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Ontological Engineering in Medical Informatics

Review Article

Abdel-Badeeh M. Salem*, Mostafa M. Aref, Moawad IF, Marco Alfonse, Galal AL Marzoqi

Faculty of Computer and Information Sciences, Ain Shams University, Cairo, Egypt.

Abstract

Ontologies were developed in Artificial Intelligence (AI) to facilitate knowledge sharing and reuse. Since the beginning of the nineties ontologies have become a popular research topic investigated by several AI research communities, including knowledge engineering, natural-language processing and knowledge representation. Ontologies are now ubiquitous in many intelligent information-systems enterprises; they are used in e-health and in various tasks of biological and medical sciences. Ontologies are becoming so popular in large part due to what they promise: a shared and common understanding of some domain that can be communicated between people and application systems. This paper presents some examples of the developed ontologies by the author and his colleagues at Medical Informatics and Knowledge Engineering Research Labs, Ain Shams University, Cairo, Egypt.

Keywords: Medical Knowledge Management; Ontological Engineering; Medical Informatics; e-Health.

Ontological Engineering

Ontological Engineering (OE) refers to the set of activities that concern the ontology development process, the ontology life cycle, the methods and methodologies for building ontologies, and the tool suites and languages that support them. During the last decade, increasing attention has been focused on ontologies [1]. The main objective of using ontologies is to share knowledge between computers or computers and human. Computers are capable to transmit and present the information stored in files with different formats, but they are not yet compatible to interpret them.

To facilitate communication and intelligent processing of information, it is necessary that all actors of the digital space (computers and humans) have the same vocabulary. Ontologies are the foundation of cooperation and the semantical understanding between computers (running a lot of non-homogenous software programs) and of the cooperation between computers and humans.

Medical informatics is concerned with the development of the science and technologies needed for collecting, sharing, reporting, analyzing, and visualizing medical data; and for providing decision-making support for disease prevention, detection, and management. However, the lack of standard vocabulary of medical data hinder effective classification. To meet this challenge, an appropriated way to describe these data is required. Many alternatives have been developed, however, there is not a consensus to use one or a small set of them yet. On the other side, medical ontology provides a common vocabulary for young physicians and researchers who need to share information in specific domain. Some of the reasons to create an ontology are, (1) to share common understanding of the structure of information among people or software agents, (2) to enable reuse of domain knowledge, (3) to make domain assumptions explicit, (4) to separate domain knowledge from operational knowledge and (5) to analyze domain knowledge. Ontologies may be categorized according to the domain they represent. General ontologies represent knowledge at an intermediate level of detail independently of a specific task. Domain ontologies represent knowledge about a particular part of the world. Finally, ontologies designed for specific tasks are called application ontologies. At present, there are many applications in biomedical domain [2, 3]. From the artificial intelligence perspective OE includes the following technical aspects; (a) methodology for the design

***Corresponding Author**:
Abdel-Badeeh M. Salem,
Faculty of Computer and Information Sciences, Ain Shams University, Cairo, Egypt.
E-mail: absalem@cis.asu.edu.eg

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and evaluation of ontologies, (b) intelligent development tools, (c) ontologies integration and validation techniques, and (d) ontologies merging and mapping. For more technical details we refer to the books [4, 5].

Intelligent Meical Informatics

Based on our analysis of the research work published in the scientific journals and international conferences which held during the last ten years, Figure 1 shows the main areas of research of the intelligent medical informatics as a since and technology. From this figure it can be seen that the research in the field consists of five main areas, namely: Robotic Surgery, Medical Imaging, Medical Knowledge Engineering, Medical Expert Systems, and Medical Education and Training.

Also, it can be seen that, the roots/concepts/theories of such interdisciplinary field/tree are; medical sciences, informatics and artificial intelligence. Moreover the figure shows some points and applications of research in this respect.

The Proposed Methodology of Developing Medical Ontology

Organizing and scoping: establishes the objectives and requirements. The scope defines the boundaries of the ontology.
Data collection: the raw data needed for ontology development is acquired.
Data analysis: the ontology is extracted from the results of data collection process. In this step, we perform the following tasks; (a) define the classes and class hierarchy, (b) the objects of interest in the domain are listed, followed by identification of objects on the boundaries of the ontology, (c) identification of the relations between objects, and (c) creation of the individual instances of the classes.
Initial ontology development: developing the preliminary ontology (i.e. initial implementation).
Ontology refinement: the initial development is iteratively refined.

Developing of Web-Based Liver Cancer Ontology

Liver cancer is the third most common cancer in the world. Liver cancer is much more common in developing countries within Africa and East Asia. In some countries, it is the most common cancer type [6, 7]. This section presents the process of building a web-based liver cancer ontology. The main goals behind building this ontology are to allow finding and locating information about liver cancer needed for interested users and domain experts and providing a semantic representation of liver cancer information over the web.

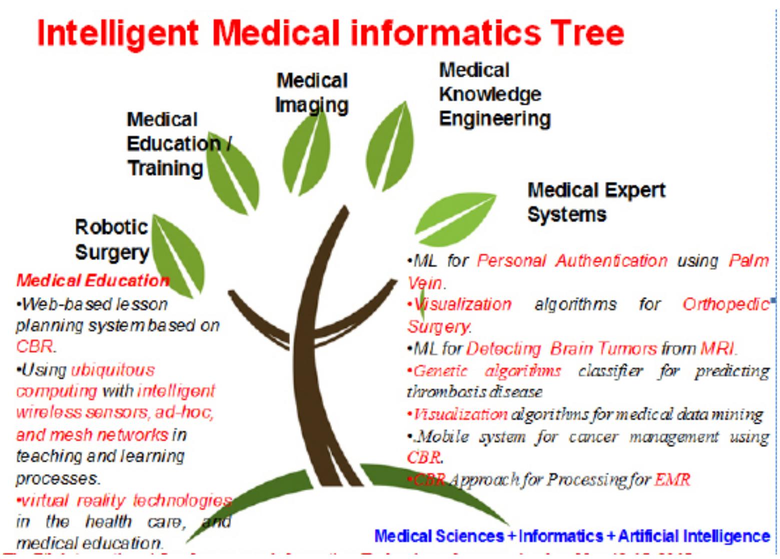
Knowledge Acquisition Process

Cancer.Net: Cancer.Net provides timely, oncologist-approved information to help patients and families make informed health care decisions [8].
MedicineNet: MedicineNet.com is an online, healthcare media publishing company. It provides easy-to-read, in-depth, authoritative medical information for consumers via its robust, user-friendly, interactive website [9].
The National Cancer Institute (NCI): NCI is part of the National Institutes of Health [10].

Building the Liver Cancer Ontology

Liver cancer ontology was built by Protégé-OWL editor [10-12] and in [13] you will find the details of the technical and computing aspects about the developing tasks. The subtypes of primary liver cancer are named for the type of cell from which they develop. There are three subtypes of primary liver cancer namely, Hepatocellular carcinoma, Cholangiocarcinoma, and Angiosarcoma. For more information about the medical description see [10]. The liver cancer is described in terms of its risk factors, symptoms, diagnosis, staging and treatment. Staging is a way of describing a cancer, such as where it is located, if or

Figure 1. The main areas of Intelligent Medical Informatics.



where it has spread, and whether it is affecting the functions of other organs in the body. One tool that doctors use to describe the stage is the TNM system.

Liver Cancer Ontology Classes

Figure 2 shows the class hierarchy of the liver cancer ontology. From this figure it can be seen that, the liver cancer ontology has three main classes; Disease, Medical_Intervention and References. The Disease class contains the LiverCancer class with its types. The Medical_Intervention class contains the Staging, Diagnosis and Treatment classes. The References class contains the Symptoms, Stage, Risk_Facors and TNM_System classes.

Instances of the Classes of Liver Cancer Ontology

In the liver cancer ontology, the diagnostic tools of the liver cancer are described as instances of the class Diagnosis. Figure 3 shows the instances of classes as described in the liver cancer ontology. The classes M, N and T are defined as enumerated classes. Each one of these classes is described in terms of its instances (no more instances can be added to any of these classes).

Web-Based Breast Cancer Ontology

The breast cancer ontology was encoded in OWL-DL format using the Protégé-OWL editing environment. The knowledge was collected from: MedicineNet, World Health Organization, breastcancer.org, ehealthMD and National Comprehensive Cancer Network [14]. Figure 4 shows the developed ontology. From this figure it can be seen that, this ontology consists of two main super classes namely; (a) "MedicalThings" which has the following four sub-classes; Diseases, Medical_Interventions, Pathological_ Category, References, and (b) "People" which has the sub classes; men and women. The main benefits from this ontology are to allow finding and locating information about breast cancer needed for interested users and domain experts.

Viral Hepatitis (VH) Ontology

In this application, the proposed methodology includes the following three phases.

The VH Ontology Extraction Phase

In this phase, we have extracted the needed knowledge from several medical sources such as domain experts, medical books, and a set of trusted medical websites [15, 16]. The bottom-up approach has been followed in this research to build the VH Ontology. In this respect there are 18 distinct symptoms and 15 distinct signs caused by the Viral Hepatitis diseases (A, B, C and D). Also, there are 16 laboratory-findings can be used to differentiate between those Viral Hepatitis diseases In our analysis, we have classified the Viral Hepatitis results into three main classes: "Symptoms", "Signs" and "Lab Result", where each main class is then have classified into subclasses a shown in Figure 5.

The VH Ontology Validation Phase

In this phase, the domain experts have been consulted to review the results of both VH Diseases and Symptoms/Signs/Laboratory-findings Mapping step and VH Disease and Symptoms/Signs/

Figure 2. The Liver Cancer Class Ontology.

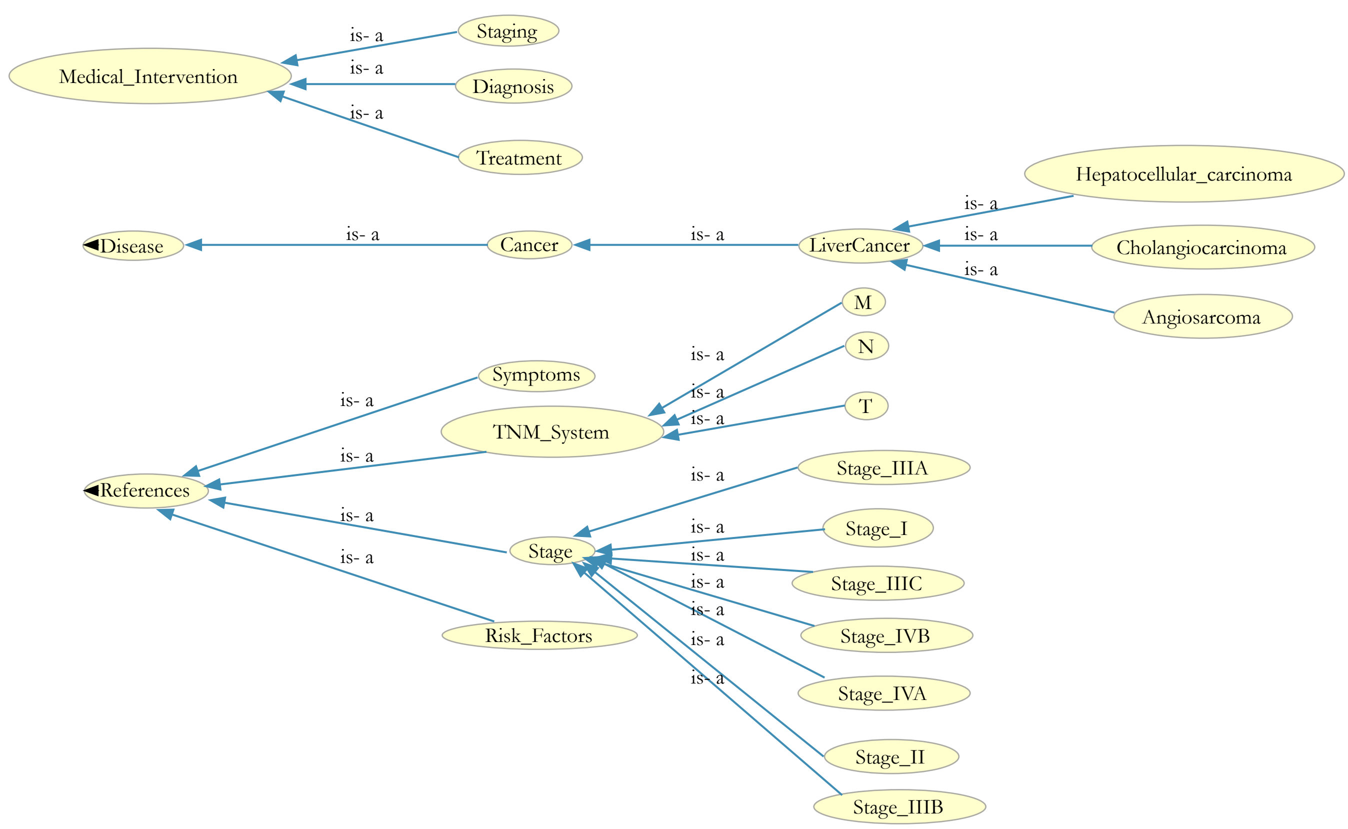


Figure 3. The Instances of the Classes of the Liver Cancer Ontology [13].

Diagnosis: Angiogram, Biopsy, Blood_tests, CT_scan, MRI, Physical_exam, Ultrasound_test
Staging: Bone_scan, CT_scan_of_the_chest, PET_scan
Treatment: Ablation, Chemoembolization, Chemotherapy, Cryoablation,Hepatectomy, Hepatic_arterial_infusion, Targeted_Therapy, Immunotherapy_therapy, Liver_transplant, Proton_beam_therapy, Radiation_Therapy, Radioembolization, Stereotactic_radiosurgery, Surgery
Risk_Factors: Aflatoxin, Alcohol, Cirrhosis, Hemochromatosis, Hepatitis_B, Hepatitis_C, Iron_storage_disease, Obesity_and_diabetes
Symptoms: A lump or a feeling of heaviness in the upper abdomen, Fever, Loss of appetite and feelings of fullness, Nausea and vomiting, Weight loss, Pain in the upper abdomen on the right side, Swollen abdomen, Weakness or feeling very tired,
T: TX, T0, T1, T2, T3a, T3b, T4
N: NX, N0, N1
M: MX, M0, M1

Figure 4. The Developed Breast Cancer Ontology.

Laboratory-findings Classification step. The domain experts have validated the classification trees generated from those steps by editing some of terminologies, and by rephrasing some of the classes' names. Also, they have added other classes to those classification trees to be fit in the OBR framework.

The VH Ontology Representing in OWL Phase

To develop the Viral Hepatitis ontology in OWL, two processes have been performed: (a) representing the classes and relations of the Viral Hepatitis Result Classification Tree in OWL, and (b) representing the classes and relations of the merged Viral Hepatitis Diseases Classification tree with OBR in OWL. The protégé-OWL editor has been exploited to implement this phase (see Figure 6).

Conclusion

Ontological engineering offers a promising way to develop efficient medical knowledge-based systems capable to facilitate knowledge sharing, refine, search, and reuse. This paper presents the process of developing three biomedical ontologies, namely; a liver cancer ontology, breast cancer ontology, and Viral Hepatitis A, B, C and D virus's ontology. These ontologies were built using the Protégé-OWL editing environment and encoded in ontology web-based language, OWL-DL. These ontologies can be used by experts or medical researchers who want the cancer knowledge to

Figure 5. The Viral Hepatitis Result Classification Tree.

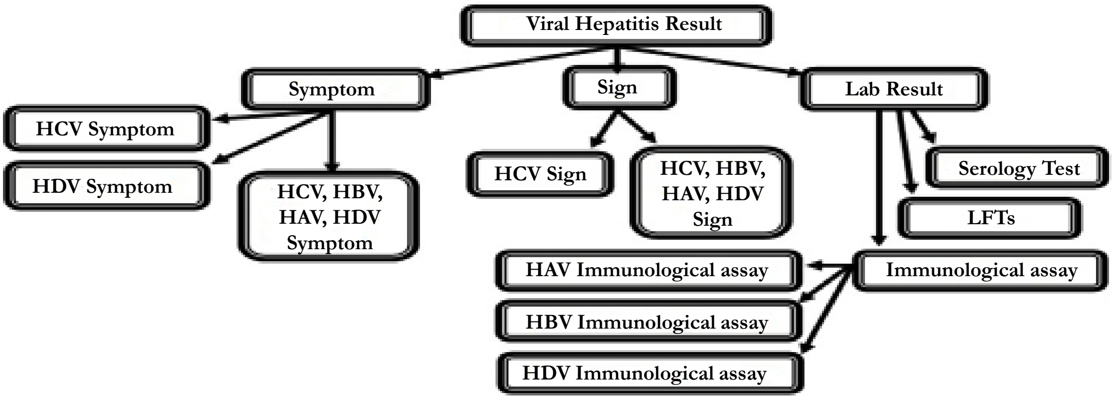
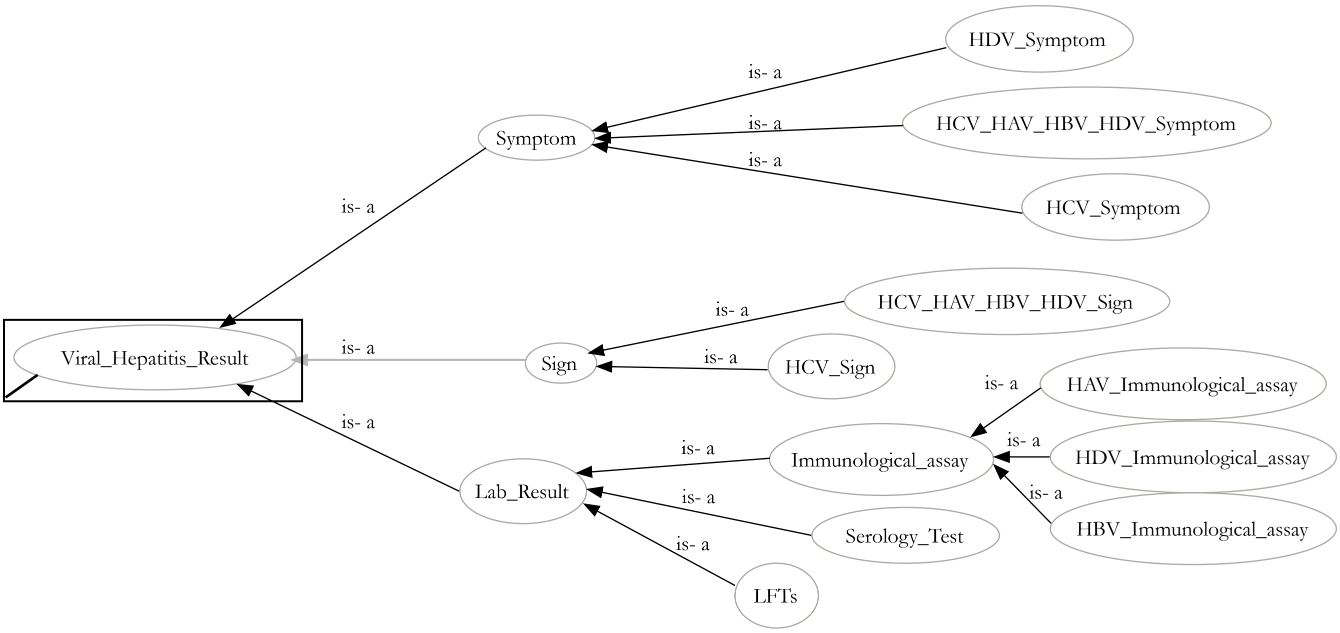


Figure 6. The OBR Viral Hepatitis Diseases Ontology.



be represented in a semantic way that allows reasoning capabilities.

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