Profiling ICD-11 Terminological Content Using the Common Terminology Services 2 (CTS2) Standard

Guoqian Jiang, Harold R. Solbrig, Christopher G. Chute Mayo Clinic College of Medicine Rochester, MN, USA

Abstract—In the beta phase of the 11th revision of International Classification of Diseases (ICD-11), the World Health Organization (WHO) exposes ICD-11 content through a collection of web services. The emerging Common Terminology Services 2 (CTS2) standard provides a common model and semantics for representation, interchange, and federation of terminological resources. We hypothesize that the CTS2 standard can provide service layer of standardization that could potentially aid in the interoperability among authoring applications for the ICD-11 revision. This paper examines the existing WHO ICD-11 content services from the perspective of the CTS2 standard. The content in the WHO ICD-11 content services was mapped to the Code System Catalog, Code System Version Catalog, Entity Description and Association models in the CTS2 specification. It proposes mappings for existing ICD-11 properties and suggests additional CTS2 properties may be important for ICD-11. The mapping effort was used to develop a prototype of the CTS2 Services Wrapper for the ICD-11. In conclusion, the CTS2 standard is useful in exposing ICD-11 content representation through predictable and familiar services.

Keywords—ICD-11; CTS2; Medical Classification; Data Standards; Biomedical Ontologies

I. INTRODUCTION

The International Classification of Diseases (ICD) is an international data standard presently maintained and curated by the World Health Organization (WHO) [1], and is widely used to support international comparison of mortality statistics. In March 2007, WHO officially launched the 11th revision of ICD (ICD-11) that aims to digitize the content of ICD using modern knowledge representation methods and standards instead of a traditional manual revision process.

To facilitate the ICD-11 revision, WHO developed a content model to present the knowledge that underlies the definitions of an ICD entity. The content model is composed of three layers: a foundation layer, a linearization layer, and an ontological layer [2]. The foundation layer is the core product of the ICD-11 revision that stores the full range of knowledge of all classification units in ICD. Each linearization, of which there may be many such as Mortality, Morbidity and Primary Care, corresponds to the classical print versions of ICD. The ontological layer provides references to formal definition of terms and relationships through two components: 1) Reference Ontologies and 2) The Common Ontology. The Common Ontology forms the semantic backbone of the ICD Foundation Layer and is a shared subset of the Systematized Nomenclature

Can Celik, Bedirhan T. Ustun World Health Organization Geneva, Switzerland

of Medicine – Clinical Terms (SNOMED CT). SNOMED CT is owned and maintained by the International Health Terminology Standard Development Organization (IHTSDO). The IHTSDO and the WHO signed a collaborative agreement in July 2010, aiming at enabling harmonization—as complementary tools—of WHO Classifications and SNOMED CT, which essentially establishes SNOMED CT as the core of the ontological component of ICD [3].

The beta phase of the ICD-11 revision started in May 2012, and WHO intends to accept public input through a distributed model of authoring. It is anticipated that the public reviewer community will potentially be large, as reviewers will want to be able to access and download target ICD-11 contents into their own evaluation tools and/or try alternatives in their own environment. To enable social computation and collaboration among broader communities, it is imperative to have an opendata access solution for ICD-11 contents. In a previous study, we proposed to build Simple Knowledge Organization System (SKOS)-compliant Semantic Web RESTful services that aim to support the authoring application development of ICD-11 [4]. In a recent development, WHO has exposed the ICD-11 content through a collection of web services, which include the SKOS-based Semantic Web Resource Description Framework (RDF) services (below).

The Common Terminology Services 2 (CTS2) [5] is an emerging Object Management Group (OMG) standard [6] that defines the functional requirements of a set of service interfaces to allow the representation, access, and maintenance of taxonomy content either locally, or across a federation of terminology service nodes. We consider that building CTS2 services on the top of the WHO ICD-11 content services will potentially 1) provide standards-based representation of ICD-11 content; 2) provide sophisticated terminology services (e.g., lexical/semantic query services; value set definition and management services); 3) facilitate the interactions with other terminologies such as SNOMED CT; and 4) optimize the interoperability among downstream authoring applications for ICD-11 revision.

The objective of this paper is to describe the existing WHO ICD-11 content services using the CTS2 standard specifications. We profiled the WHO ICD-11 content services using the information models of Code System Catalog, Code System Version Catalog, Entity Description and Association from the CTS2 specification. We developed the mappings for the elements between the two service models and identified the

CTS2 elements that are missing from the WHO services but important for the ICD-11 content representation and services. We also developed a prototype of the CTS2 Wrapper Services for ICD-11.

II. BACKGROUND

A. WHO ICD content services

A Unified Resource Identifier (URI) is commonly used to name and identify a resource, and is treated as a core principle of Semantic Web Linked Open Data by Tim Berners-Lee [7]. A RESTful, resource-oriented service exposes a URI for every piece of data the client might need to interact with. A welldesigned URI structure/scheme would allow users to easily explore and invoke ICD content through an implementation of the RESTful services. Recently, an ICD URI scheme was proposed for naming and supporting web services by WHO. The base URI of <u>http://id.who.int</u> has been adopted, with <u>http://id.who.int/icd/schema</u> as the prefix for the vocabulary terms that are related to ICD classification efforts maintained by WHO and <u>http://id.who.int/icd/entity</u> for the foundation entities related to ICD concepts.

Table 1 shows the service categories, URI scheme and returned properties modeled and implemented in the existing WHO ICD content services. There are two content service categories: Foundation and Release. Under the release category, the URI Schemes are defined for the subcategories of the ICD-11 Linearizations and ICD-10 with/without minor version. For each category, the URI Schemes for the Top Level and Individual Entities are defined separately. Fig. 1 shows a HTML view of the RDF rendering for an individual entity in the Foundation category – "Acute myocardial infarction".

Table 1.The service categories, URI scheme and returned properties modeled in the WHO ICD content services.

Service Categories URI Scheme		Returned Properties		
Foundation				
Top Level	http://id.who.int/icd/entity	Title, Definition, Child		
Individual Entity		Parent, Child, Title, Definition, Long Definition, Synonym, Narrower Term, Inclusion, Exclusion, Body Site, Body System, Causal Agents, Causal Mechanisms, Signs And Symptoms, Genomic Characteristics, Investigation Findings, Type, Intent, Activity when Injured, Object or Substance Producing Injury, Mechanism of Injury, Place of Occurrence, Substance Use, Children and Youth Impact, Communication Impact, Interpersonal Relations Impact, Household Activities Impact, Life Management Activities Impact, School Activities Impact, Work Activities Impact, Mobility Impact, Self Care		
Release	http://id.who.int/icd/entity/{id}	Impact, Social Participation Impact, Understanding Impact		
ICD-11 Linearizations				
WITHOUT minor version				
Top Level	http://id.who.int/release/11/{Linearization			
	Name}	Title, Latest Version, Version		
Individual Entity	http://id.who.int/release/11/{Linearization Name}/{id}	Title, Latest Version, Version		
ICD-11 Linearizations		The, Edest Version, Version		
WITH minor version				
Top Level	http://id.who.int/release/11/{Minor			
Version}/{Linearization Name}		Title, Definition, Child		
Individual Entity http://id.who.int/release/11/{Minor		Code, Parent, Child, Title, Definition, Long Definition,		
ICD-10 WITHOUT minor	Version}/{Linearization Name}/{id}	Inclusion, Exclusion, Index Terms, Class Kind, Source		
version				
Top Level http://id.who.int/release/10		Title, Latest Version, Version		
Individual Entity	http://id.who.int/release/10/{CODE}	Title, Latest Version, Version		
ICD-10 WITH minor				
version				
Top Level http://id.who.int/release/10/{Minor Version}		Title, Definition, Child		
http://id.who.int/release/10/{Minor Individual Entity Version}/{CODE}		Code, Parent, Child, Title, Definition, Inclusion, Exclusion, Class Kind, Coding Hint, Note		

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	http://id.who.int/ icd/ entity/ 1334938734		
icd:bodySite	Myocardium structure (body structure)		
icd:bodySystem	Circulatory System (Cardiovascular System)		
skos:broaderTransitive	http://id.who.int/ icd/ entity/ 1964269418		
icd:causalMechanisms	Infarct (morphologic abnormality)		
skos:definition	The term acute myocardial infarction (MI) should be used when there is evidence of myocardial necrosis in a clinical setting consistent with acute myocardial ischemia. Under these conditions any one of the following criteria meets the diagnosis for MI;		
icd:exclusion	myocardial infarction: old certain current complications following acute myocardial infarction myocardial infarction: specified as chronic or with a stated duration of more than 4 weeks (more than 28 days) from onset myocardial infarction: subsequent postmyocardial infarction syndrome		
icd:householdActivitiesImpact	d630. Preparing meals		
icd:inclusion	ST elevation (STEMI) and non-ST elevation (NSTEMI) myocardial infarction myocardial infarction specified as acute or with a stated duration of 4 weeks (28 days) or less from onset		
icd:interpersonalRelationsImpact	cd.interpersonalRelationsImpact d7702. Sexual relationships		
icd:lifeManagementActivitiesImpac	${ m t}$ d230. Carrying out daily routine		
<u>icd:narrowerTerm</u>	ST elevation (STEMI) and non-ST elevation (NSTEMI) myocardial infarction myocardial infarction specified as acute or with a stated duration of 4 weeks (28 days) or less from onset		
<u>skos:narrowerTransitive</u>	http://id.who.int/ icd/ entity/ 680129526 http://id.who.int/ icd/ entity/ 2016376826 http://id.who.int/ icd/ entity/ 391388807 http://id.who.int/ icd/ entity/ 1679681893		
skos:prefLabel	Acute myocardial infarction		
icd:type	Disease Type		
icd:workActivitiesImpact	d850. Remunerative employment		

Fig. 1. A HTML view of the RDF rendering of the individual foundation entity "Acute myocardial infarction (http://id.who.int/icd/entity/1334938734)". (icd prefix: http://id.who.int/icd/schema/; skos: http://www.w3.org/2004/02/skos/core#.)

B. CTS2 Standard Specification

CTS2 is an OMG specification [6] for representing, accessing, and disseminating terminological content. The CTS2 platform independent model (PIM) describes the formal model and semantics of the various components. The PIM model is rendered in the Unified Modeling Language (UML) and its specification is publicly accessible at the OMG website. As of April 2014, the CTS2 version 1.1 is the latest version released [6].

The CTS2 specification specifies the information models for the terminological artifacts. These artifacts include:

- Code System Catalog metadata about code systems (ontologies, code sets, thesauri, classification systems, etc.) (See Fig. 2)
- Code System Version Catalog metadata about specific versions of code systems.
- Entity Description a set of entity (aka. "class", "category", "concept", "predicate", "property", "term", "individual") identifiers known to the service along with information about which code system versions make assertions about these identifiers and what they say.
- Association sets of "semantic" assertions about entity identifiers, in which the entity identifier may play the role of subject, predicate (verb) or object in the assertions.
- Value Set Catalog metadata about sets of entity identifiers (value sets) that have been grouped for some purpose.
- Value Set Definition information about how value sets are constructed.

- Concept Domain Catalog a catalog of abstract "concept domains" that represent a collection of possible meanings.
- Map Catalog a catalog of "maps" collection of rules that allow human or machine assisted transformation between the codes in one value set or code system and those in a second.

The CTS2 specification also supports a number of functional areas including the Read, Query, Update, History, Maintenance, and Temporal services.

AbstractResourceDescription
CodeSystemCatalogEntry
codeSystemName :CodeSystemName {readOnly} codeSystemCategory :CodeSystemCategoryReference [01] ontologyDomain :OntologyDomainReference [0*] ontologyType :OntologyTypeReference [01] designedForOntologyTask :OntologyTaskReference [0*] hasOntologyLanguage :OntologyLanguageReference [01] includes :CodeSystemReference [0*] versions :CodeSystemVersionCatalogEntryDirectoryURI [01] {readOnly} currentVersion :CodeSystemVersionCatalogEntryDirectorgURI [01] {readOnly} usedOntologyEngineeringTool :OntologyEngineeringToolReference [0*]
::AbstractResourceDescription releaseDocumentation :OpaqueData[01] releaseFormat :SourceAndNotation[0*]
::ResourceDescription about :ExternalURI {readOnly} describedResourceType :CTS2ResourceType {readOnly} resourceID :Localidentifier {readOnly} formalName :String [01] keyword :String [01] resourceSynopsis :EntryDescription [01] additionalDocumentation :PersistentURI [0*] sourceAndRole :SourceAndRoleReference [0*] rights :OpaqueData [01] note :Comment [0*] property :Property [0*] alternateID :ExternalURI [0*] sourceStatements :StatementDirectoryURI [01] {readOnly}
::Changeable entryID :PersistentURI {readOnly} entryState :EntryState status :StatusReference [01]

Fig. 2. Code System Catalog Information Model.

III. PROFILING ICD TERMINOLOGICAL CONTENT

We analyzed the existing WHO ICD content services, and profiled the ICD-11 terminological content using the CTS2

information models of Code System Catalog, Code System Version Catalog, Entity Description and Association. We identified the mappings of the properties between the two service models and suggested the CTS2 properties that are important for ICD-11 but currently missing from the existing service implementation.

A. Code System Catalog

We identified three code systems from the existing WHO ICD content services: 1) **ICD-11** Foundation (http://id.who.int/icd/entity) that corresponds to the Top Level of the Foundation category in Table 1; 2) ICD-11 Morbidity (http://id.who.int/release/11/morbidity) that corresponds to the Top Level of the ICD-11 Linerizations without minor version in Table 1; and 3) ICD10 (http://id.who.int/release/10) that corresponds to the Top Level of the ICD10 without minor version. Table 2 shows the mappings of the properties between the ICD code systems and the CTS2 Code System Catalog model. We also suggested 5 properties shown in red (codeSystemName, resourceType, resourceSynopsis, rights and entryState) that may be important for describing the ICD code systems.

Properties in CTS2	Properties in ICD
codeSystemName	Suggested for ICD
codeSystemCategory	
ontologyDomain	
ontologyType	
designedForOntologyTask	
hasOntologyLanguage	
includes	
versions	version
currentVersion	latestVersion
usedOntologyEngineeringTool	
releaseDocumenation	
releaseFormat	
about	URI
describedResourceType	
resourceID	
formalName	Title
keyword	
resourceType	Suggested for ICD
resourceSynopsis	Suggested for ICD
additionalDocumentations	
sourceAndRole	
rights	Suggested for ICD
note	
property	
alternateID	
sourceStatements	
entryID	
entryState	Suggested for ICD
status	

B. Code System Version Catalog

We identified the code system versions for each of three existing ICD code systems. 1) ICD-11 Foundation Beta (<u>http://id.who.int/icd/entity</u>) is the current version of the code system ICD-11 Foundation. Note that for the code system ICD-11 Foundation, the existing WHO services do not provide any

versioning information, so the same URI is used for both the code system and the code system version for now. 2) ICD-11 Morbidity Beta (http://id.who.int/release/11/beta/morbidity) is the current version of the code system ICD-11 Morbidity; 3) ICD10 has two version: ICD10 2010 (http://id.who.int/release/10/2010) ICD10 2008 and (http://id.who.int/release/10/2008), in which ICD10 2010 is the current version of the code system ICD10.

Table 3. Profiling by the CTS2 Code System Version Catalog model.

Properties in CTS1	Properties in ICD
Properties in CTS2 codeSystemVersionName	Suggested for ICD
versionOf	Suggested for ICD
imports	Suggested for ICD
defaultLanguage	Suggested for ICD
	Suggested for ICD
supportedLanguage assoications	Suggested for ICD
entityDescriptions	
classes	
individuals	
roles	
documentURI	
sourceAndNotatoin	
predecessor	
officialResourceVersionId	Title
officialReleaseDate	Suggested for ICD
officialActivationDate	Suggested for ICD
state	
about	URI
describedResourceType	UKI
resourceID	
formalName	Title
keyword resourceType	Suggested for ICD
resourceSynopsis	Suggested for ICD Definition
additionalDocumentations	
sourceAndRole	
	Suggested for ICD
rights	Suggested for ICD
note	
alternateID	
sourceStatements	
entyrID	Suggested for ICD
entryState	Suggested for ICD
status	Suggested for ICD

Table 3 shows the mappings of the properties between the ICD code system versions and the CTS2 Code System Version Catalog model. We also suggested 9 CTS2 properties shown in red that may be important for describing the ICD code system versions.

C. Entity Description

For the individual entities in each code system version, we used the CTS2 Entity Description model for the profiling. Table 4 shows the mappings of the properties between the individual entities and the CTS2 Entity Description model. We also suggest four additional CTS2 properties shown in red (describingCodeSystemVersion, ancestors, descendants and entityType) that may be important for describing an individual ICD-11 entity.

Table 4	. Profiling	by the	CTS2	Entity	Description model.
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Property in CTS2	Property in ICD
entityDescriptionType	
about	URI
entityID	ID, Code
alternateEntityID	
describingCodeSystemVersion	Suggested for ICD
codeSystemRole	
designation	Title, Synonym
definition	Definition, Long Definition
example	
note	
	Narrower Term, Inclusion,
	Exclusion, Index Terms, Class
property	Kind, Coding Hint
sourceStatements	
	Body Site, Body System, Causal Agents, Causal Mechanisms,
	Signs And Symptoms, Genomic Characteristics, Investigation
	Findings, Type, Intent, Activity
	when Injured, Object or
	Substance Producing Injury,
	Mechanism of Injury, Place of
	Occurrence, Substance Use (by
subjectOf	CTS2 Association Directory)
predicateOf	
targetOf	
parent	Parent (by CTS2 Entity Reference)
parents	Parent (by CTS2 Entity Directory)
ancestors	Suggested for ICD
children	Child (by CTS2 Entity Directory)
descendants	Suggested for ICD
entityType	Suggested for ICD
instances	
equivalentEntity	

D. Association

As mentioned in the section above, the CTS2 Association represents sets of "semantic" assertions about entity identifiers, in which the entity identifier may play the role of subject, predicate (verb) or object in the assertions. In fact, the properties "subjectOf", "predicateOf", "targetOf", "parents", "children", "ancestors" and "descendants" in the CTS2 Entity Description model (see Table 4) are such sets of assertions utilizing the Association model. Specifically, the CTS2 Association Directory model is used for the semantics asserted for the property "subjectOf" and the CTS2 Entity Directory model is used for the semantics asserted for the rest of four properties.

IV. PROTOYPE IMPLEMENTATION

Based on the mappings identified and the new properties suggested in the section above, we have initiated a prototype implementation by building a CTS2 services wrapper for ICD-11. The wrapper takes a URI from existing WHO ICD content services as input, and extracts RDF-based content (by content negotiation) using the Jena API [9], and develops the CTS2compliant rendering using CTS2 Development Framework APIs [10].

Note that the CTS2 Development Framework is a development kit for rapidly creating CTS2 compliant applications. The CTS2 Development Framework provides a REST web application, along with URL routing and parameter handling. The CTS2 Development Framework has been successfully used to build a NCBO BioPortal Wrapper service that provide a layer of standardized CTS2 signatures to existing biomedical ontologies available in BioPortal.

Table 5 shows a set of service signatures proposed for the implementation of an ICD-11 CTS2 Services Wrapper. The wrapper services cover those terminological artifacts presented in the section III. The detailed examples of each service for the prototype implementation can be accessed at:

http://informatics.mayo.edu/rest2/project/icd11/cts2/doc.

Table 5. A set of services proposed for the implementation of the ICD-11 CTS2 Services Wrapper.

Signature	Description		
Code System Catalog	-		
codesystems	Return all named code systems in ICD11		
codesystembyuri?uri={uri}	Read catalog entry for {uri}		
Code System Version Catalog			
codesystemversionsbyuri?uri={uri	Return all known versions of the named code system by {uri}		
codesystemversionbyuri?uri={uri}	Read code system version catalog entry for {uri}		
Entity Description			
entitybyuri?uri={uri}	Read a synopsis for the referenced entity URI		
Association Directories			
entitysubjectofbyuri?uri={uri}	Return all associations having the named entity as the subject.		
Entity Directories			
entityparentsbyuri?uri={uri}	Return a list of entities that are "parents" (target of skos:broaderTransitive relationship) of the named concept.		
entityancestorsbyuri?uri={uri}	Return a list of entities that are ancestors (transitive target of skos:broaderTransitive relationship) of the named concept.		
entitychildrenbyuri?uri={uri}	Return a list of entities that are direct children (target of skos:narrowerTransitive relationship) of the named concept.		
entitydescendantsbyuri?uri={uri}	Return a list of entities that are descendants (transitive target of skos:narrowerTransitive relationship) of the named concept.		

V. DISCUSSION

In this paper, we describe our efforts in profiling the ICD-11 terminological content exposed by existing WHO services using the CTS2 standard specification. The profiling results revealed a number of areas in the existing WHO ICD content services, which can potentially be enriched by the CTS2 standard.

First, we found that ICD-11 Foundation, as a code system from the CTS2 perspective, does not have a versioning mechanism in the existing WHO services. The existing WHO service implementation was based on a specific use case assumption that all the linearization releases (e.g., ICD-11 Morbidity) should refer to versionless foundation entity URIs so that different linearizations created at different times could be anchored by the foundation entity URIs. However, when we consider the entire life cycle of ICD-11 content authoring, the versioning mechanism is a critical component for the authoring applications that would need to access version specific foundation entities or history of changes. We propose that WHO may consider a model similar to that used by SNOMED CT which allows for both fine-grained and coarse versioning. Specifically, revisions to the SNOMED CT are released twice a year and marked by the release date, e.g., the version 20140131. In a fine-grained level in the Release Format 2 (RF2) of SNOMED CT [8], each component has its effective time and active status to indicate versioning mechanism. The CTS2 Code System and Code System Version models contain rich metadata for representing such versioning mechanism.

Second, we identify three code systems and their corresponding code system versions from the existing WHO services, while we also find that these important terminological artifacts are generally underspecified. In addition to identifying the property mappings, we suggest 5 properties from the CTS2 Code System Catalog model and 9 properties from the CTS2 Code System Version Catalog model for describing these ICD-11 terminological artifacts. For example, we suggest property "rights" for the artifacts because it will make the WHO rights statement explicit. Such statement will be critical for providing the guidance to the user community for the use of ICD-11 that is critical to the users. We also suggest the properties "defaultLanguage" and "supportedLanguage" for the ICD-11 because the multilingual support is one of major goals for the ICD-11 revision and eventually at least the six official United Nations languages will need to be supported.

Third, those ICD properties (such as Body System, Causal Agents, etc.) that will have values from external terminologies (e.g., SNOMED CT, ICF) are mapped to the CTS2 property "subjectOf", which can be elegantly described by the Association Directory model. Note that the existing WHO services have not exposed the external identifiers for those values annotated by external terminologies. For example, as shown in Fig. 1, the property icd:bodySite in the foundation entitv "Acute myocardial infarction" has the value "Myocardium structure (body structure)" in a textual string. The original unique identifier from SNOMED CT (i.e., http://snomed.info/id/74281007) is not exposed in the existing services. In addition, CTS2 uses the Entity Directory model to describe the semantics for the properties "parents", "children", "ancestors" and "descendants" in the CTS2 Entity Description model. We found that the existing WHO services do not have a mechanism to retrieve the transitive closure for parent (i.e., skos:broaderTransitive) or child (skos:narrowerTransitive) relationships to get all ancestors or descendants for a specific ICD entity. This is an area that would need to be enhanced in the future.

Fourth, CTS2 is designed to address a broad range of requirements within the ontology and terminology community. In addition to the specification about the four kinds of terminological artifacts addressed by this paper, CTS2 specifications also have the notions of Value Set Catalog, Value Set Definition and Map Catalog. Although the existing WHO services have not exposed any such terminological artifacts, these artifacts (e.g., value sets) have been actively utilized in the ICD-11 revision process. Actually, a collection of value sets (including Body Systems, Anatomical Sites, Functioning, etc.) had been developed and utilized in support of the ICD-11 revision. In a previous study, we developed and evaluated approaches to value set extraction from SNOMED CT for the ICD-11 anatomy use case [11]. We consider that CTS2 would play a key role in enabling the standard representation and dissemination of such terminological artifacts in the future.

In summary, CTS2 standard specification is a very useful tool for profiling the ICD-11 terminology content. The outcome produced by this profiling would potentially enrich the WHO ICD-11 content representation and enable a robust and standards-based content services to better support the ICD-11 revision applications.

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