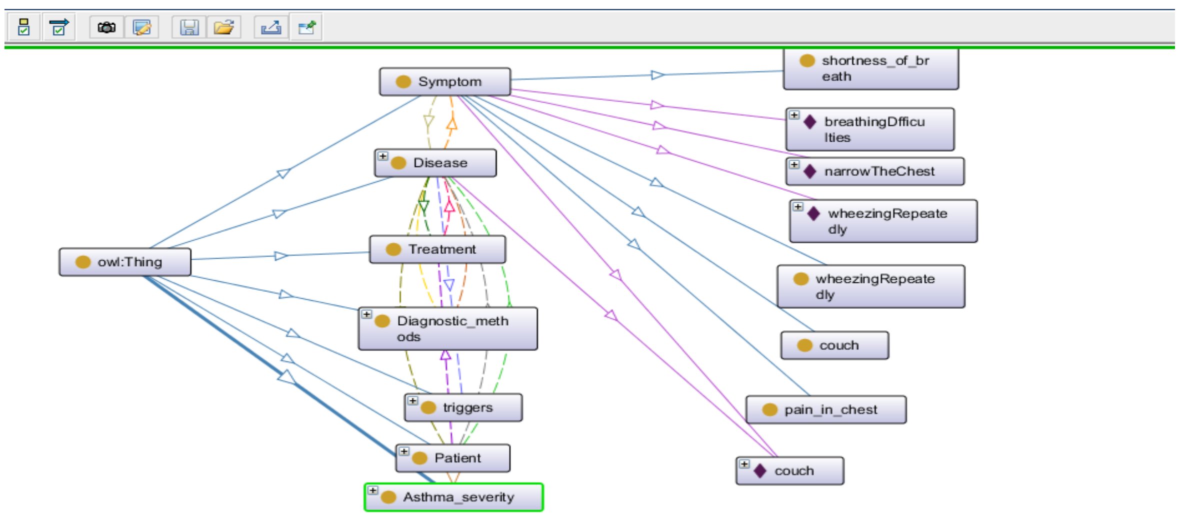


Fig 3: AsthmaONTO Tree Structure Produced By Protégé

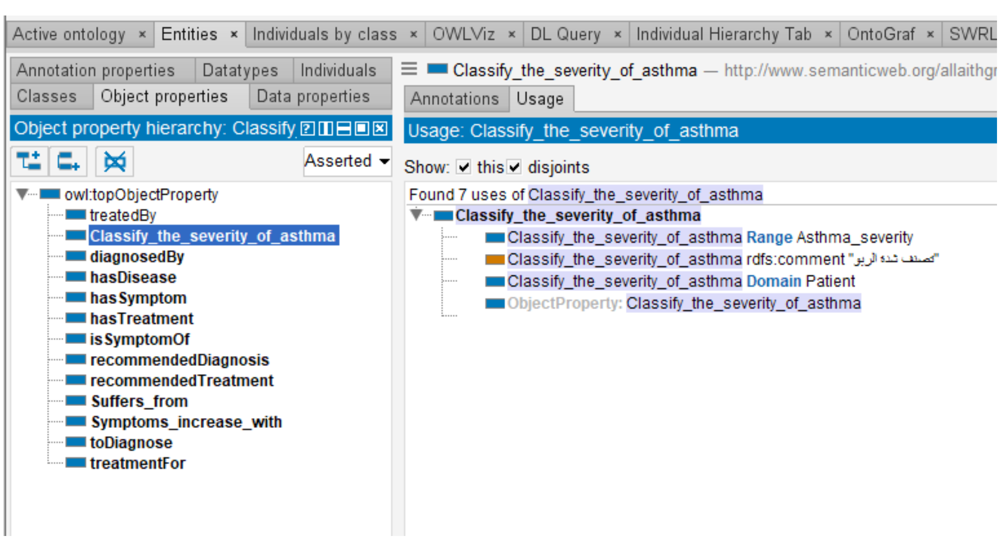


Fig 4: AsthmaONTO Object Properties in Protégé

6. Evaluation: It is not a standalone stage but it is the one that interfere with all the previous stages. As seen in Error! Reference source not found., the evaluation stage depth starts to be high at the first stages of the development process and then it decreases as the process goes farther. In the first stages the important thing is to guarantee the verification of the developed ontology which is done by checking the correctness of the captured knowledge and the conceptualizing table and its tree using interviews with the experts in the domain. While in the later stages, the validation of (AsthmaONTO) has been checked by professionals to ensure that the created system fits their needs

7. Documentation: this stage is going in parallel with all other stages since the output of each stage is a document which is important to available as an input for the next stage. Moreover, publishing papers in journals or conferences in considered being a kind of documentation.

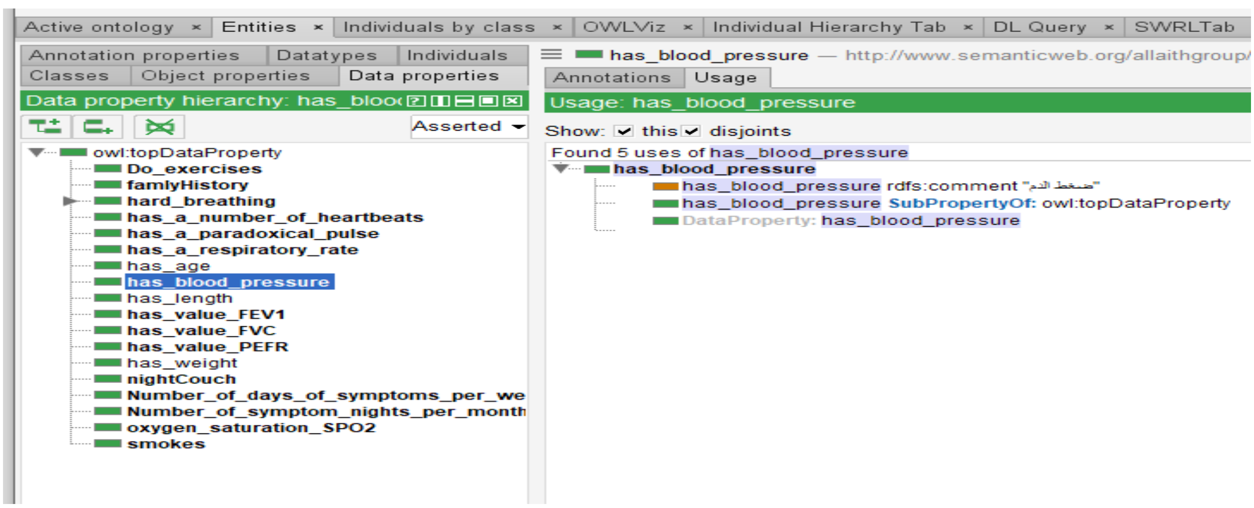
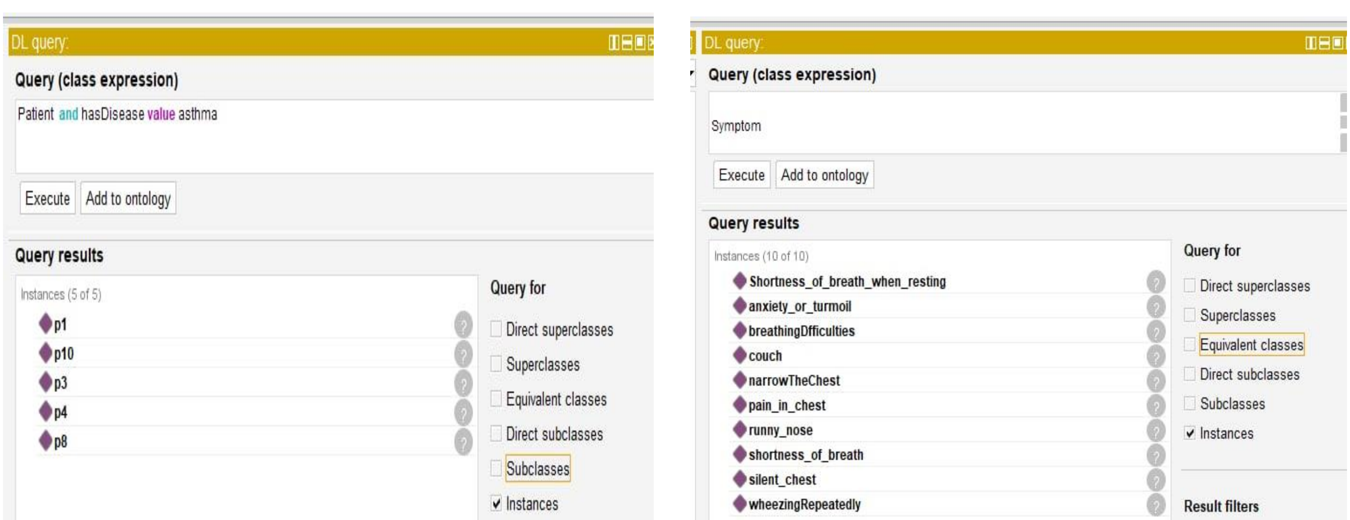
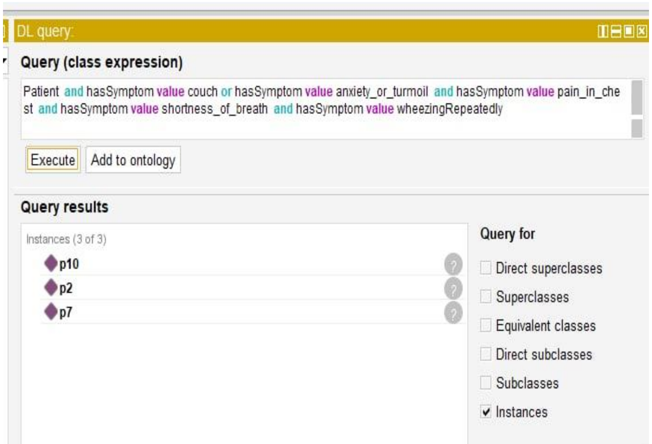


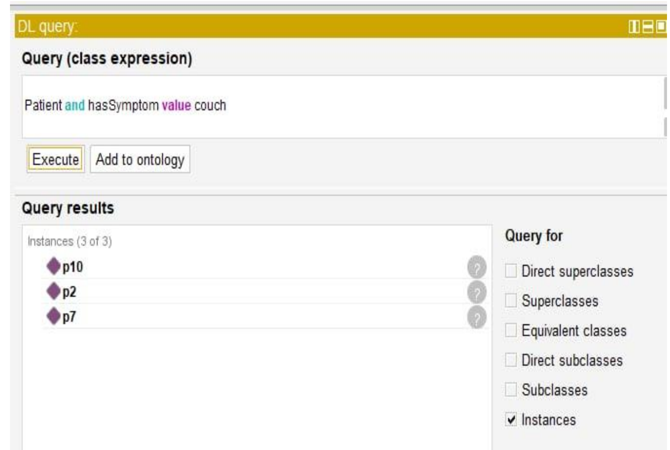
Fig 5: AsthmaONTO Data properties in Protégé



(a) **Fig 6: Show DL Query in the protégé** (b)

a- Answers the following query: Who are the patients with asthma?
b- Answers the following query: What are the symptoms of Asthma?





(a) **Fig 7: Show DL Query in the protégé** (b)

- c- **Answers the following query: Who are the patients who have the following symptoms: Coughing, anxiety or trouble, shortness of breath, chest pain, and wheezing??**
- d- **Answers the following query: Who are the patients who have cough symptoms?**

4. Discussion

Technical assessment and user evaluation are the two phases in the evaluation of an expert system. Technical evaluation should undergo verification and validation, as was indicated in the sections before this one. All of the knowledge gathered should be guaranteed to be accurate during the verification stage, and the designed system should be guaranteed to function properly. A number of methods were employed to examine the validity of an expert system One of these methods is to use the DL Query as shown in Fig 6 and Fig 7, validation based on case studies was by far the most popular one . To verify the system's validation, it is essential to choose test instances. These cases ought to include every diagnostic that is currently conceivable for a particular instance. To test (AsthmaONTO)there were 54 test cases that had previously been diagnosed as asthma patients Each test case has input parameters that include general patient information, symptoms, and diagnostic test results. Out of the 48 cases, our method gave the same diagnosis, indicating that the devised method properly detected 88.8% percent of asthma patients. However, because the asthma domain is a difficult area, this study gave a viable strategy for diagnosing asthma with few resources, according to the domain expert. the proposed technique's produced prototype is useable, and the accuracy of the (AsthmaONTO) Ontology-based approach It was an appropriate accuracy makes it work as a domain expert in the asthma diagnosis sector. Based on the research findings and the ontology approach, the created method could be a critical component in asthma diagnosis.

5-Conclusion and future work

An ontology-based approach for asthma diagnosis has been developed in this research study, It is founded on a formal ontological definition of a health-related topic. All essential concepts connected to asthma disease are included in the proposed ontology, including the patient's personal information, activity, symptoms, risk factors, laboratory examination findings, treatment plan, and appropriate recommendations. The knowledge base and approach are the two fundamental modules of the (AsthmaONTO)Ontology system. A test dataset of 54 cases for a group of persons who had previously been diagnosed was used to evaluate an ontology-based diagnosis approach. The findings revealed that 48 of the 54 patients had a diagnosis that matched. We met our research objectives at the end of the study, and the recommended approach is right and provides appropriate accuracy.In the future, Making X-rays a part of the tests for diagnosing asthma to be more accurate in the diagnosis Expand the number of patients to get a more comprehensive and accurate of the system. Finally, we want to create system mobile app versions.

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