Heart failure ontology

Authors: Alan Jovic[a], Dragan Gamberger[b*], Goran Krstacic[c]

[a] Faculty of Electrical Engineering and Computing, University of Zagreb, Unska 3, 10000 Zagreb, Croatia; e-mail: alan.jovic@fer.hr

[b] Rudjer Boskovic Institute, Bijenicka 54, 10000 Zagreb, Croatia; email: dragan.gamberger@irb.hr

[c] Institute for Cardiovascular Diseases and Rehabilitation, Draskoviceva 13, 10000 Zagreb, Croatia; e-mail: goran.krstacic@zg.t-com.hr

[*] Corresponding author: Dragan Gamberger, Rudjer Boskovic Institute, Bijenicka 54, 10000 Zagreb, Croatia

Email: dragan.gamberger@irb.hr

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Abstract

Ontology represents explicit specification of knowledge in a specific domain of interest in the form of concepts and relations among them. This paper presents a medical ontology describing the domain of heart failure (HF). Construction of ontology for a domain like HF is recognized as an important step in systematization of existing medical knowledge. The main virtue of ontology is that the represented knowledge is both computer and human-readable. The current development of the HF ontology is one of the main results of the EU Heartfaid project. The ontology has been implemented using Ontology Web Language and Protégé editing tool. It consists of roughly 200 classes, 100 relations and 2000 instances. The ontology is a precise, voluminous, portable, and upgradable representation of the HF domain. It is also a useful framework for building knowledge based systems in the HF domain, as well as for unambiguous communication between professionals. In the process of developing the HF ontology there have been significant technical and medical dilemmas. The current result should not be treated as the ultimate solution but as a starting point that will stimulate further research and development activities that can be very relevant for both intelligent computer systems and precise communication of medical knowledge.

1. Introduction

Representation of medical knowledge in a form which enables its use by medical computer-based systems is a long-term goal for clinical researchers and technical people developing such systems. The topic is important due to the complexity of the contemporary medical knowledge and the necessity for unambiguous, consistent, and reliable reasoning using clinical data [1]. Effective knowledge representation is also a hot research topic both in theory and practice of computer science related to artificial intelligence [2].

Ontologies are widely accepted as an appropriate form for the conceptualization of knowledge [3]. They represent a basic step in the knowledge representation process which integrates

a) domain vocabulary (terminology),

b) organization of concepts expressed in the chosen terminology into a hierarchical structure (also known as the taxonomy), and

c) description of relations among concepts and/or classes of concepts.

Ontologies usually do not include procedural knowledge which defines how a specific task can be realized, how some problem can be solved, or what has to be done in a specific situation. Complex manipulations with medical knowledge can be and already have been implemented without ontologies. The examples are diverse expert systems [4-6]. However, construction of ontologies and their usage may have a few decisive advantages:

a) Starting from the available ontology which defines the concepts and relations among them, it is much easier to implement any complex system for data and/or knowledge manipulation. The reason is that using the ontology enables appropriate organization of procedural knowledge, regardless if these procedures and functions are loosely or tightly connected with the ontology, and that can be beneficial for the implementation and maintenance of these systems [1,7].

b) Ontologies are reusable in various patient data transformations in diverse applications. This facilitates interoperability among the applications, enables easier verification and comparison, and perhaps most significantly, ensures comparability of results coming from applications using the same ontology [8].

c) The development of the ontologies requires that medical concepts are precisely defined. The result may be significant not only for the implementation of technical systems but also as a basis for precise and unambiguous inter-human communication including scientific publications, text books, and guidelines [9-11].

The work in building medical ontologies started with general taxonomies like SNOMED-CT [12] and general ontologies like foundational model of anatomy (FMA) [13]. The main reason for the development of domain-specific ontologies is that general ontologies may not include all domain-related concepts and relations necessary for a particular use case [14]. Another important reason is the level of the granularity of knowledge that is requested by different medical applications. In general, there is no agreement in respect to what type of knowledge a domain-specific medical ontology should include, the optimal organization of the hierarchy of classes, and how relations with general ontologies should be implemented [15].

In spite of significant interest in developing domain-specific ontologies [16], and recognition of the potential benefits of their application [15], there are practically no domain-specific medical ontologies for clinical decision-making that are publicly accepted and widely used [17].

Heart failure (HF) is a one of the leading causes of morbidity and mortality in the world. Despite significant progress in the treatment, HF incidence and prevalence continue to increase, which represents a serious medical problem [18,19]. There has already been some effort to develop domain-specific medical ontologies. Several approaches have been recently proposed by authors [8,20,21].

This paper presents heart failure ontology as one of the main research results of the EU FP6 project Heartfaid [22].

The structure of the work paper is as follows. In section 2 we describe data sources and tools employed in the construction of the ontology. In section 3 we give an in-depth overview of the ontology. Section 4 discusses the significance of the ontology from the standpoint of human communication, patient data analysis, and decision support. We give an overview of the works of other authors in section 5. Section 6 concludes the paper.

2. Materials and methods

The starting objective for developing the HF ontology was the need for an ontology that will be reusable for different decision support tasks in the heart failure domain. Such an ontology was necessary for the EU Heartfaid project, which required conceptualization and codification of the knowledge from guidelines for handling congestive and acute heart failure patients published by the European Society of Cardiology [19,23-26]. Other sources of information have been used in its development, including, but not limited to, the Unified Medical Language System (UMLS) [27], the Mayo clinic web site [28] and the Open Clinical web site [29].

The ontology is implemented in the Web Ontology Language (OWL) [30] using the Protégé editing tool [31]. OWL has been developed specifically for web applications (i.e. semantic web) but it also has some other favorable properties: it supports a very expressive description logic which enables specification of complex conditions that may be used for the definition of concepts, it supports both object and datatype properties, and it nicely integrates with Semantic Web Rule Language (SWRL) that can be used for procedural knowledge description in ontologies [32,33]. There are publicly available open-source reasoners for OWL [34]. The Protégé tool presents a useful and intuitive visual interface for human editing and validation of the ontology and, by its publicly available Java library, it supports and enables development of various software applications that may use the developed ontologies [35]. All these reasons have been decisive for using OWL in this project.

The information contained in the ontology has been added manually. Several currently available automatic tools for ontology construction have been considered in the project, with the most promissing one being Guideline Interchange Format (GLIF) [36]. The automatic tools failed to combine accurately and efficiently the information coming from diverse sources. The content of the ontology has been proof-read by several medical experts collaborating on the project.

The current version of the ontology is in accordance with the last guidelines of European Society of Cardiology for the diagnosis and treatment of acute and chronic heart failure published in the year 2008 [19]. It has been exported into the HTML format and is available as a series of web pages at http://lis.irb.hr/heartfaid/ontology/. Besides for the Heartfaid project, the ontology has been till now used for scientific research purposes by Division of Biomedical Informatics Research at Stanford University School of Medicine [37] and by Heart + Lung Institute at St. Paul's Hospital at University of British Columbia.

3. Results

3.1. Heart failure ontology description

The HF ontology presents the formalized description of concepts for the HF domain. It includes basic HF concepts, properties that characterize patients, all relevant diagnostic examinations and tests, as well as treatment procedures. The ontology also includes other cardiovascular system related concepts as well as concepts related to other organs connected with HF.

The ontology presents a detailed taxonomic overview of the HF domain with around 200 classes describing HF related concepts. Examples are "Cardiac_hypertrophy", "Blood_pressure_signs" or "Heart_murmurs". These concepts are interconnected with super-class and sub-class properties into a hierarchical tree-like structure. At the basic level there are five super-classes: "HF_concept", "Patient_characteristic", "Patient", "Testing", and "Treatment".

Instances are members of the classes and typically represent list of concrete concepts relevant for the class. For example, the "Cardiac_hypertrophy" class has the following six instances: "Cardiomegaly", "Combined ventricular hypertrophy", "Left_atrial_hypertrophy", "Left_ventricular-hypertrophy", "Right_atrial_hypertrophy", and "Right_ventricular_hypertrophy". In total, the HF ontology includes more than 2000 instances. When possible, instances and classes are connected by their UMLS Concept Unique Identifier (CUI number) and with a list of synonyms. Fig. 1 presents the class "Echocardiography_tests" with seven instances including "Doppler_echocardiography" which has the CUI "C0013520".

[Figure 1 about here]

Like all ontologies, the HF ontology contains properties that connect instances in different classes. These properties enable representation of relations between two concepts. For example, the relationship between instance "Valvular_heart_disease" from the class "Heart_valve_diseases" and the instance "Dyspnea" from the class of "Signs_and_symptoms" may be represented by the property "CouldBeRelatedTo". Similarly, "Hyperkalemia" from the class "Potassium_disorder" may be linked to "Potassium_sparing_diuretics" or "Spironolactone" by the property "MayBeCausedByMedication". The HF ontology includes definitions of more than 100 different properties. In Fig. 1 we can see that the instance "Doppler_echocardiography" has properties "Definition", "Measures" and "CanDetect". "Definition" is a datatype property which contains a text description of the instance while "Measures" and "CanDetect" are object properties which represent relationships to other instances in the ontology. Specifically, for the instance "Doppler_echocardiography" property "Measures" includes 45 instances starting with "Left_atrial_pressure" while property "CanDetect" includes 9 instances starting with "Right_ventricular_diastolic_dysfunction". Besides that, instance "Doppler_echocardiography" has 13 additional properties that are not presented in Fig. 1.

3.2. Heart failure ontology structure

Class "HF_concept" is one of the five main ontology super-classes. It consists of a hierarchy of classes which describe HF terminology, including the risks for congestive heart failure, medical synonyms, and types of classification. One can consider it as a backbone of the whole ontology (Fig. 2). In parentheses, the number of instances in the corresponding class is given.

[Figure 2 about here]

Class "Patient_characteristics" contains patient's demographical characteristics, possible diagnoses, possible signs and symptoms, prognosis and other characteristics. In fact, this hierarchy defines clinical data in the patient's HF medical record. It is interesting to note that both diagnosis and signs and symptoms have been placed in this class. (Fig. 3). Class hierarchy is shown down to the third-level subclasses due to space limitations.

[Figure 3 about here]

Class "Testing" represents knowledge regarding tests performed in medical institutions. This includes a list of tests, usual measurements, measurements normal ranges and relevant results. Physical examination has also been placed within this class. Each test relevant to HF has properties that denote the measurements for that test and also which disorders it can detect. Some tests are invasive or used in combination with other tests and this information is also included. The specification of test measurements is as thorough as possible (Fig 4).

[Figure 4 about here]

Class "Treatment" consists of medical procedures used in the healing process, including medications, devices, invasive and non-invasive procedures, and recommendations regarding HF. Medications are organized into medication groups. Most of the medications relevant for HF symptoms and common comorbidities have been included in the ontology, along with medication dosages and their contraindications (Fig. 5).

[Figure 5 about here]

The last super-class is "Patient" which is the place reserved for factual knowledge about particular patients. Class “Patient” has no subclasses, so there is no class hierarchy that can be displayed. At the moment, for illustrative purposes, this class contains only three patients, but this is the place where in real applications typically many patient data will be present in the same format. The data may be extracted from medical records or they can be inferred as the result of the reasoning process. Patients have roughly 40 properties in the ontology.

For a more detailed representation of the ontology structure than the ones shown in the Fig. 2-5 we refer the reader to the ontology web site [http://lis.irb.hr/heartfaid/ontology/].

3.3. Most significant classes

In this section we present a few most important classes in the HF ontology.

3.3.1. Class "Diagnosis"

Class "Diagnosis" is a subclass of the class "Patient_characteristics". It consists of four subclasses: "Cardiovascular_system_related", "Effects", "Related_to_other_organs", and "Syndromes". "Cardiovascular_system_related" contains also four important subclasses as shown in Fig. 3. These are "Blood_cell_disorder", "Circulation_disorder", "Directly_HF_related" and "Heart_diseases". Each of these classes is further divided into many subclasses and instances.

Class "Heart_diseases" contains the list of all heart-related disorders, excluding HF. For example, instances "Left_atrial_hypertrophy", "Myocardial_fibrosis", "Cardiomegaly", "Aortic_valve_insufficiency", "Sick_sinus_syndrome", "Left_bundle_branch_block", and many others are members of the subclasses of the class "Heart_diseases".

Class "Directly_HF_related" contains very specific diagnoses related directly to HF, such as "Chronic_heart_failure", "Acute_heart_failure", "Left_ventricular_systolic_dysfunction", "Diastolic_heart_failure", and others.

Some of the most interesting classes in the ontology are "Blood_cell_disorder" and "Circulation_disorder", because heart failure is often in direct relation with the dynamics of the blood flow and with its content. Examples of included concepts are: "Hypovolemia", "Sepsis", "Polycythemia", "Thromboembolic_event" and

"Hemorrhage". However, there are lot of other important blood and circulation related disorders and not all of them could be included in this ontology.

We added many other disorders that are not cardiovascular because they are related to the functioning of the heart, relevant as possible causes of the HF symptoms, or relevant to the treatment of HF. These are included in the class "Related_to_other_organs". Some of the examples are: "Skeletal_muscle_problems", "Anaemia", "Cerebral_hemorrhage", "Drug_abuse", "Pneumonia", etc.

Some of patient statuses that can not be exactly considered as a diagnosis and some known HF syndromes are given in classes "Syndromes" and "Effects". Examples include: "Lack_of_adequate_sleep", "Meningism", "Overeating" and "Reduced_sudden_death". The class "Diagnosis" thus contains many different possible aspects of the heart failure disorder and even a wider range of diseases. Most of the significant diseases which are considered to be relevant to heart problems in any way are members of this class.

3.3.2. Class "Medication"

"Medication" is a subclass of the root class "Treatment" and it contains HF related medications and medication groups. It also contains some of the other generic medications used in treatment of heart related problems, such as medications for atrial fibrillation or high blood pressure. This class is divided into three classes: "Avoid_or_use_with_caution_medications", "Heart_failure_medication_group", and "Other_medications_groups".

Classes for the specific medication group include individual generic medications. Medication groups may also have a specific instance, e.g. "ACE_inhibitors" or "Nitrates" or "Angiotensin_II_receptor_blockers". Individual medications and medication groups contain about a dozen important properties such as properties related to dosage: "InitiatingDose", "TargetDose", "MaximumRecommendedDailyDose", "MaintenanceDose", "SideEffect", "Indicated", "Contraindicated", etc. These properties link medications with signs and symptoms, diagnosis and other medications.

Class, “Avoid_or_use_with_caution_medications,” contains specific medications that should not be prescribed to the patient if the patient has HF, as recommended by chronic heart failure guidelines, such as "Corticosteroids", "Diltiazem" and "Verapamil". [19,23-25]. Finally, "Other_medication_groups" includes those groups of medications and individual medications not directly used in the treatment of HF, but rather in the treatment of the most common comorbidities. Currently, there are a total of 37 medication groups and about 100 individual generic medications in the HF ontology.

3.3.3. Class "Testing"

The sub-class "Test_list" contains a thorough list of tests spanned through four classes: "Echocardiography_tests", "Electrocardiography_tests", "Hematology_and_biochemistry_tests" and "Other_tests". There are seven individual tests under "Echocardiography_tests" as shown in Fig. 1. There are two electrocardiography based tests: "Electrocardiogram_at_rest" (12-lead) and "Holter_electrocardiography_24_hour". There are 27 hematology and biochemistry tests, for example: "C-reactive_protein_test", "Complete_blood_count", "Leukocytes", "Lipid_panel", "S-glucose" etc. "Other_tests" include 18 tests, e.g. "Cardiac_MRI", "Cardiovascular_monitoring", "Chest_CT", "6_minute_walk_test" and "Thoracic_radiography". Each test has its measurements specified in a separate class. All of these classes are placed in the class "Test_measurements", under the class "Testing". Test results relevant for inference of some disorders are placed in the separate class "Relevant_test_results", which enumerates 107 instances. For example: "Cardiothoracic_ratio_greater_than_0.5", "BNP_value_higher_than_100_pg_per_ml", "E_A_ratio_less_than_1" etc.

There also exists a separate class "Normal_ranges" used to specify the normal values, most often in mg/dl or mmol/l, but also in other measure units.

3.3.4. Class "UMLS_syn"

Finally, we consider the class "UMLS_syn", which is a subclass of class "Synonym", subclass of "Terms" and subclass of "HF_concept" (see Fig. 2). Class "UMLS_syn" contains many of the synonyms taken from Unified Medical Language System. In OWL, there can be only one instance with each unique name in the whole ontology despite the fact that there may be many synonyms for any given concept. This is solved by creating instances of significant synonyms in the class "UMLS_syn". Each instance of this class has its name and CUI, which identifies it in UMLS. For example, instance "Dehydration" of the class "Effects" has UMLS synonyms "Exsiccosis" and "Dehydrated" in class "UMLS_syn" and all possess the same CUI: C0011175.

Class "UMLS_syn" differs from a more general class "Synonym" in that an instance located in the class "UMLS_syn" must be found in the UMLS while instances in superclass "Synonym" need not be present in UMLS. An example is the instance "Acute_heart_failure" in the class "Heart failure". It has synonyms: "Decompensated_heart_failure" and "AHF" in class "Synonym" and "Cardiac_failure_acute" in class "UMLS_syn". "Decompensated_heart_failure" and "AHF" do not exist in UMLS (at present), but are important concepts in guidelines for the acute heart failure and are thus added as synonyms for the acute heart failure (although decompensated heart failure is not strictly a synonym for the acute heart failure, it is its most common case).

4. Discussion about applications

The primary application of this HF ontology is in human professional communication [9]. For this purpose it is enough to verify that a term exists in the ontology and that the relations as described by the ontology correspond to our understanding of the underlying concept in a concrete situation. If these conditions are met, then we can be fairly certain that other humans will be able to correctly understand our statements. If a term is not in the ontology or its relations to other concepts are different from those that we assume, a good practice is to specify explicitly the term and its relations with those concepts that are present in the HF ontology. If such a concept and its relations are important for many users, it can be added to revised versions of the ontology.

4.1. Patient data transformation

Very useful is the application of the ontology for systematic and reproducible patient data analysis tasks. For this purpose, an addition of some procedural knowledge for the transformation or abstraction of patient data is necessary. By connecting these procedural relations with the ontology we ensure that implemented relations can be verified and potentially reused on different datasets. This approach is especially relevant for scientific research tasks in clinical studies because it is possible to ensure transparency and reproducibility of the obtained results [7,37]. Currently, the most popular is the use of SWRL for coding procedural relation in the form of rules. Publicly available description logic reasoners such as Pellet [34] can be used for the execution of the data transformation and data preparation process. A schematic illustration of this type of application is presented in Fig. 6. The figure demonstrates that the actual data transformation is performed by the reasoning process performed on patient data previously extracted from patient records and presented in the ontology as factual knowledge. The transformation definitions are in the form of SWRL rules which use concepts defined by the HF ontology.

[Figure 6 about here]

4.2. Decision support

The most sophisticated is the application of the HF ontology in intelligent decision support systems (DSS). In this type of application, a complete expert system for patient related warnings, suggestions, and/or decisions may be implemented.

The starting point is descriptive knowledge about the medical domain (HF ontology) into which procedural knowledge (also called actionable knowledge) presented in the form of rules is added [38]. The rules connect some patient properties with relevant conclusions. An example is the rule for systolic HF which states:

Patient has systolic HF if

a) he has performed echocardiography and

b) has either decreased left ventricular contractility or left ventricular ejection fraction below 40% and

c) has some HF signs or symptoms.

SWRL may be used for the presentation of these rules in the same way as described in the data transformation applications. Because of such integration, the procedural knowledge uses the taxonomy of the HF ontology and, what is even more relevant, it may use information from the descriptive knowledge part. An example is that in the case of the rule for systolic HF we do not need to list all HF signs and symptoms in this rule. We only need to test if a patient has any instance from the classes defining HF signs and HF symptoms. Another example is that in the HF ontology there are classifications of medications and their initiating and target doses. This information may be used to implement rules that warn about potentially conflicting situations when there are two instances from the same class for the same patient (for example in the class "Angiotensin_II_receptor_blocker"), to suggest initiating dose when introduction of a medication is suggested, and to warn when current dose is higher than the target dose.

There are two drawbacks that we are aware of when using SWRL as a rule-based system. The first one is that the Pellet reasoner supports only decidable (DL-safe) SWRL rules. This limits the application of rules to only those instances that are contained in the ontology. A solution is a translator between a database and the ontology that transforms database entries into a set of allowed instances. The second problem with SWRL is that negation-as-failure is not supported. This problem can be solved by applying quantifiers on the properties or by introducing instances representing negative results.

In the realized HF expert system for the Heartfaid platform, procedural knowledge is organized in eight groups of rules including: diagnosis, alternative diagnosis, severity assessment, prognosis, medication prescription and medication related warnings, and acute decompensation detection. In total, the expert system consists of about 200 rules that are in form similar to the presented rule for systolic HF.

The third necessary component of an expert system, besides descriptive and procedural knowledge, is factual knowledge about real patient data. The data are extracted from patient records and inserted into the ontology in the form of instances into the class "Patient". The data may be categorical (like "Patient_performed_echo" or "Patient_has_low_EA_ratio") or they may be numerical (like "Measured_ejection_fraction" equals 30%). In the latter case additional SWRL rules are used to generate categorical instances necessary for decision support tasks (like "Ejection_fraction_above_40", or "Ejection_fraction_30-40"). The conclusions obtained as a result of the reasoning process (like "Patient_has_systolic_HF") are also instances of the class "Patient".

The process and complexity of the transformation of patient data into ontological form of factual knowledge and the process of presentation of results of the reasoning from the ontological form into human interpretable suggestions depend on the particular application. The reasoning part of the expert system may be implemented by general reasoning tools in the same way as for data transformation tasks as shown in Fig. 6. The only differences between the applications for patient data analysis tasks and the decision support applications are that a) the latter have as their input data about one patient only and b) their output is not only the transformed input information but concrete warnings, suggestions, and/or decisions that may be interpreted by humans.

The similarities between these two types of apparently very different applications and the utility of a single HF ontology for both of them, demonstrate the reusability of the implemented ontology. This was a primary motivating factor in choosing to use ontologies as the knowledge-representation method for the EU Heartfaid project. The approach is suggested also for applications in various clinical studies.

In the Heartfaid project we have also made an experiment that is applicable only for decision support applications of ontologies. In this experiment we have introduced actionable subclasses of the class "Patient" that are defined by necessary and sufficient conditions representing rules in the description logic formalism. The approach is illustrated in Fig. 7. The motivation has been to construct a hierarchy of actionable classes in a way similar to the ontology for descriptive knowledge. The goal has been natural ordering and clustering of rules with the aim to enable more effective maintenance of procedural knowledge and more human intuitive coding of rules than in the implementation using SWRL. In this setting the result of reasoning is not introduction of an additional instance like "Patient_has_systolic_HF" but the result of reasoning is placement of the patient, which is already an existing instance and which is the object of reasoning, into the class "Systolic_positive" that is defined with the rule describing systolic HF patients (Fig. 7). The characteristics and problems related to this approach are the topic of further research and are out of scope of this work.

[Figure 7 about here]

4.3. Applications in other domains

The HF ontology presented in this work has already been used for such applications by Stanford University School of Medicine [34]. In this application, the ontology has been used for the research in the field of etiology and risk factors of the nonischemic heart failure. For this application three different knowledge bases have been developed on the top of the HF ontology: a HF pathophysiology knowledge base, a diagnostic criteria knowledge base, and a nonischemic HF etiology knowledge base. These knowledge bases are then applied to patients' clinical data using SWRL for patient phenotype classification, data set generation, and hypothesis validation and discovery in the framework as presented in Fig. 6.

5. Related work

This section gives an overview of several recently proposed approaches to ontology-based, domain-specific modeling in cardiovascular medicine [8,20,21].

The work by Eccher et al. [8] promotes a very flexible architecture for supporting a health care process and its interface with medical knowledge bases for the case of HF. Also interesting is the approach for building the ontology that starts with archetypes that are developed for the concrete application, which are then integrated into general reference ontology of medicine (DOLCE). The work performed by Esposito [20] deals with ontology-based reasoning applied to patients with congenital heart disease (CHD). By using some specific parts of the SNOMED medical terminology, the author managed to construct a small domain-specific ontology used to detect congenital malformations of the heart and of its blood vessels.

The authors Chiarugi et al. [21] recently proposed a DSS for heart failure patients' management based on the knowledge acquired through continuous collaboration on the Heartfaid project. The smaller ontologies used by the DSS were developed for the purpose of faster computer reasoning as well as for easier maintenance. The authors designed the DSS in such a way that it takes signal and image processing patient data, stores them in a database, transforms and imports the data in the ontologies by using Jena framework, reasons on the data by using Pellet reasoner, and displays various suggestions to medical personnel.

6. Conclusions

In our work we have been confronted with many dilemmas and the current version of the HF ontology may be interesting also as a prototype for building similar medical ontologies for other domains. With the discussion on potential benefits of using ontologies, we also try to motivate the medical community for stronger participation in building and refinement of medical ontologies. Nobody can better define the ontology than the HF experts, and the heart failure community can significantly profit if both technical systems and humans in their communication start to use this standard.

This HF ontology has been developed mainly by technical people by reading medical literature, primarily HF guidelines published by European Society of Cardiology. In this sense the current version is more an effort to demonstrate how a useful ontology may look like than the final product. In order to stimulate its application and its further development, the complete HF ontology is made public. Constructive criticism may help us to improve the ontology iteratively. How the process of the ontology maintenance should be organized in order to ensure its public relevance and constant improvements at the same time, is still an open issue.

By connecting the procedural tasks with the ontology as part of data preprocessing, we ensure transparency, consistency, and reusability of the procedures, characteristics that are important for scientific research tasks and medical trials. Integration of procedural knowledge (rules) with ontological representation of domain related descriptive knowledge in decision support tasks enables direct use of descriptive knowledge in the decision making process. A nice property of such integration is that the reasoning result may change automatically when the knowledge present in the HF ontology is updated. The approach has been demonstrated as useful within the Heartfaid platform and is suggested also for other similar projects.

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Figure captions

Fig. 1. Instances in class "Echocardiography_tests" and some properties of the instance "Doppler_echocardiography". It can be seen that the instance has CUI and "Definition" as obligatory parts and some other properties like "Measures" and "CanDetect".

Fig. 2. The "HF_concept" super-class includes following subclasses: "Terms", "CHF_risks", and "Classification".

Fig. 3. "Patient_characteristics" super-class consists of: "Demographic_characteristics", "Diagnosis", "Other_patient_characteristics", and "Signs_and_symptoms". Each of these classes includes many different concepts.

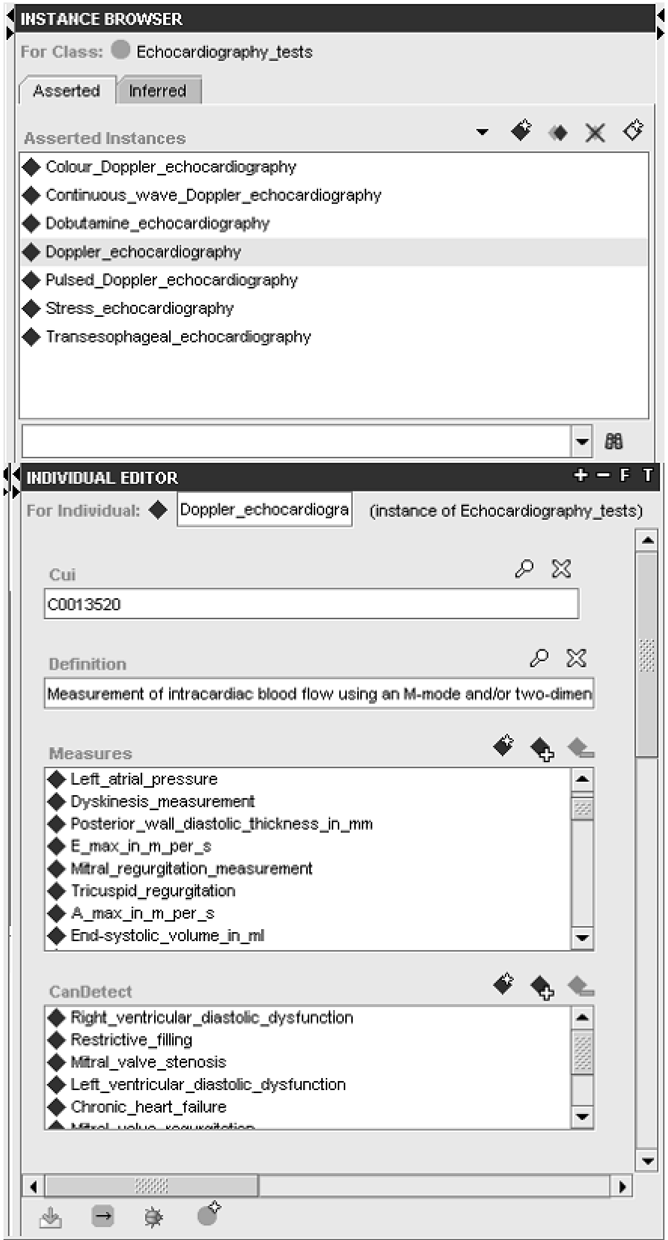
Fig. 4. The hierarchy of the class "Testing".

Fig. 5. Super-class "Treatment" consists of three sub-classes: "Additional_therapy", "Medical_devices_and_surgical_procedures", and "Medication". This last one is relevant because of the information about many specific medications.

Fig. 6. Application of the ontology for systematic and reproducible patient data analysis tasks.

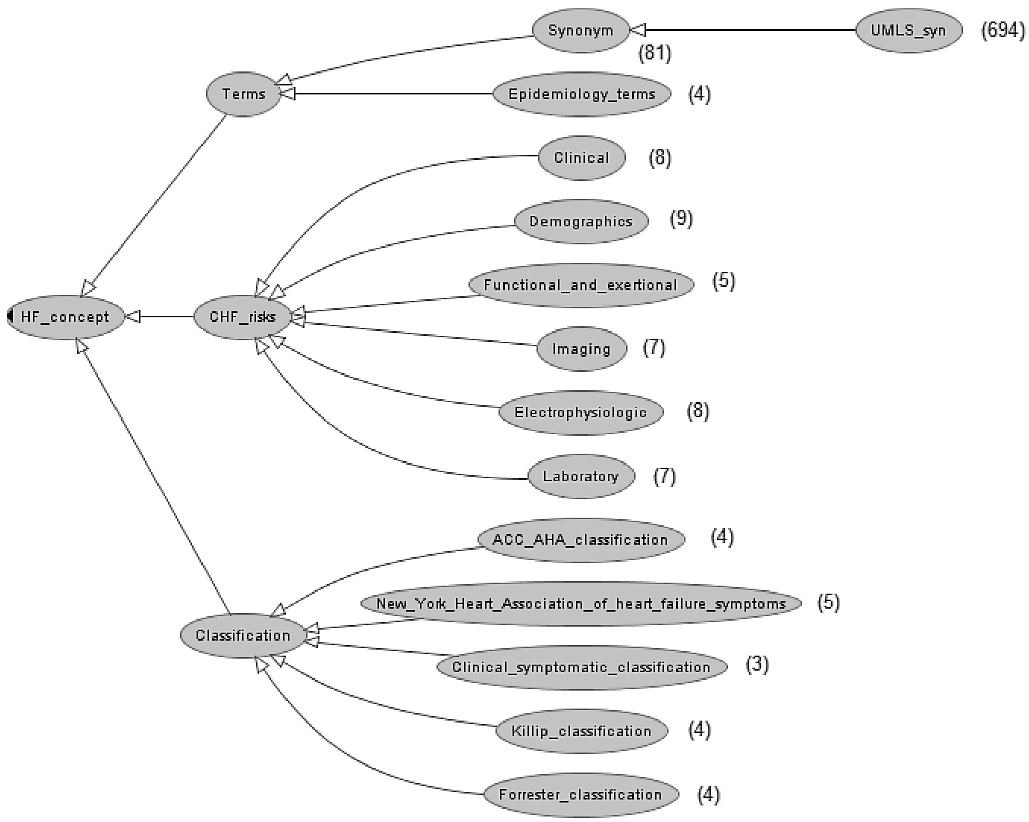
Fig. 7. Procedural knowledge integrated into HF ontology as actionable classes for the implementation of the Heartfaid expert system. The central part of the figure presents definition of a rule for the diagnosis of the systolic heart failure by using description logic. The left part of the figure presents eight super-classes in which actionable knowledge is included in a hierarchical order.

Figure 1



INSTANCE BROWSER
For Class: Echocardiography_tests
Asserted
Inferred
Asserted Instances
Colour_Doppler_echocardiography
Continuous_wave_Doppler_echocardiography
Dobutamine_echocardiography
Doppler_echocardiography
Pulsed_Doppler_echocardiography
Stress_echocardiography
Transesophageal_echocardiography
INDIVIDUAL EDITOR
For Individual: Doppler_echocardiogra
(instance of Echocardiography_tests)
Cui
C0013520
Definition
Measurement of intracardiac blood flow using an M-mode and/or two-dimen
Measures
Left_atrial_pressure
Dyskinesis_measurement
Posterior_wall_diastolic_thickness_in_mm
E_max_in_m_per_s
Mitral_regurgitation_measurement
Tricuspid_regurgitation
A_max_in_m_per_s
End-systolic_volume_in_ml
CanDetect
Right_ventricular_diastolic_dysfunction
Restrictive_filling
Mitral_valve_stenosis
Left_ventricular_diastolic_dysfunction
Chronic_heart_failure

Figure 2



Synonym
(81)
UMLS_syn
(694)
Terms
Epidemiology_terms
(4)
Clinical
(8)
Demographics
(9)
Functional_and_exertional
(5)
HF_concept
CHF_risks
Imaging
(7)
Electrophysiologic
(8)
Laboratory
(7)
ACC_AHA_classification
(4)
New_York_Heart_Association_of_heart_failure_symptoms
(5)
Classification
Clinical_symptomatic_classification
(3)
Killip_classification
(4)
Forrester_classification
(4)

Figure 3


Patient_characteristics
Demographic_characteristics
Age_group (7)
Employment_status (3)
Gender (2)
Marital_status (4)
Racial_group (4)
Diagnosis (2)
Cardiovascular_system_related
Blood_cell_disorder (16)
Circulation_disorder (15)
Directly_HF_related (2)
Heart_diseases (4)
Effects (36)
Related_to_other_organs
Bone_or_muscular_diseases (6)
Brain_nervous_system_and_mental_disorders (27)
Gastroenterological_disorder (6)
Kidney_diseases (13)
Liver_diseases (6)
Malignant_neoplasms (9)
Nutritional_disorder (7)
Other_disorder (33)
Pulmonary_diseases (47)
Thyroid_disorder (3)
Syndromes (4)
Other_patient_characteristics (14)
Health_status_type (2)
Medical_attention (9)
Patient_condition (4)
Patient_feeling (2)
Physical_activity (5)
Smoking_alcohol_and_drugs_status (7)
Treatment_outcome (9)
Prognosis (5)
Signs_and_symptoms (4)
Signs
Symptoms (25)

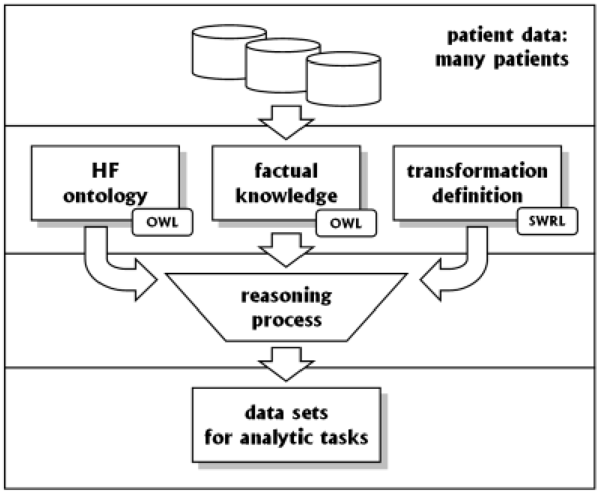
Figure 4

- Testing
  - Normal_ranges (33)
  - Physical_examination (1)
  - Relevant_test_results (107)
  - Test_characteristics (3)
  - Test_list
    - Echocardiography_tests (7)
    - Electrocardiography_tests (2)
    - Hematology_and_biochemistry_tests (27)
    - Other_tests (18)
  - Test_measurements (2)
    - Cardiac_magnetic_resonance_imaging_measurements (9)
    - Cardiac_output_measurements (1)
    - Cardiopulmonary_stress_test_measurements (8)
    - Chest_X-ray_measurements (12)
    - Echocardiography_measurements (24)
    - Electrocardiography_measurements (13)
    - Exercise_test_measurements (14)
    - Hematology_and_biochemistry_measurements (34)
    - Measurements_obtainable_from_more_than_one_test (3)
    - Natriuretic_peptides_measurements (2)
    - Physical_examination_measurements
    - Pulmonary_function_tests_measurements (3)
    - Radionuclide_angiography_measurements (3)
    - Six-minute_walk_test_measurements (3)

Figure 5

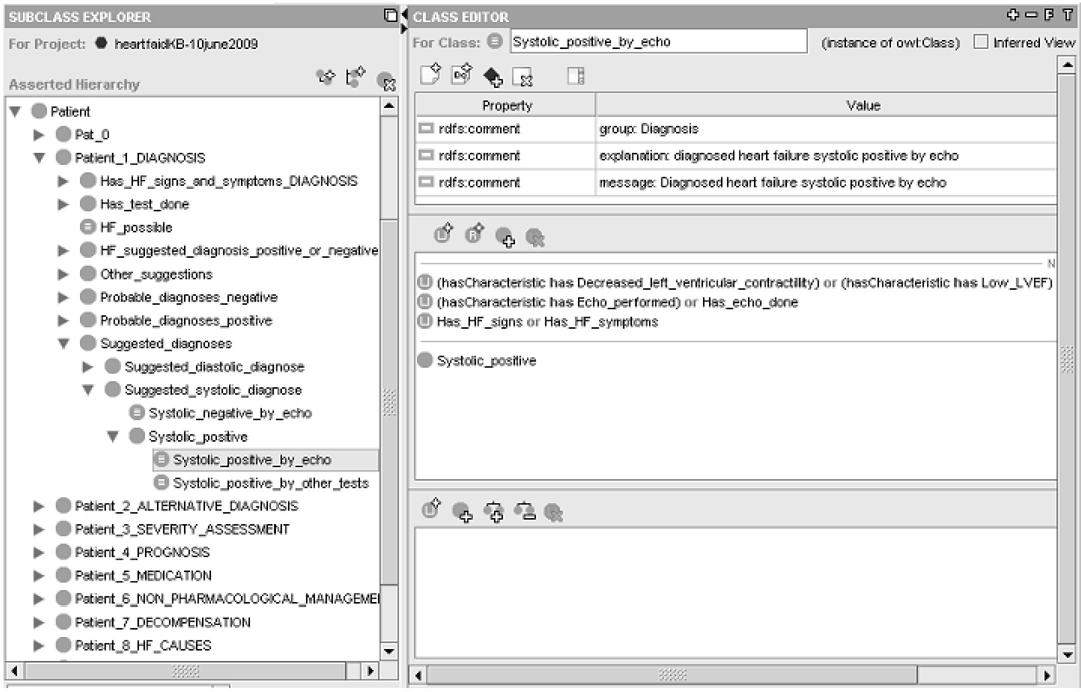
- Treatment (3)
  - Additional_therapy (21)
  - Medical_devices_and_surgical_procedures
    - Medical_device (12)
      - Artificial_cardiac_pacemaker (3)
      - Cardiac_resynchronization_therapy (2)
    - Surgical_procedure (5)
      - Heart_valve_surgery (4)
      - Left_ventricular_restoration (4)
      - Revascularization (3)
  - Medical_procedure (13)
  - Medication
    - Avoid_or_use_with_caution_medications (9)
    - Heart_failure_medication_group (20)
      - ACE_inhibitor (5)
      - Adrenergic_beta_antagonist (8)
      - Aldosterone_receptor_antagonist (2)
      - Angiotensin_II_receptor_blocker (6)
      - Antiarrhythmic_agents
      - Calcium_antagonist (2)
      - Cardiac_glycoside (1)
      - Diuretics (9)
      - Fibrinolytic_agent (4)
      - Inotropic_agent (9)
      - Oxygen_therapy (1)
      - Statin_agents (1)
      - Vasodilator_agent_and_nitrate (5)
    - Other_medications_groups (17)
      - Anticholinergics (1)
      - Antidiabetic_treatment (3)
      - Antihistamines (1)
      - AVP_receptor_antagonists (2)
      - Specific_medications (23)
      - Steroids (1)
      - Toxins (7)
  - Patient_education (12)
  - Recommendations (15)

Figure 6



patient data:
many patients
HF
ontology
OWL
factual
knowledge
OWL
transformation
definition
SWRL
reasoning
process
data sets
for analytic tasks

Figure 7


SUBCLASS EXPLORER
For Project: heartfaidKB-10june2009
Asserted Hierarchy
Patient
Pat_0
Patient_1_DIAGNOSIS
Has_HF_signs_and_symptoms_DIAGNOSIS
Has_test_done
HF_possible
HF_suggested_diagnosis_positive_or_negative
Other_suggestions
Probable_diagnoses_negative
Probable_diagnoses_positive
Suggested_diagnoses
Suggested_diastolic_diagnose
Suggested_systolic_diagnose
Systolic_negative_by_echo
Systolic_positive
Systolic_positive_by_echo
Systolic_positive_by_other_tests
Patient_2_ALTERNATIVE_DIAGNOSIS
Patient_3_SEVERITY_ASSESSMENT
Patient_4_PROGNOSIS
Patient_5_MEDICATION
Patient_6_NON_PHARMACOLOGICAL_MANAGEME
Patient_7_DECOMPENSATION
Patient_8_HF_CAUSES
CLASS EDITOR
For Class: Systolic_positive_by_echo
(instance of owl:Class)
Inferred View
Property
Value
rdfs:comment
group: Diagnosis
rdfs:comment
explanation: diagnosed heart failure systolic positive by echo
rdfs:comment
message: Diagnosed heart failure systolic positive by echo
(hasCharacteristic has Decreased_left_ventricular_contractility) or (hasCharacteristic has Low_LVEF)
(hasCharacteristic has Echo_performed) or Has_echo_done
Has_HF_signs or Has_HF_symptoms
Systolic_positive