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

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Abstract

This study describes 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 this study, we extend the Semantic Web-based framework of a previous study that provides a standards-based, semantically annotated, machine-readable rendering of the QDM [2], and develop a semantic metadata repository and associated Web services by integrating HL7 FHIR data element models [3]. Integrating the data elements provides a more comprehensive coverage for clinical phenotype applications.

Methods

Our system is comprised of first a semantic data element repository layer, in which 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. The second layer is a semantic services layer, while the third layer is a phenotype application layer.

In our previous study [2], we developed a QDM schema in OWL representing the QDM reference model. In this study, we extended the schema with the notions of HL7 FHIR Datatypes and Resources, and is designed as a natural extension of the ISO 11179 standard. We populated the schema with data elements from HL7 FHIR models as QDM schema instances (Table 1), and developed RESTful services on the repository (https://github.com/PheMA/phema-mdr), being utilized by a phenotype authoring tool under active development.

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.

Acknowledgments

References

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Table 1

Populated data elements

	QDM	HL7 FHIR	Examples (FHIR)
Category	18	99	Medication
Datatype	76	99	Medication
Attribute	528	1021	Medication Kind
Value Set	-	180	Medication Kind
Logic Element	53	-	-