Codification and Application of a Well-Founded Heart-ECG Ontology

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Abstract. *Despite the fact that many authors in the literature defend the need of ontologically well-founded languages for ontology representation, this approach has not yet been broadly adopted. We present in this paper a codification of a well-founded heart-ECG domain ontology in OWL+SWRL. The lightweight ontology produced is then applied to a web environment for heart electrophysiology reasoning and visualization. We also reflect on this codification process to argue in favor of the view that two classes of languages are needed for ontology engineering: (i) a theoretically well-founded representation language for creating conceptual domain ontologies; (ii) a lightweight representation language for codifying these conceptual ontologies.*

1. Introduction

The purpose of using ontological distinctions for building domain ontologies has long been recognized in the literature [Gruber 1995], [Guarino 1997]. These distinctions are significant for making the real-world meaning of concepts and relations precise and explicit. By placing this discussion in the realm of languages, it has also been evidenced in the literature the existence of different language levels [Brachman 1979], [Guarino 1994]. Languages which are to be at the so-called *ontological level* must explicitly commit to fundamental ontological distinctions in their metamodels comprising categories such as kinds, roles and mixins. Languages that follow this criterion are termed ontological level languages or ontologically well-founded languages (e.g., see [Evermann 2003] or [Guizzardi 2005]).

Although recent research initiatives such as [Fielding et. al 2004] and [Guizzardi 2006] have elaborated on why domain ontologies must be represented with the support of a foundational theory, such an approach has still not been broadly adopted. As reported by Jones et al. (1998) and Wache et al. (2001), most existing methodologies do not emphasize or even completely ignore this aspect. We believe that the main reasons include: (i) the call for some expertise in handling philosophical issues [Jarrar and Meersman 2007] and using logical axiomatization; (ii) the contrast between the need of high expressivivity when using ontologically well-founded language languages and the well-known computational efficiency required in the target applications.

With respect to (i), in fact the theoretical notions which are required for suitable characterizations of domain conceptualizations are of a complex nature. This puts emphasizes on the need for appropriate computational support (e.g. design tools) for hiding as much as possible this inherent complexity from conceptual modeling practioneers. Regarding (ii), and in pace with [Guizzardi 2007] and [Guizzardi and

Halpin 2008], we argue that two classes of languages are required to fulfill two sets of requirements in ontology engineering. On one side, a **reference ontology** is to be a special kind of conceptual model, an engineering artifact with the additional requirement of representing a model of consensus within a community. In other words, it is an off-line solution-independent specification whose aim is to make a clear and precise description of the domain elements for the purposes of communication, learning and problem-solving. As a consequence, *domain ontologies must be represented by ontologically well-founded languages*. On the other side, a **lightweight ontology** is to be a model for computation, amenable to be used, say, in knowledge-based systems as serving to represent the universe of discourse with reasoning purposes. It is then an on-line shareable software artifact that must strive for performance, scalability, adaptability, interoperability and so on. With this in mind, a number of languages could be used offering different trade-offs involving different non-functional requirements. As we further elaborate, among other language alternatives, we have been experimenting with the semantic web technologies OWL DL and SWRL (Semantic Web Rule Language) to produce lightweight ontologies as codifications of reference ontologies.

In [Gonçalves et al. 2007], we have published a heart-ECG reference ontology represented in OntoUML [Guizzardi 2005]. OntoUML is a UML profile that augments the UML expressiveness based on Unified Foundational Ontology (UFO) [Guizzardi and Wagner 2005]. In other words, it is an ontologically well-founded language. In the present article, we introduce a codification of this heart-ECG reference ontology using the ontology implementation languages OWL DL + SWRL. The lightweight ontology produced is then applied in an interactive web environment for heart electrophysiology reasoning and visualization.

The remainder of this paper is organized as follows. Section 2 introduces the heart-ECG domain ontology by briefly discussing the subject domain and posing the ontology competence questions. In Section 3 we discuss the languages chosen for producing the heart-ECG lightweight ontology in view of non-functional requirements of a target application. In Section 4, we firstly discuss some relevant issues we have experienced in the mapping between reference ontologies and their codification using this specific target environment; and, secondly, we evaluate this codification in terms of its capability to answer the ontology competence questions. Section 5 then presents a web environment in which the lightweight ontology is applied for heart electrophysiology reasoning and visualization. Finally, in Section 6 we provide final considerations.

2. The Heart-ECG Reference Ontology

In [Gonçalves et al. 2007] we propose a heart-ECG reference ontology that strives for meeting the issues highlighted in the introduction concerning the purpose of reference ontologies. In building this ontology, we have tackled many non-trivial philosophical concerns that naturally arise in dealing with such a biomedical domain. Some examples are: (i) how to represent that although the electrical impulse that triggers a ventricular contraction is the sinoatrial (SA) impulse, the atrioventricular (AV) impulse also contributes (and often indispensably) in such a contraction; (ii) how to state that the heart's role as a blood pump is continually played as long as ventricular contractions are continually performed as result of subtle bioelectric phenomena; or (iii) how to interconnect events inherent to bioelectric phenomena to ECG waveform patterns measured by means of a recording device. This experience has showed not only that ontological distinctions are significant in dealing with this biomedical domain, but also that they contribute a lot for pushing the developer to think much deeper about the universe of discourse.

On the other hand, existing ECG standards such as AHA/MITBIH [Goldberger et al. 2000], SCP-ECG [SCP 2002], HL7 [HL7 2003], FDADF [Brown et al. 2002] and ecgML [Wang et al. 2003] are developed by using standard conceptual modeling languages such as UML that allow the production of models of poor expressivity and clarity. This becomes, in fact, a challenge to overcome as far as these standardization initiatives are mainly concerned to foster semantic interoperability, in general, and data integration, in particular, between heterogeneous health information systems.

The main goal of the heart-ECG domain ontology, which is inherent to a reference ontology, is to provide a domain theory striving for independence of codification languages as much as specific applications. The purpose of this initiative is: (i) conveying a heart-ECG knowledge repository; (ii) addressing semantic interoperability and data/standard integration between health systems and also (iii) supporting AI knowledge-based systems. The ontology covers the domain by connecting multiple albeit complementary levels of granularity. As said before, it is represented in OntoUML, comprising concepts, relations and FOL axioms. It was conceived by following guidelines that have been tested for the last ten years in the development of a number of domain ontologies [Falbo 2004]. As a definition of the purpose of the heart-ECG ontology, as much as serving to an evaluation resource, we have the following <u>competence questions</u>.

CQ1. What conditions must be satisfied for the heart to play the role of a blood pump?

CQ2. What conditions must be satisfied for the heart being able to pump blood to both systemic and pulmonary circulation?

CQ3. What is in the background of an ECG recording session?

CQ4. What is the source of an ECG record?

CQ5. How can one obtain the ECG records acquired in the scope of one treatment?

CQ6. How does an ECG recording device acquire an ECG record?

CQ7. What does the P wave represent in the ECG waveform?

CQ8. What does the QRS complex represent in the ECG waveform?

CQ9. What kind of information does a physician use to identify variations in the morphology and timing of events in the ECG waveform for inferring an interpretation?

These competence questions have also been formalized into FOL axioms in [Gonçalves et al. 2007]. They are supposed to be embedded in the codification, in such a way that allows the implemented ontology to answer them. Moreover, they provide a mapping between the heart activity and electrocardiography concepts.

The heart-ECG reference ontology is divided into sub-ontologies as follows: (i) heart, (ii) bioelectric phenomena, (iii) circulatory phenomenon, (iv) human protocol and (v) ECG. They are interconnected by imports relationships. For brevity, we do not show here all the ontology diagrams nor all its FOL axioms, since it falls completely out of the scope of this paper. A detailed presentation of this ontology can be found elsewhere in [Gonçalves et al. 2007]. Figure 1, however, depicts an indicative part of one of the heart-ECG sub-ontologies. This part comprises the interconnection between two of the most relevant sets of concepts and relations. They are the *heart electrical impulses* and the *ECG elementary forms*, which lie in the bioelectric phenomena and ECG sub-ontologies, respectively. Such a mapping is a key point to relate elementary forms from a *cycle* of the ECG *waveform* to the correlated heart bioelectric phenomena. The usefulness of these associations is further evidenced in Section 5 in the context of the web multimedia environment. In the next section we discuss the choice of a codification environment and some issues that are inherent to this choice.

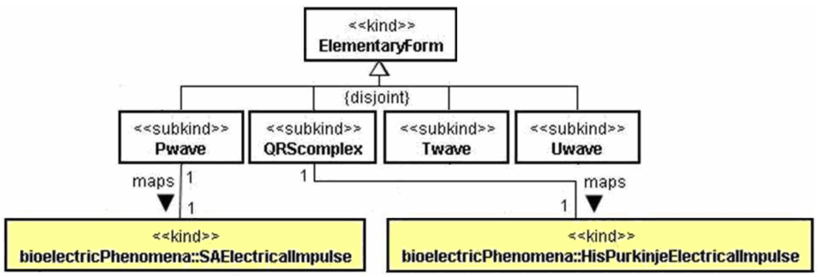

Figure 1. Bioelectric phenomena and ECG ontologies mapping.

3. Codification Environment

The choice of a codification environment must be guided by the end-application non-functional requirements. The main issues of mapping a domain ontology into a lightweight ontology are related to handling the reduction in semantic precision and expressivity. As we have mentioned, the reason is that while languages used for creating reference ontologies focus on representation appropriateness, languages for building lightweight ontologies concentrate on formal reasoning [Guizzardi 2007].

By considering the interactive web environment we have as target, the most relevant requirements are (i) reasonable computational efficiency and (ii) compatibility with semantic web standards. Therefore OWL DL and SWRL have been elected as our codification languages. The latter requirement is met since these technologies are W3C recommendations that constitute noteworthy technologies in the semantic web effort. Regarding the former, OWL DL is based on the Description Logic SHOIN(D), strictly designed to be decidable [Horrocks et al. 2003]. Besides, it can be combined to the rule language SWRL as an extension to include horn-like rules. Although this combination might lead to undecidability in interesting reasoning problems, recent research efforts have proposed alternatives for overcoming this issue. They propose to restrict the use of rules for handling only rules that are *DL-safe* [Motik et al. 2005]. We have applied this principle and reached results that fit well in our application purpose (see Section 5). This has been possible due to the availability of efficient off-the-shelf semantic web reasoners such as Pellet [Sirin et al. 2007].

Concerning the mapping issues, especially the pursuit for maintaining the expressiveness reduction acceptable, the choice for OWL DL and SWRL have also shown to be amenable. The main losses we have found are: (a) decreasing from OntoUML expressivity (quantified intensional modal logics) to OWL DL expressivity

(SHOIN(D)), and (b) transforming all ontologically well-founded concepts and relations into OWL classes and OWL properties respectively. Meanwhile the purposefulness of using a reference ontology remains emphatic once the hierarchical structure of the reference ontology remains correctly designed in the implemented model.

As an attempt to make this tangible, consider the example shown in Figure 2. It depicts three different possible models for representing the concept of customer, which can be either a person or an organization. The first models are ontologically incorrect since: (i) in 2.a, it is not the case that all instances of person (or organization) are customers; (ii) according to 2.b, every instance of Customer is both Person and Organization, thus, the extension of Customer is empty. The model 2.c, otherwise, is a design pattern that provides an ontological solution to the person-organization-customer case which is proposed in [Guizzardi 2006]. Indeed, the correctly designed hierarquical structure of 2.c can be preserved in an implemented model.

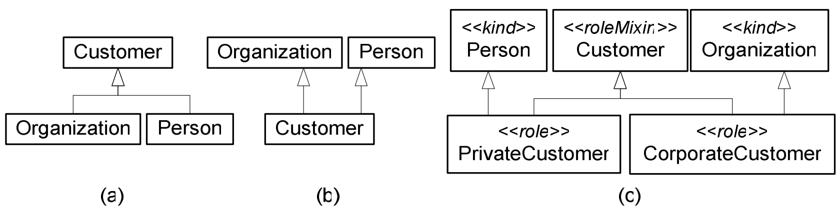

Figure 2. Example of models representing person-organization-customer case.

Nonetheless, we have verified that these losses in expressiveness do not compromise the results expected by our end-application. As a first evidence, the next section elaborates on the heart-ECG ontology implementation in the codification environment.

4. The Lightweight Heart-ECG Ontology

The heart-ECG ontology has been implemented by taking advantage of the Protégé editor. It provides a friendly environment for supporting design in both languages OWL and SWRL. It also has ease integration with the reasoners Racer Pro and Jess. The former can be used for verifying the consistency, inferring class subsumption and classifying individuals. The latter in turn is able to reason with SWRL rules[1].

In handling the mapping between two radically different languages such as OntoUML and OWL+SWRL, there is a need for customizing which characteristics to represent from a specific domain element. As an example, consider the part-whole relation in OntoUML depicted in Figure 3. When mapping this relation into OWL, we immediately lose its modal characteristics, as discussed in Section 3. It is then necessary to consider the information contained in this notation: range, domain, cardinality restriction and transitivity. All these things are embedded in a visual pattern that simply does not exist in OWL. Instead, the OWL concrete syntax requires from one to explicitly include each one of these characteristics a specific relation holds on. Moreover, in OWL it is not possible to represent both transitivity and cardinality

[1] These tools are available at protege.stanford.edu, www.racer-systems.com and www.jessrules.com.

restriction [Bechhofer et. al. 2004]. In this case, one must choose which one to represent. We have deemed that cardinality restriction is more relevant in our case. Another issue that arises concerns how to represent and organize many relations. If a generic relation *isPartOf* is created, it is not possible to restrict the range and domain, and thus representing cardinality restrictions does not make sense. Our choice is to use specific relations like *isPartOf_LeftVentricle_HumanHeart* and *isPartOf_HumanHeart_ HumanBody*, which are represented as subrelations of a generic relation *isPartOf*. This is for improving both organization and query answering. Finally, with respect to inverse relations, Rector and Welty (2005) advice not to represent unless it is necessary, since it increases the reasoning complexity significantly. Nonetheless, we used it in order to represent the cardinality restriction in both directions[2].

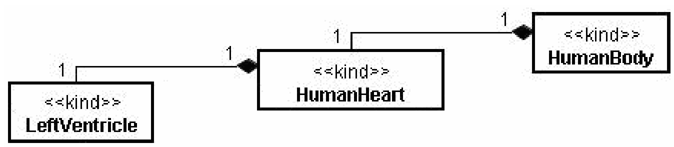

Figure 3. Part-whole relations in the heart-ECG reference ontology.

For obtaining the example results, we have populated the ontology with arbitrary individuals. As follows, for each (selected) competence question, the original axioms written in FOL are presented in combination with its codification in either OWL (using the Protégé syntax) or SWRL.

CQ1. What conditions must be satisfied for the heart to play the role of a blood pump?

Original Axioms:

(A1.1) $\forall$x (LeftVentricleAsAPump(x) $\leftrightarrow$ $\exists$y,z (PurkinjeElectricalImpulse(y) $\wedge$ LVContraction(z) $\wedge$ mediates(z,x) $\wedge$ mediates(z,y)))

(A1.2) $\forall$x (RightVentricleAsAPump(x) $\leftrightarrow$ $\exists$y,z (PurkinjeElectricalImpulse(y) $\wedge$ RVContraction(z) $\wedge$ mediates(z,x) $\wedge$ mediates(z,y)))

(A1.3) $\forall$z (HeartAsPump(z) $\leftrightarrow$ $\exists$x,y (LeftVentricleAsAPump(x) $\wedge$ RightVentricleAsAPump(y) $\wedge$ isPartOf(x,z) $\wedge$ isPartOf(y,z)))

Implemented Axioms:

(A1.1) *LeftVentricleAsPump*: isMediatedBy **some** (LVContraction
and (mediates **some** PurkinjeElectricalImpulse))

(A1.2) *RightVentricleAsPump*: isMediatedBy **some** (RVContraction
and (mediates **some** PurkinjeElectricalImpulse))

(A1.3) *HeartAsPump*: hasPart **some** LeftVentricleAsPump
hasPart **some** RightVentricleAsPump

The implemented axioms are codified as necessary and sufficient conditions for the classes depicted in italic. Thus, the reasoner is able to conclude if a ventricle works as a pump. Furthermore, in case the heart has both its right and left ventricles working as pumps the reasoner also concludes the heart is working as pump.

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[2] Although it is true that in this specifc example the cardinality restriction can be represented as a functional and inverse functional relation, we have chosen for using the same standard in every case.

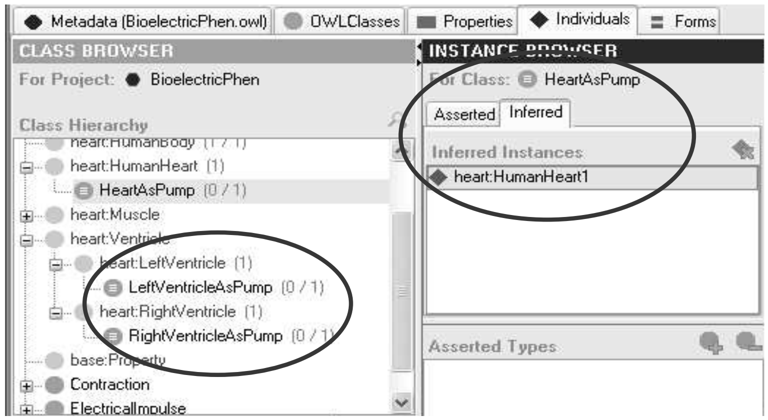

Figure 4. Answer for competence question 1.

The Figure 4 shows the Protégé individuals tab after running Racer for classifying individuals. As a result, the individual *HumanHeart1*, initially created as instance of *HumanHeart* class, is classified as an instance of *HeartAsPump*. Another individual's classification can be noticed by looking at the numbers between backets in front of the classes of right and left ventricle. It has the form (a) or (a / b), which represents how many instances have been created manually (a) and how many have been inferred by the reasoner (b). Thus, one can observe that the individuals created as Left (or Righ) Ventricle have been classified as Left (or Right) VentricleAsPump.

CQ8. What does the QRS complex represent in the ECG waveform?

Original Axiom:

(A8.1) $\forall x$ (QRScomplex(x) $\rightarrow \exists! y$ (HisPurkinjeElectricalImpulse(y) $\wedge$ maps(x,y)))

Implemented Axiom:

(A8.1) QRScomplex(?qrs) $\wedge$ ElectricalImpulse(?ei) $\wedge$ maps(?qrs, ?ei)
$\rightarrow$ query:select(?qrs, ?ei)

This axiom A8.1 is implemented as a SWRL query, used for retrieving some piece of information. It has been reconfigured by focusing on answering the question. Thus, for each instance of QRS complex it is possible to identify the electrical impulse it maps. The Figure 5 shows the results of runing the query A8.1.

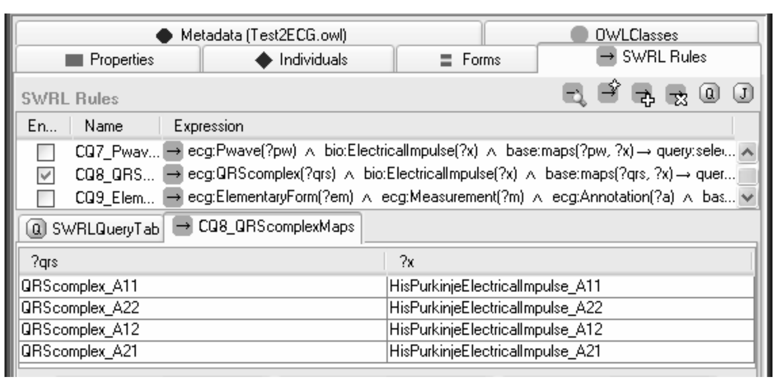

Figure 5. Answer for competence question 8.

This lightweight ontology has then been applied to an ontology-based web environment. The next section aims to describe the basic features of this environment. Our purpose is to give a brief account of how the lightweight ontology is used and, additionally, to highlight its potential in serving as enhanced knowledge base for reasoning services.

5. The Heart Electrophysiology Web Environment

The ontology-based web environment (see Figure 6) is a prototype developed to validate and demonstrate use of the heart-ECG ontology. They are applied for reasoning and visualization of ECG records as well as the human heart electrical behavior. This is an effort to aid teaching the heart electrophysiology by exploiting the potential of ontology in both senses of enhanced representation technique and base for reasoning. In trying to learn heart electrophysiology, students can be limited not only by their imagination, but also by their experiences. In general, the heart bioelectric phenomena are very abstract to human cognition and also require good spatial perception. By using simulations, however, one could actually “see” the electrical currents generated by the heart pacemaker cells. Moreover, interactive simulations allow students to explore, e.g., the heart conduction system, in order to better comprehend the ideas.

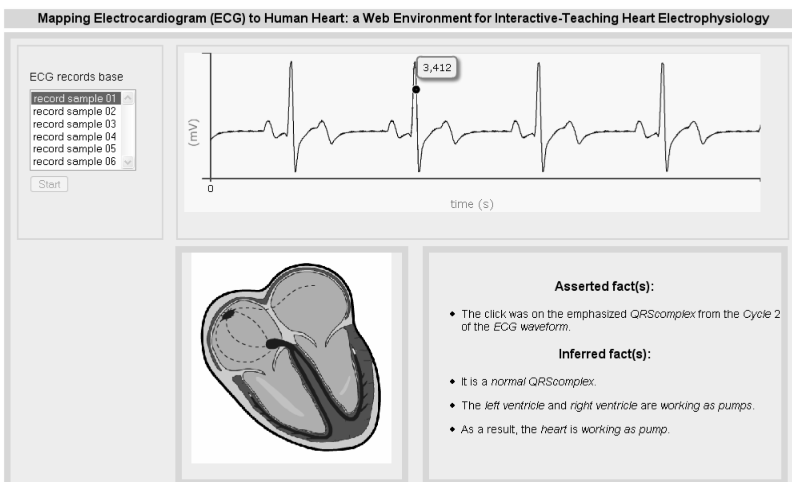

Figure 6. The ontology-based web multimedia environment.

All this has been our motivation for applying the heart-ECG ontology in such a web environment. Besides the ontology, its main components are two flash objects: (i) a chart for presenting the ECG waveform, and (ii) a media to simulate the heart activity. The web application is implemented in Java by taking advantage of the GWT framework[3]. We have adopted the Jena[4] framework and Pellet [Sirin et al. 2007] as Java APIs for handling ontologies. While the former serves in holding OWL ontology in memory, the latter is an OWL reasoner that has exhibited results that seem to be the best in the literature. Pellet is efficient, customizable and also can generate reasoning log information [Neto and Pimentel 2006]. Moreover, it also supports decidability even

[3] Google Web Toolkit (GWT). http://code.google.com/webtoolkit
[4] Jena framework. http://jena.sourceforge.net/index.html

using SWRL rules (since they are DL-safe) as well as consistency validation between OWL restrictions and facts produced by SWRL rules.

The application allows three basic user interactions: **(1)** choosing a record sample for having its ECG data loaded into the chart; **(2)** clicking a point on the ECG waveform chart; which enables reasoning results and reloading the chart for emphasizing the clicked pattern as well as enabling simulation; and **(3)** clicking an specific point of the heart flash object, which enables the simulation of the correspondent heart electrical event and makes the correlated ECG pattern emphasized on the reloaded chart. On the following these features are described in detail in the context of the two client-server RPC's they produce.

5.1. ECG Chart Service

When the user clicks a record sample **(1)** (see Figure 6 on the left), an RPC is triggered by the client requesting a URL. This URL locates a temporary file required by the flash object to plot the ECG waveform on the chart. This file contains the chart data generated by the server from the chosen record sample, which is represented by the ECG web ontology as an OWL A-Box. As far the client receives a successful RPC callback from the server, the flash object is loaded and the ECG data is populated on the chart. This flash chart is interactive. It can receive user clicks that enable self-instructive events.

On the other hand, when the user click comes from the heart flash object **(2)**, an extended version of the RPC just mentioned is triggered with an additional parameter. It informs the clicked part of the heart electrical conduction system. The server then generates the correlated ECG chart, but with an additional feature: the ECG waveform pattern associated with the clicked part of the heart electrical system is emphasized in all *cycles*[5] it is present.

5.2. Inference Service

This service is requested by the client as answer to a click performed on the ECG waveform **(3)**. The service comprises an RPC passing as parameters the current selected record sample as well as the **x** coordinate of the chart clicked point. The server-side logic is then:

(i) searching in the ECG ontology A-Box that holds the record sample what is the ECG pattern where the click has been performed;

(ii) reasoning about the fetched ECG pattern and infer new facts according to the pattern properties (e.g. whether it is a an elementary form, or just heart resting state);

(iii) requesting the ECG chart service for reloading the chart with the recognized pattern emphasized (just in case it is an *elementary form*);

(iv) and, finally, enabling a simulation of the pattern-correlated heart electrical phenomena;

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[5] Heart beats are represented in the ECG waveform by periodic cycles. Each normal cycle has known patterns called elementary forms.

As an example, consider the case illustrated in Figure 6. The click was on the second cycle of the waveform. As far the server has fetched the clicked pattern in the ECG ontology and recognized it as a QRScomplex in the *Cycle 2*, one fact indicating the *QRScomplex 2* was clicked is asserted into the Jena ontology model[6]. The reasoning concerning the selected elementary form is then performed. If the conditions are satisfied, the SWRL rules that map such an ECG elementary form to the related heart concepts are fired. This example illustrates that the ontology implementation answers one of the competence questions (see Section 2), which is about the effectiveness of heart's role of blood pump. As a result, the heart behavior associated to the elementary form is inferred. Such a result is then shown in a log box on the right of the screen. Besides, the ECG chart is reloaded showing only the QRScomplex in the second cycle emphasized. At last, the correlated heart electrical phenomenon is simulated through the flash animation (see Figure 6). All the correlations between ECG patterns and heart electrical phenomena are thus made explicit to the human observer.

6. Final Considerations

In this paper we present the codification and application of a well-founded heart-ECG ontology for heart electrophysiology reasoning and visualization. This work then extends the contribution of [Gonçalves et al. 2007] in providing evidence for the following statements:

(i) Despite a reference ontology is a solution-independent artifact that above all aims to be reusable, in further steps of a development process it can be a reference for producing a number of lightweight ontologies addressing different target-application purposes.

(ii) Although such a design activity inevitably leads to a reduction in the reference ontology expressiveness, the application needs can still be fulfilled; moreover, the ontological distinctions that are inherent to a domain representation in a reference ontology are still purposeful since they impose a hierarquical structure that remains in the lightweight ontology.

As it has been pointed out in this paper, we defend that ontology engineering should account for two classes of languages with different purposes. In this trend, we argue that research efforts concerning semantic interoperability must consider the added-value in using ontologically well-founded ontologies as off-line models. For instance, [Guizzardi 2006] illustrates examples of semantic interoperability problems that can pass undetected when interoperating lightweight ontologies. Likewise, Fielding et al. (2004) discuss how a principled foundational ontology can be used to spot inconsistencies and provide solutions for problems in lightweight biomedical ontologies. As a final example, the need for methodological support in establishing precise meaning agreements is recognized in the Harvard Business Review report of October 2001, which claims that "*one of the main reasons that so many online market makers have foundered [is that] the transactions they had viewed as simple and routine actually*

[6] The heart-ECG sub-ontologies make up a composition hierarchy. When the application is initiated, all them are loaded into a Jena model. For reasoning, in turn, this Jena model is converted into a Pellet model.

involved many subtle distinctions in terminology and meaning". Finally, we advocate that the translation from a reference off-line ontology to an on-line lightweight ontology must be guided by following systematic guidelines. This is indeed a relevant topic of research for which we have started to investigate.

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