Congenital Heart Disease: An Ontology-Based Approach for the Examination of the Cardiovascular System

M. Esposito

Institute for High-Performance Computing and Networking (ICAR)
National Research Council (CNR)
Via Castellino 111, 80131 Naples, Italy
massimo.esposito@na.icar.cnr.it

Abstract. Congenital Heart Disease (CHD) represents the most common group of congenital malformations of the heart and of its blood vessels. In this paper, we present an ontology-based approach to detect abnormalities and malformations due to CHD. In particular, we propose a formal and well-defined model to represent the anatomy of the cardiovascular system, based on the SNOMED vocabulary. The model defines either the anatomy of the cardiovascular system in normal patients or the anatomy characterized by malformations and abnormalities in CHD patients. We have formalized this model in OWL ontologies and SWRL rules and, then, we have used a logic reasoner to identify either CHD patients or the heart abnormalities and malformations they are affected by.

Keywords: Congenital heart disease, ontologies and rules, model checking.

1 Introduction and Related Work

1.1 Introduction

Congenital Heart Disease (CHD) represents the most common group of congenital malformations of the heart and of its blood vessels that affect between 7 and 8 per 1000 live-born infants [1].

In many cases, CHD contributes significantly to infant mortality and morbidity and may only be recognized when the affected infant develops life-threatening symptoms of cardiovascular collapse. The clinical examination of the cardiovascular system at the time of routine clinical newborn examination can enable to identify early those infants who are at risk of adverse or irreversible outcomes as a consequence of congenital heart defects, whilst they are still pre-symptomatic.

The large number of routine clinical investigations on the one hand, and the need of a sound examination of every single case on the other hand have necessarily decreased the cardiologist productivity and the quality of the diagnosis reports.

This highlights the need of an automated approach for the examination of the cardiovascular system that could support the cardiologists, providing outputs which can be used as a 'second opinion' in detecting malformations and abnormalities. Moreover it could also increase the cardiologist productivity and improve the quality of the

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diagnosis reports. Such an approach should automatically combine the lower-level information, coming from a preliminary segmentation step, with a higher level domain-specific knowledge, closing the existing semantic gap.

We think the Semantic Web languages and technologies, and in particular ontologies and rules, could close this gap for the following reasons:

- Ontologies and rules enable to represent, explicitly and formally, the domain-specific knowledge a cardiologist uses in his clinical investigations. Ontologies and rules can be formalized using semantic representation languages as OWL[1], SWRL [2] and RDF[3]. These languages, characterized by a high degree of expressiveness and modeling power, enable to formalize complex models in an accurate and sound way.
- OWL ontologies and SWRL rules can be processed by logic reasoners. These reasoners perform inference patterns that can be exploited to automatically examine the cardiovascular system and detect abnormalities.

As a result, in this paper, we propose an automated approach, based on the use of ontologies and rules, to examine the cardiovascular system and identify abnormalities and malformations due to CHD.

More precisely, we have realized a formal and well-defined model to represent the anatomy of the cardiovascular system, based on the SNOMED[4] vocabulary. This model defines either the anatomy of the cardiovascular system in normal patients or the anatomy characterized by malformations or abnormalities in CHD patients. Moreover, we have formalized this model in OWL ontologies and SWRL rules and used a logic reasoner to identify either CHD patients or the heart abnormalities and malformations they are affected by.

The rest of this work is organized as follows. Section 2 describes the semantic approach. Section 3 presents our cardiovascular examination method and overviews some application examples. Finally, section 4 concludes the work.

1.2 Related Work

In the past, many authors have proposed the use of Expert Systems as Clinical Decision Support Systems (CDSS) in order to directly assist physicians with decision making tasks [2], [3],[4],[5].

In [2] the authors investigate the application of artificial neural networks in medical diagnosis, presenting a hybrid fuzzy-neural automatic system and a simple and applied method. In [3] the authors present a rule-based CDSS for the diagnosis of Coronary Artery Disease, based on the development of a fuzzy model. In [4] the authors focus on using decision-theoretic networks for decision-making and discuss, as a typical example, the treatment of patients with aortic coarctation, a kind of CHD. In [5] the authors propose a mechanism for reasoning about the differential diagnosis of cases involving the symptoms of heart failure defining a causal model.

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[1] http://www.w3.org/TR/owl-semantics/
[2] http://www.w3.org/Submission/2004/SWRL/
[3] http://www.w3.org/RDF/
[4] http://www.snomed.org/

The weakness of all these approaches relies on the poor expressiveness and modeling power of the Expert Systems, that do not enable to formalize a well-defined, unambiguous and structured representation of the domain-specific knowledge. Differently, our approach, exploiting the modeling power of ontologies and rules, enables to describe the knowledge in a structured, organized and more human-understandable manner, facilitating the information formalization and interpretation.

Recently, approaches using ontologies and rules for modeling domain-specific medical knowledge have been proposed [6],[7],[8]. More specifically, in [6],[7] the authors focus on the brain examination and, in particular, they aim at modeling brain knowledge in order to label brain images. These works, similarly to us, use i) ontologies and rules to represent a domain-specific knowledge and ii) logic reasoners to perform reasoning mechanisms.

In [8] the authors use clinical and spatial ontologies representing the human heart to automatically generate a diagram based on a patient's information in cardiology databases. This work, similarly to us, also defines a model for the cardiovascular system, but it is not related to any shared and well-defined collection of medical terminology. Anyway, none of these approaches aim at supporting physicians in the medical diagnosis.

2 The Semantic Approach

2.1 Our Proposal of a Cardiovascular Model

The approach presented in this paper relies on a model that we have defined to provide a unique and uniform representation for the anatomy of the cardiovascular system. This model is fundamentally based on the SNOMED (Systematized Nomenclature of Medicine) vocabulary, that is a systematically organized and computer readable collection of medical terminology covering most areas of clinical information. Our model has been formalized in OWL ontologies and SWRL rules and represents the cardiovascular knowledge by specifying anatomical concepts and relationships between them.

Fundamentally, the cardiovascular system consists of three sub-systems, the heart, the arterial system and the venous system. The heart has four chambers, separated by grooves. Blood is pumped through the chambers, aided by four heart valves, to all parts of the body through a series of vessels, termed arteries. The arteries undergo enormous ramification in their course and end in minute vessels, called arterioles, which in their turn open into a close-meshed network of microscopic vessels, termed capillaries. After the blood has passed through the capillaries, it is collected first into a series of minute vessel, termed venule, and then into a series of larger vessels, called veins, by which it is returned to the heart.

It is worth noting that our model takes into account only the part of the arterial and venous systems that are strictly related to the heart and that can be affected by CHD. First, we have identified the main cardiovascular concepts, that are shown in figure 1, by taking into account the medical terminology specified in SNOMED.

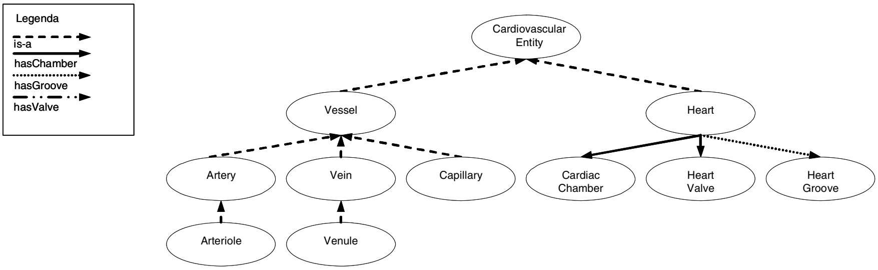

Fig. 1. The main cardiovascular concepts

Table 1. Mereological roles

Role	Domain	Range	Inverse	Trans.	Sym.
HasSegment	Vessel	Vessel	isSegmentOf	yes	no
HasBranch	Vessel	Vessel	isBranchOf	no	no
HasVisceralBranch	Vessel	Vessel	isVisceralBranchOf	no	no
HasParietalBranch	Vessel	Vessel	isParietalBranchOf	no	no
HasTerminalBranch	Vessel	Vessel	isTerminalBranchOf	no	no
HasChamber	Heart	Cardiac Chamber	isChamberOf	no	no
HasGroove	Heart	Heart Groove	isGrooveOf	no	no
HasValve	Heart	Heart Valve	isValveOf	no	no

Table 2. Topological roles

Role	Domain	Range	Inverse	Trans.	Sym.
isConnectedTo	Vessel	Vessel	No	no	Yes
isSeparatedFrom	Cardiac Chamber	Cardiac Chamber	No	no	Yes
isSeparatedFrom VentricleBy	Cardiac Chamber	Heart Groove	Separates VentricleFrom	no	No
isSeparatedFrom AtriumBy	Cardiac Chamber	Heart Groove	Separates AtriumFrom	no	No
emptyIn	Vessel	Cardiac Chamber	isEmptiedBy	no	No
takesOriginFrom	Vessel	Cardiac Chamber	givesOrigin	no	No
isIncludedIn	Heart Valve	Cardiovascular Entity	includes	no	No

Table 3. Relationship between roles

Rule	Antecedent	Consequent
R1	**if**(Cardiac_Chamber_X isSeparatedFromVentricleBy Heart_GrooveY **AND** Cardiac_Chamber_Z isSeparatedFromVentricleBy Heart_GrooveY)	Cardiac_Chamber_X isSeparatedFrom Cardiac_Chamber_Z
R2	**if**(Cardiac_Chamber_X isSeparatedFromAtriumBy Heart_GrooveY **AND** Cardiac_Chamber_Z isSeparatedFromAtriumBy Heart_GrooveY)	Cardiac_Chamber_X isSeparatedFrom Cardiac_Chamber_Z
R3	**if** (Vessel_X HasSegment Vessel_Y **AND** Vessel_Y HasBranch Vessel_Z)	Vessel_X HasBranch Vessel_Z
R4	**if** (Vessel_X HasSegment Vessel_Y **AND** Vessel_Y HasVisceralBranch Vessel_Z)	Vessel_X HasVisceralBranch Vessel_Z
R5	**if** (Vessel_X HasSegment Vessel_Y **AND** Vessel_Y HasParietalBranch Vessel_Z)	Vessel_X HasParietalBranch Vessel_Z
R6	**if** (Vessel_X HasSegment Vessel_Y **AND** Vessel_Y HasTerminalBranch Vessel_Z)	Vessel_X HasTerminalBranch Vessel_Z
R7	**if** (Vessel_T HasSegment Vessel_X **AND** Vessel_T HasSegment Vessel_Y **AND** Vessel_T HasSegment Vessel_Z **AND** Vessel_X isConnectedT Vessel_Y **AND** Vessel_Y isConnectedTo Vessel_Z)	Vessel_X isConnectedTo Vessel_Z

Moreover, we have identified three typologies of relationship: i) the subsumption relationship concerns the generalization/specialization between cardiovascular concepts; ii) the mereological relationship concerns part-whole relations between cardiovascular concepts; iii) the topological relationship concerns neighborhood relations between cardiovascular concepts.

We have modeled these typologies of relationship by a set of roles. Each role has a domain (the set of possible subject concepts) and a range (the set of possible object concepts). Besides, each role can have a corresponding inverse role and it can be transitive (if it remains true across chains of links) or symmetric (if it can be applied in both the directions). Moreover, we have realized a set of SWRL rules, reported in table 3, in order to capture relationships between roles.

2.2 CHD: A Typical Heart Abnormality

The presented model takes into account only the normal anatomy of the cardiovascular system. Nevertheless, the cardiovascular system in a patient affected by CHD is characterized by a different anatomy. In this paper, we consider, as an illustrative example, a typical congenital abnormality, that is the Interrupted Aortic Arch (IAA) defect. We have also taken into account other typical heart abnormalities, but we have not reported their descriptions in this paper for sake of brevity.

Below, we highlight the anatomical differences existing respectively between the cardiovascular systems in a normal patient and in a patient affected by IAA.

In normal patients, the aorta consists of three segments: an ascending segment, a transverse segment or arch, and a descending segment.

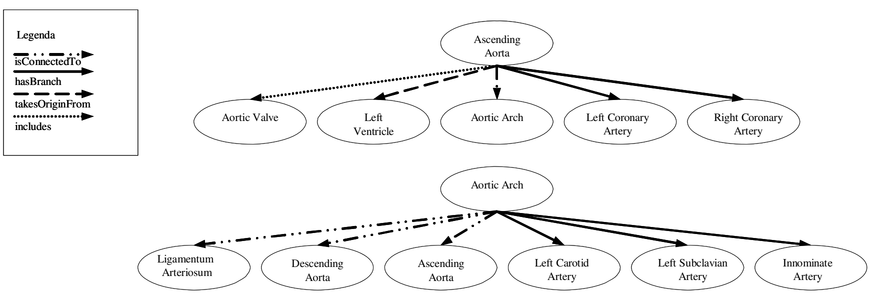

Fig. 2. The Ascending Aorta and the Aortic Arch

The ascending aorta takes its origin from the left ventricle, it gives rise to the left and right coronary arteries and its proximal portion includes the aortic valve.

The aortic arch begins at the innominate artery and ends at the ligamentum or ductus arteriosum. It gives rise to the innominate, left carotid and left subclavian arteries. The descending aorta begins at the ligamentum or ductus and consists of two segments, the thoracic aorta and abdominal aorta.

When a patient is affected by IAA, the aorta does not develop completely in the area of the arch. As a result, the aorta is divided only into two parts, the ascending aorta and descending aorta, that are not connected to each other. IAA can be classified on the basis of the site of interruption. Type A is distal to the left subclavian artery, type B is proximal to the left subclavian artery, and type C occurs between the innominate artery and the left common carotid artery.

As a result, we have modified the presented model, taking into account these differences, in order to describe the anatomy of the cardiovascular system in a patient affected by IAA.

Specifically, the cardiovascular model for patients affected by IAA presents the following changes with respect to the presented one: i) the aorta has two segments, the ascending aorta and the descending aorta (the aortic arch is not present); ii) the ascending aorta is not connected to the descending aorta; iii) IAA Type A: the ascending aorta has the innominate artery, the left subclavian artery and the left carotid artery as its branches; iv) IAA Type B: the ascending aorta has the left subclavian artery and the left carotid artery as its branches; v) IAA Type C: the ascending aorta has the innominate artery as its branch.

3 The Cardiovascular Examination Procedure

3.1 The Method

The method we have used to perform the examination of the cardiovascular system consists of two main steps.

The first step is the segmentation of the cardiac images. We suppose the segmentation procedure is able to identify the anatomical structure of the heart and of its blood vessels and the topological relations existing among them. It is worth noting that, in this paper, we focus our attention only on the next step.

The second step is fundamentally a model checking procedure that performs an automatic formal verification of the outputs of the segmentation step with respect to the defined cardiovascular models. We make use of the logic reasoner presented in [9], that integrates and reasons about ontologies and rules to perform this procedure.

More precisely, we use the presented model and the information produced after the segmentation step to populate the knowledge base of the reasoner. The knowledge base consists of i) the Tbox, populated by using the ontologies formalized in OWL, ii) the Rule-box, populated by using the rules formalized in SWRL, and iii) the Abox, populated with the outputs of the segmentation step, formalized in RDF.

Moreover, we exploit two inference patterns provided by the reasoner, that are the instance checking and the consistency checking. The instance checking determines whether an individual from the Abox is always an instance of a certain concept. The consistency checking determines whether two assertions about a same individual are inconsistent with respect to the TBox. We use these inference patterns to determine whether the current Abox contains a consistent instance of the Tbox.

Our model checking procedure consists of a set of model checking executions that, in turn, perform the instance and consistency checkings on a different cardiovascular model.

We start populating the Tbox with the ontologies and rules that describe a normal cardiovascular system The Abox is populated with the information coming from the segmentation step, that describe the cardiovascular system under exam.

The reasoner performs the instance and consistency checkings to verify whether the cardiovascular system under exam, loaded in the Abox, does not violate the normal cardiovascular model, loaded in the Tbox.

If the reasoner provides a no answer, this means that the cardiovascular system under exam is not coherent and consistent with the normal cardiovascular model. Hence, the patient under exam is affected by a kind of abnormality. Otherwise, the patient is affected by no abnormality. If the patient is affected by a kind of abnormality, we have to change the model loaded in the Tbox with the one describing the anatomy of a cardiovascular system affected, for an example, by IAA Type A and, then, verify whether the individuals in the Abox do not violate it.

We have to change the model in the Tbox and launch a new model checking execution until the reasoner gives a positive answer. When it happens, it is possible to query the reasoner in order to determine which model is currently loaded in the Tbox. This enables to find out the abnormality that affects the patient in exam.

3.2 Some Application Examples

In this subsection we describe two application examples of the cardiovascular examination procedure described above. In particular, suppose that the segmentation has detected the anatomical structure of the heart and of its blood vessels. Moreover, suppose the initial set of information submitted to the reasoner is the following:

I1. *Aorta(aor0)*
I2. *AscendingAorta(asc_aor0)*
I3. *AorticArch(aor_arc0)*
I4. *DescendingAorta(des_aor0)*
I5. *hasSegment(aor0, asc_aor0)*
I6. *hasSegment(aor0, aor_arc0)*
I7. *hasSegment(aor0, des_aor0)*
I8. *InnominateArtery(inn_art0)*
I9. *LeftArtery(lef_art0)*
I10. *LeftSubclavianArtery(lef_sub_art0)*
I11. *isConnectedTo(asc_aor0, aor_arc0)*
I12. *isConnectedTo(aor_arc0, des_aor0)*
I13. *isConnectedTo(asc_aor0, des_aor0)*
I14. *hasBranch (aor_arc0, inn_art0)*
I15. *hasBranch (aor_arc0, lef_art0)*
I16. *hasBranch (aor_arc0, lef_sub_art0)*

This set of information represents a fragment of our current Abox. The information I13 is derived from the application of the rule R7 on the individuals I5, I6, I7, I11, I12. The reasoner performs the instance and consistency checkings and verifies that this information does not violate the normal cardiovascular model loaded in the Tbox. Hence, we can conclude that the patient under exam is normal.

Instead, suppose the initial set of information is the following:

I1. *Aorta(aor0)*
I2. *AscendingAorta(asc_aor0)*
I3. *DescendingAorta(des_aor0)*
I4. *hasSegment(aor0, asc_aor0)*
I5. *hasSegment(aor0, des_aor0)*
I6. *InnominateArtery(inn_art0)*
I7. *LeftArtery(lef_art0)*
I8. *LeftSubclavianArtery(lef_sub_art0)*
I9. *hasBranch (asc_aor0, inn_art0)*
I10. *hasBranch (asc_aor0, lef_art0)*
I11. *hasBranch (asc_aor0, lef_sub_art0)*

Now, this is a fragment of our current Abox. The reasoner performs the instance and consistency checkings and verifies that this information violates the normal cardiovascular model loaded in the Tbox. Hence, we can conclude that the patient under exam is affected by a kind of abnormality.

Now, we load the model describing the IAA Type A in the Tbox. Then, the reasoner performs the instance and consistency checkings and verifies that the individuals in the Abox do not violate the IAA Type A model. Hence, we can conclude that the cardiovascular system of the patient under exam is affected by IAA Type A.

4 Conclusions and Directions for Future Works

In this paper, we have described an ontology-based approach for the examination of the cardiovascular system that aims at detecting abnormalities due to CHD.

The main goal of this work has been to highlight the possible applicability of a novel approach in order to support cardiologists in the medical diagnosis. For this reason, the paper is very descriptive and illustrative and often omits many technical details. Moreover, this work represents only a part of an ongoing research and, in particular, it describes only a first step. As a matter of fact, at the moment, the method has been tested only over a few proof data, and so experimental results and performance evaluations are still missing. Next step of our research will be to investigate the applicability and reliability of the proposed approach in real cases.

Future work will also study i) how to overcome present Semantic Web language and tools limitations and ii) how to extend the approach with uncertainty processing.

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