A Standards-based Semantic Metadata Repository to Support EHR-driven Phenotype Authoring and Execution

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Abstract

The objective of the study is to describe our efforts in developing a standards-based semantic metadata repository for supporting electronic health record (EHR)-driven phenotype authoring and execution. Our system comprises three layers: 1) a semantic data element repository layer; 2) a semantic services layer; and 3) a phenotype application layer. In a prototype implementation, we developed the repository and services through integrating the data elements from both Quality Data Model (QDM) and HL7 Fast Healthcare Inteoroperability Resources (FHIR) models. We discuss the modeling challenges and the potential of our system to support EHR phenotype authoring and execution applications.

Keywords:

Metadata Repository; Clinical Phenotyping; Quality Data Model (QDM); HL7 FHIR.

Introduction

The Quality Data Model (QDM) is an information model developed by the National Quality Forum (NQF) and a promising candidate for representing EHR-driven phenotyping algorithms for clinical research [1]. In a previous study, we developed a Semantic Web-based framework that provides a standards-based, semantically annotated, machine-readable rendering of the QDM [2]. In this study, we extend the framework and develop a semantic metadata repository and associated Web services by integrating the data elements from HL7 FHIR models [3]. The purpose of the integration is to provide a more comprehensive coverage of data elements available for clinical phenotype applications.

System Architecture

Our system comprises three layers: 1) a semantic data element repository layer; 2) a semantic services layer; and 3) a phenotype application layer. In the repository layer, we leverage both W3C standards, such as Resource Description Framework (RDF) and Web Ontology Language (OWL), and the meta-data standard ISO 11179 [4] to describe the QDM reference model, data model elements and logic elements.

Prototype Implementation

In our previous study [2], we developed a QDM schema in OWL representing the QDM reference model with the notions of Data Model Element (including Category, Datatype, Attribute, Valueset, etc.) and Logic Element (including Logic Operator, Function, Comparison Operator, Temporal Operator, etc). In this study, we extended the schema with the notions of HL7 FHIR Datatypes and Resources. The schema are designed as a natural extension of the ISO 11179 standard. We then populated the schema with data elements from HL7 FHIR models as QDM schema instances (Table 1). We also developed RESTful services on the top of the repository (https://github.com/PheMA/phema-mdr), being utilized by a phenotype authoring tool under active development.

Table 1. The number of data elements populated and examples

	QDM	HL7 FHIR	Examples (FHIR)
Category	18	99	*Medication*
Datatype	76	99	*Medication*
Attribute	528	1021	*Medication.kind*
ValueSet	-	180	*MedicationKind*
Logic Element	53	-	-

Conclusion

Our system provides a standards-based semantic infrastructure in enabling data element services to support phenotype authoring and execution. In future work, we plan to develop a standard interface mechanism with Clinical Information Modeling Initiative (CIMI)-compliant clinical models.

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