

Chapter 28

HOD²MLC: Hybrid Ontology Design and Development Model With Lifecycle

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ABSTRACT

Ontologies have gained increasing usage to augment an application with domain knowledge, particularly in healthcare, where they represent knowledge ranging from: bioinformatics data such as protein, gene, etc. to biomedical informatics such as diseases, diagnosis, symptoms, etc. However, the current ontology development efforts and process are data intensive and construction based, creating ontologies for specific applications/requirements, rather than designing an abstract ontological solution(s) that can be reusable across the domain using a well-defined design process. To address this deficiency, the work presented herein positions ontologies as software engineering artifact that allows them to be placed into the position to share the captured domain conceptualization and its vocabulary involving disparate domain backgrounds, that can then be created, imported, exported and re-used using different frameworks, tools and techniques. Towards this end, the authors propose an agile software process for ontologies referred to as the Hybrid Ontology Design & Development Model with Lifecycle, HOD²MLC. To place HOD²MLC into a proper perspective, they explore, compare, and contrast it to existing ontology design and development alternatives with respect their various phases as related to the authors' work and phases in varied SDP models.

1. INTRODUCTION

An ontology is a compilation of *concepts* that can be represented as a class composed of *attributes* coupled with *relationships* or *associations* that define interactions between the classes and augmented with *axioms* which are rules or constraints on classes, attributes, or relationships. Ontologies are pro-

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moted as a part of the Semantic Web to attach knowledge to information thereby aiding users (humans and agents) in various ways. Ontologies are heavily utilized in research such as knowledge engineering and representation, domain modeling (Saripalle, Demurjian, & Behre, 2011), database integration (Konstantinou, Spanos, & Mitrou, 2008), natural language processing (Kaihong, Hogan, & Crowley, 2011), etc. For our purposes, we have focused on clinical informatics that involves medical organizations (e.g., private practices, hospitals, etc.) that represent, store patient data (e.g., demographics, diagnosis, vitals, etc.) electronically using standards such as Continuity of Care Record (CCR)¹, Health Language Seven (HL7) Standard Clinical Document Architecture (CDA) (Boone, 2011), etc. and share the data in the form of Electronic Health Record (EHR) or Personal Health Record (PHR). As shown in the top portion of Figure 1, EHRs, PHRs, and other health IT systems are brought together by Health Information Exchange (HIE) (Demurjian, Saripalle, & Behre, 2009) (promoted by the HITECH Act²) into a conceptual setting called the Virtual Chart (VC) which provides a common global view for all potential users. For semantic knowledge, each of these systems employs standard medical ontologies such as: Logical Observation Identifiers Names and Codes (LOINC)³ a standard for identifying medical laboratory observations; International Classification of Disease (ICD)⁴ to hierarchically organize medical concepts such as diseases, symptoms, injuries, procedure, etc., Unified Medical Language System (UMLS) (Bodenreider, 2004) aggregation of medical ontologies and terminology such as ICD, LOINC, SNOMED, etc.; and have a semantic network and metathesaurus. The main objective of our work is to achieve interoperability among the health systems by unifying not only the data, but most importantly, unify their ontologies through their schema or model integration, as shown in the third horizontal box from the top of Figure 1.

As we seek to achieve this interoperability, we have identified a number of problems that significantly affect the process. First, individual EHRs or healthcare applications are built using different ontologies organizing domain knowledge in different ways to suit the specific business and medical requirements. This current approach is often an ad-hoc process, with minimal attention given to process and to a consideration of the impact of the ontology on other software components or ontologies. As a result, the developed ontologies are often incompatible and difficult to integrate. For example, UMLS Metathesaurus is the most comprehensive effort for integrating independent/disparate medical ontologies. The integration of a new ontology into UMLS is ad-hoc and exhaustive process that includes the following steps: automatic techniques - mostly a combination of natural language processing, corpus processing, linguistic analysis, etc., expert assessment – human intervention mostly for validating automatic techniques, and auditing protocols- for verifying the correctness of the resulting system. Second, the existing ontology building process is predominantly data intensive and construction based, often dictated by the talent and expertise of the designer rather than using any concrete well-defined process. For example, UMLS and FMA (Formal Model of Anatomy) comprise of millions of concepts and relationships, where the focus is on the acquiring the domain data rather than an ontological model that is reusable, modular and importantly, that describes the data. Third, the current ontology development processes and knowledge representational frameworks lack an ability to design solutions that are broader in scope i.e. they have a convoluted design and development processes that are clearly disjoint in a conventional software modeling approach (Kuhn, 2010; Guizzardi, 2010). Gonzalez-Perez and Henderson-Seller also state that for achieving interoperable ontological systems, the ontological domain must have an approach that executes ontology modeling and development process simultaneously (Gonzalez-Perez & Henderson-Seller, 2006). This further supports our statement on ontology design/modeling and its existing development methodologies.

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