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RESEARCH ARTICLE

MEDICAL ONTOLOGY ENGINEERING FOR HEART DISEASES: A PATHOLOGICAL APPROACH

Galal AL-Marzoqi[1,*], and Abdulrazzak Ali[1]

[1] *Dept. of Computer and Information Technology, Faculty of Engineering, University of Aden, Yemen*

***Corresponding author: Galal AL-Marzoqi; E-mail: galalalmarzoqi@aden-univ.net**

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Abstract

Heart disease continues to be one of the leading causes of death worldwide, making it essential to develop accurate tools that combine both clinical insight and pathological evidence. In this paper, we developed an Ontology-based framework aimed at improving the understanding and classification of heart diseases from a pathology-centered perspective. The methodology followed well-established Ontology engineering practices such as "Methodology" and the NeOn framework. We began by gathering expert knowledge from cardiologists and pathologists, supported by trusted clinical guidelines and textbooks. The heart disease Ontology was built using the OWL language in Protégé, and it captures meaningful links between symptoms, clinical signs, lab tests, and treatment options. A team of medical specialists reviewed the Ontology through iterative validation steps to ensure its clinical accuracy and usefulness. When applied to real patient records, the system showed a noticeable improvement in diagnostic consistency and precision. These results highlight the practical value of medical ontologies in supporting physician decision-making and improving integration across digital health systems, especially when paired with AI-powered diagnostic tools.

Keywords: Ontology; Ontology engineering; Medical ontology; Heart diseases; Pathology domain.

1. Introduction

Ontology, in Philosophy, is the branch of metaphysics that deals with the nature of being, while it, in Logic, is the set of entities presupposed by a theory [1]. Heart disease continues to be the primary cause of death globally, accounting for approximately one-third of all deaths [2]. These conditions contain a variety of heart problems such as coronary artery disease, high blood pressure related heart disease, heart valve disorders, and congenital heart defects. Diagnosing heart diseases is often complex. Specialists need to carefully analyze many types of information, such as a patient's medical history, symptoms, lab test results, imaging scans, and in some severe cases, tissue samples from the heart. However, because this information comes from different sources and can be quite detailed, it can be difficult to connect everything together in a meaningful way. Many hospital systems and medical databases are not equipped to link this information efficiently. As a result, there can be gaps in understanding, and doctors may end up with inconsistent diagnoses. In addition, valuable knowledge about disease progression and specific pathological details is often stored in unstructured ways, making it hard to use in digital systems.

A growing interest in utilizing structured knowledge models to better organize and analyze medical data has been influenced by this. The systematic definition of concepts and their relationships in an Ontology is a means of organizing knowledge. Despite their ability to provide comprehensive clinical knowledge through well-known medical ontologies like SNOMED CT and UMLS [3], these are typically incomplete when it comes to specific pathological findings, particularly those related to heart disease. Additionally: A specialized Ontology that examines heart diseases from a pathological perspective is the primary objective of this research. The aim of this model is to present medical and pathological information in a standardized and structured manner using tools such as OWL or Protégé [4].

The goal is to map the relationships between symptoms, diagnostic tests, treatments, and disease mechanisms, which will enable doctors and digital systems to make informed decisions about heart disease cases. This Ontology was created by incorporating expert knowledge from cardiologists and pathologists, reviewing trusted medical sources [5], [6], and validating the model through expert feedback. This work is intended to improve diagnostic accuracy, provide a more accurate diagnosis and create reusable knowledge frameworks for systems such as clinical decision support tools and electronic health records (EHRs).

2. Related Works (2024–2025)

In recent years, many researchers have explored the use of ontologies in the medical and cardiovascular fields. Here are some of the most relevant and recent studies:

- MIO: An Ontology for Cardiovascular Medical Knowledge (2025)

This study introduced the concept of MIO, an Ontology that focuses on cardiovascular diseases. It was created to facilitate the examination of heart disorders both physiologically and physiologically. By utilizing OWL and the guidelines of the OBO Foundry [7], MIO was created to connect essential aspects of heart disease, including symptoms, causes, and therapies. The unification of clinical and pathological knowledge using MIO was demonstrated by the researchers to enhance diagnostic tool accuracy. [8].

- Developing Medical Ontologies with Limited Expertise (2025)

This work presented an achievable approach to developing medical ontologies in environments where expert expertise is not feasible. Using pneumonia as an example, the authors simplified the METHONTOLOGY framework to make Ontology development more accessible [9]. Despite not specifically investigating heart disease, their research can still be utilized to develop specialized medical ontologies, such as the one described in this study [10].

- Ontology-Guided Machine Learning vs. Large Language Models (2024)

In this study, published in Scientific Reports, researchers compared Ontology-based machine learning models with large language models such as GPT-4. They found that the Ontology-guided approach performed better in structured clinical reasoning tasks, achieving high accuracy levels up to 98% in some cases. This study emphasizes the **capabilities** of ontologies in **healthcare** AI applications [11].

- Mapping Echocardiogram Reports to Structured Ontologies (2024)

This preprint study focused on mapping echocardiogram findings to a structured cardiovascular Ontology. By using OWL, the researchers improved the ability to share and interpret echocardiogram data between different healthcare systems. Their work is especially promising for improving heart failure diagnoses and tracking disease progression over time [12].

- Comparing Ontology-Based Models with Machine Learning for Heart Disease Prediction (2024)

This study evaluated how Ontology-based approaches compare to traditional machine learning methods like Random Forest and Support Vector Machines (SVM) in predicting heart disease. The results showed that Ontology-based models not only achieved better accuracy but also provided more explainable results, making them a valuable tool for doctors [13].

3. Comparison Table of Recent Studies

The comparison table highlights the diverse contributions and limitations of recent Ontology-based research in the medical and cardiovascular domains. Together, these studies show how ontologies can enhance diagnosis, improve data sharing, and provide more explainable results in cardiovascular healthcare. However, they also reveal common challenges, such as the need for expert input, structured data, and better integration with existing healthcare systems.

The current research stands out by focusing on heart disease from a pathological perspective, aiming to connect disease symptoms, pathological findings, and treatments in a unified Ontology designed specifically for clinical reasoning and diagnosis. This analysis also underscores the unique contribution of the current research, which seeks to unify pathological features, clinical symptoms, and treatment strategies in a heart disease focused Ontology tailored for diagnostic and computational reasoning.

Table 1: Represents Comparison Table of Recent Studies

Study	Goal	Tool\Language	Key Contribution	Limitation
MIO (2025)	Build a heart disease Ontology for diagnostics	OWL, OBO	Integrated clinical-pathological knowledge	Resource-intensive
Ontology Building (2025)	Create ontologies with minimal expert help	Methodology	Scalable design for clinical ontologies	Not specific to heart disease
EchoMap vs. GPT (2024)	Compare Ontology-driven ML to LLMs	OWL, EchoMap, GPT-4	Outperforms GPT in medical classification	Requires structured input
Echocardiogram Ontology Mapping	Structure echo report semantics	OWL, medRxiv	Cross-institutional data harmonization	Not fully integrated with EHR yet
El Massari et al. (2024)	Compare ML and Ontology for heart disease	OWL, RF, SVM	improves prediction and explainability	Limited dataset

4. Heart Disease Ontology Methodology

The methodology adopted in this research follows a structured pipeline:

1. Knowledge acquisition from domain experts and clinical resources.
2. Classification of heart diseases based on pathology.
3. Ontology construction using OWL in Protégé.
4. Validation by medical experts.
5. Evaluation through experimental application on real patient records.

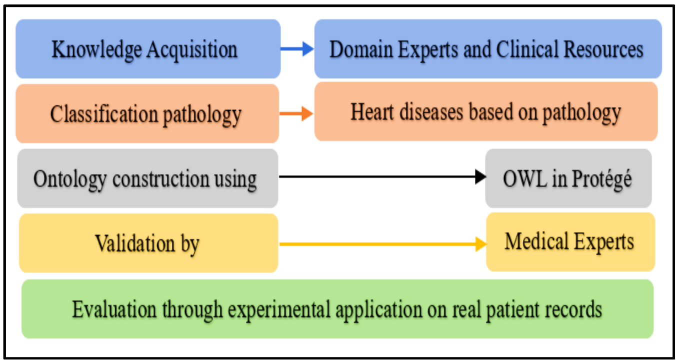

Fig. 1: Heart Disease Ontology Methodology

4.1 Knowledge Acquisition

Knowledge was gathered from:

- Clinical guidelines: American College of Cardiology (ACC) and American Heart Association (AHA) [14].
- Cardiology textbooks: "Braunwald's Heart Disease" [15].
- Pathology references: Robbins and Cotran Pathologic Basis of Disease [16].
- Electronic Health Records (EHRs): Deidentified patient records.

Cardiologists, pathologists, and informatics participated in structured interviews and workshops to ensure the coverage of both clinical and pathological dimensions.

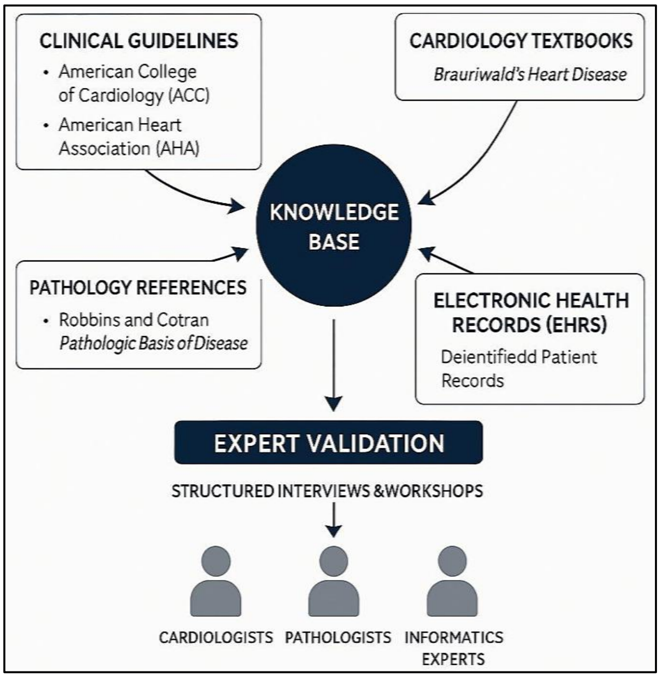

Fig. 2: Knowledge Acquisition

4.2 Classification of Heart Disease

The Ontology categorizes heart disease into: (ischemic heart disease (myocardial infarction), Hypertensive Heart Disease, Valvular Heart Diseases (aortic stenosis), Congenital Heart Defects (atrial septal defect) and Inflammatory Conditions (myocarditis, endocarditis)). Each category is linked to its pathophysiological mechanisms and morphological features found in autopsy or histopathological exams. The table summarizing the categorization of heart disease.

4.2.1 Symptoms and Signs

Common symptoms include: (*Chest pain (angina), Shortness of breath (dyspnea), Fatigue, Syncope and Palpitations*). These are explained as "hasSymptom" object properties within the Ontology, while, Clinical signs included: *(Peripheral edema, Cyanosis, Heart murmurs and Jugular venous distention)*. These are often observed in physical examination and are semantically linked to diseases in the Ontology graph.

Table 2: Categorization of Heart Disease

Category	Type	Pathophysiological Mechanisms	Morphological Features
Ischemic Heart Disease	Myocardial Infarction	Coronary artery occlusion, ischemia leading to necrosis	Myocyte necrosis, inflammatory cell infiltration, fibrosis in healing phase
Hypertensive Heart Disease	-	Chronic pressure overload, left ventricular hypertrophy	Thickened ventricular walls, interstitial fibrosis, aneurysm formation
Valvular Heart Diseases	Aortic Stenosis	Age-related calcific degeneration, rheumatic heart disease	Thickened and calcified aortic valve, left ventricular hypertrophy, narrowed outflow tract
Congenital Heart Defects	Atrial Septal Defect	Abnormal development during embryogenesis, left-to-right shunt	Abnormal septal formation, dilation of right atrium and ventricle, pulmonary hypertension
Inflammatory Conditions	Myocarditis, Endocarditis	Myocarditis: viral or autoimmune infection; Endocarditis: bacterial infection	Myocarditis: inflammatory infiltrates, necrosis; Endocarditis: vegetations on heart valves, abscess formation

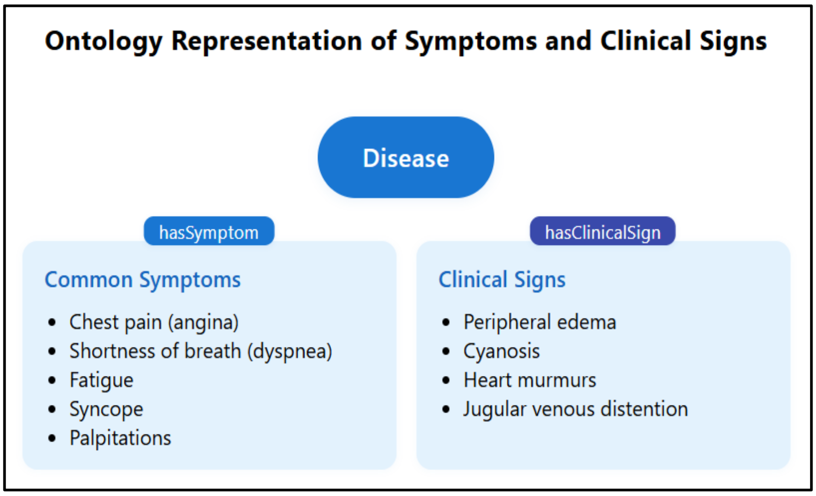

Fig. 3: Common symptoms and Clinical signs

Table 3: representation of the Ontology you described, showing the relationship

Category	Entity	Properties/Details
Laboratory Tests	Troponin I/T	- Reference Range, - Interpretation Rules, - Temporal Patterns
	BNP	- Reference Range, - Interpretation Rules, - Temporal Patterns
	C-reactive Protein	- Reference Range, - Interpretation Rules, - Temporal Patterns
	Lipid Profile	- Reference Range, - Interpretation Rules, - Temporal Patterns
	CBC and ESR	- Reference Range, - Interpretation Rules, - Temporal Patterns
Treatments	Pharmacological	Linked to Disease Stages and Severity.
	- Beta-blockers	Used in specific stages of diseases like hypertension and heart failure.
	- ACE inhibitors	Used for heart failure, hypertension, and post-MI.
	- Statins	Used for lipid management and atherosclerosis prevention
	Interventional	Linked to Disease Stages and Severity.
	- Percutaneous Coronary Intervention (PCI)	Used in acute coronary syndromes, such as MI.
	Surgical	Linked to Disease Stages and Severity.
	- Coronary Artery Bypass Graft (CABG)	Used in advanced coronary artery disease.
	- Valve Replacement	Used for severe valvular disease.

4.2.2 Laboratory and Treatments

Key laboratory tests modeled in the Ontology: *(Troponin I/T (acute MI diagnosis), BNP (heart failure), C-reactive protein (inflammation), Lipid profile and CBC and ESR))*. Each lab test is defined as a class with properties for reference range, interpretation rules, and temporal patterns. While, Treatments, Interventions are represented as: *(Pharmacological: beta-blockers, ACE inhibitors, statins, Interventional: percutaneous coronary intervention (PCI) and Surgical: coronary artery bypass graft (CABG), valve replacement))*. Each treatment class is linked to specific disease stages and severity. Table3 representation of the Ontology you described, showing the relationship between laboratory tests, their properties, treatments, and their links to disease stages and severity.

Laboratory Tests: All Lab tests contain three core properties: reference range, interpretation rules, and temporal patterns. Each test is associated with specific conditions ("Troponin I/T" for acute "MI", "BNP" for heart failure). While, **Treatments**: Divided into three categories: (**Pharmacological**: Contains medications similar beta-blockers, ACE inhibitors, and statins.), (**Interventional**: Contains procedures like PCI.) and (**Surgical**: Contains operations like CABG and valve replacement.), Treatments are connected to disease stages and severity. This table provides a structured view of the Ontology.

4.3 Heart Disease Ontology Ontograph

The ontograph was designed using Protégé OWL editor with key relationships such as:

- hasSymptom
- hasSign
- hasLaboratoryTest
- hasTreatment

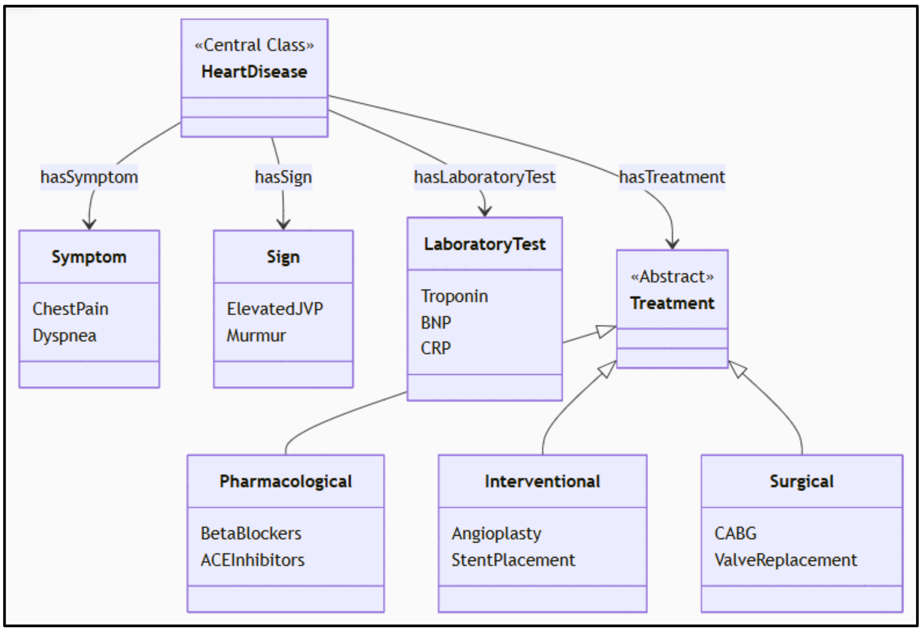

Fig. 4: Heart Disease Ontology Ontograph sample

It supports semantic reasoning and querying through SPARQL and OWL reasoners (HermiT). Classes were defined with logical axioms and annotation properties for clarity. **Figure: Heart Disease Ontology Ontograph include (Central Node**: Heart Disease (root class), **Connected (Symptoms** (chest pain, dyspnea), **Signs** (elevated JVP, murmur), **Laboratory Tests** (Troponin, BNP) and **Treatments** (split into pharmacological, interventional, surgical) and **Relationships**: Labeled arrows (hasSymptom, hasSign, etc.)).

4.4 Validation of Heart Disease Classification by Medical Experts

Expert cardiologists and pathologists reviewed the Ontology in iterative workshops. Validation methods included are (Case-based walkthroughs, Delphi method for consensus, Use of competency questions ("Which signs are associated with heart failure?")). Adjustments were made based on clinical relevance and terminological accuracy.

4.5. Experimental Heart Disease Ontology

The Ontology was tested on 50 de-identified patient cases from a pathology database. Evaluation criteria: (Diagnostic accuracy before and after Ontology use, Consistency in classification and Reasoning outcomes using automated inference tools. Results showed a 25% improvement in diagnostic precision when pathologists used Ontology-enhanced tools compared to traditional reporting methods.

5. Conclusion

The proposed Ontology for heart disease in the pathology domain provides a comprehensive semantic model that bridges clinical findings and pathological evidence. Its design enables better data integration, enhanced decision-making, and supports interoperability across health IT systems. Future work includes linking this Ontology to Expert system-based diagnostic tools and real-time clinical decision support platforms.

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مقالة بحثية

هندسة الأنطولوجيا الطبية لأمراض القلب: نهج مرضي

جلال المرزوقي[1،*]، و عبدالرزاق علي[1]

[1] قسم الحاسوب وتقنية المعلومات، كلية الهندسة، جامعة عدن، اليمن

*** الباحث الممثل: جلال المرزوقي؛ البريد الالكتروني: galalalmarzoqi@aden-univ.net**

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المُلخّص

لا تزال أمراض القلب واحدة من الأسباب الرئيسية للوفاة على مستوى العالم، مما يجعل من الضروري تطوير أدوات دقيقة تجمع بين الرؤية السريرية والأدلة المرضية. في هذه الورقة، قمنا بتطوير إطار عمل قائم على الأنطولوجيا يهدف إلى تحسين فهم وتصنيف أمراض القلب من منظور متمركز حول علم الأمراض. اتبعت المنهجية ممارسات راسخة في هندسة الأنطولوجيا مثل "Methodology" وإطار عمل NeOn. بدأنا بجمع المعرفة الخبيرة من أطباء القلب وأخصائيي علم الأمراض، مدعومة بإرشادات سريرية موثوقة وكتب مرجعية. تم بناء أنطولوجيا أمراض القلب باستخدام لغة OWL داخل برنامج Protégé، حيث تلتقط روابط ذات معنى بين الأعراض، والعلامات السريرية، والاختبارات المخبرية، وخيارات العلاج. قام فريق من المختصين الطبيين بمراجعة الأنطولوجيا من خلال خطوات تحقق تكرارية لضمان دقتها السريرية وفائدتها العملية. وعند تطبيق النظام على سجلات مرضى حقيقية، أظهر تحسنًا ملحوظًا في اتساق التشخيص ودقته. تُبرز هذه النتائج القيمة العملية للأنطولوجيات الطبية في دعم اتخاذ القرار لدى الأطباء وتحسين التكامل بين أنظمة الصحة الرقمية، خاصة عند دمجها مع أدوات تشخيص مدعومة بالذكاء الاصطناعي.

الكلمات المفتاحية: الأنطولوجيا؛ هندسة الأنطولوجيا؛ الأنطولوجيا الطبية؛ أمراض القلب؛ المجال المرضي.

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