ABSTRACT

The UMLS is a standard medical vocabulary developed by aggregating standard terminologies and sources from diverse healthcare domains, which encapsulates a broad spectrum of medical knowledge. The two primary knowledge sources of the UMLS are semantic network (SN) and metathesaurus (META). In the present state, the impact of the semantic knowledge captured by UMLS-SN on healthcare application is minimal or has no impact, as UMLS confines the UMLS-SN knowledge. Further, healthcare application design and development is achieved primarily by using sound software modeling principles and established object-oriented (OO) frameworks and languages. The goal of this chapter is to translate UMLS-SN knowledge into an OO modeling framework to utilize SN’s knowledge in designing healthcare application. The result is the design and development of UMLS-SN, a custom metamodel by leveraging UML profile and metamodel. This specialized metamodel allows developers to design healthcare applications that are coherent with object-oriented modeling and software engineering approaches.

1. INTRODUCTION

For the past decade, semantic knowledge (Guranio, 2001) sources great influenced the domain of biomedical and healthcare informatics to achieve unambiguous knowledge interpretation and interoperability among heterogeneous healthcare systems, which has led to the development of various standard medical knowledge standards. For example, the International Classification of Diseases (ICD) hierarchically organizes various medical concepts in the domains such as diseases, symptoms, injuries, and procedures. Systematized Nomenclature of Medicine Clinical Terms (SNOMED-CT) is a computer-readable ontology of medical terms and instances providing codes, terms, synonyms and definitions covering medical domains such as diseases, findings, procedures, microorganisms, and substances and Diagnostic and
Statistical (DSM) offers a common vocabulary and standard criteria for the classification of mental disorders. They are more than forty medical knowledge standards across the globe that capture semantics of various aspects of healthcare.

These medical standards have moved the healthcare informatics community towards uniform semantic interpretation and eased semantic interoperability issues when healthcare data is exchanged across heterogeneous healthcare systems. However, this has resulted in interoperability issues between the standards themselves. This is due to the unique way the semantic knowledge is designed, structured and expressed in the individual knowledge standard. To address the interoperability issues between the standards and continue to capture medical knowledge, the National Library of Medicine has initiated the Unified Medical Language System (UMLS) (Browne, Divita, & McCray, 2003; Bodenreider, 2004; UMLS, 2009). The UMLS system is formed by aggregating millions of medical concepts from diverse medical knowledge standards (e.g., ICD, LOINC, SNOMED-CT, NCBI, etc.) and sources. The UMLS provides mappings (equivalence, parent, child, etc.) structure between the aggregated concepts and this mapping is leveraged to ease interoperability issues between medical standards. The UMLS system is comprised of three components: Semantic Network (SN), Metathesaurus (META) and Specialist Lexicon (SL). The UMLS-SN is formed by connecting defined semantic types with semantic relationships and UMLS-META comprises of millions of medical concepts obtained by aggregating various medical standards and is the heart of the UMLS.

The primary purpose of UMLS-SN is to classify the medical concepts captured in the UMLS-META. For example, the concepts such as Coughing, Redness of Eye, and Wheezing are of type Finding; Myocardial Infarction and Diabetes are of semantic type Disease or Syndrome; and Hematologic Tests and Urinalysis are of semantic type Laboratory Procedure. The terms Finding, Disease of Syndrome and Laboratory Procedure are semantic types. The semantic types are connected using semantic relationships such as co-occurs_with (e.g., Finding co-occurs_with Finding), associated_with (e.g., Findings associated_with Pathologic Function), and so on. However, the success of employing the semantic knowledge captured by UMLS-SN in healthcare application research and development beyond UMLS is minimal due to the following key issues.

- The software modeling principles and metamodels (e.g., UML, ERD, and XML) dominate the application design and development process. The UMLS-SN focuses on classifying medical concepts in UMLS-META, and its scope is restricted to the UMLS environment. There is no acceptable representation (e.g., UML, XML, or ERD) of its structure and semantics that can readily be used for designing health and biomedical application that is consistent with existing best software modeling principles and practices.
- Each medical concept in UMLS-META is assigned to at least one semantic type from UMLS-SN for consistent classification. This process is complex and inconsistent (Kumar & Smith, 2003), as it requires constant expert human intervention. This process is against the principles of software modeling and design.

The software engineering emphasizes on modeling in a way that conceptualizes the problem in a fashion that promotes characteristics such as modularity, abstraction, and reuse using a standard modeling language. Further, the focus is on modeling language, its capabilities and the domain model/schema rather than the actual data. The software design never ties itself to the model data unlike to UMLS, where a medical concept is assigned one or more semantic types. For example, UML (Booch, Rum-