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Maintenance of ONC Terminology for i2b2 Metadata

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Problem

ONC terminologies are constantly adding new content and deactivating existing codes. The University of Nebraska Medical Center (UNMC) deploys three primary code sets that require regular updating to support research: SNOMED CT, RXNORM / NDC, and LOINC. A problem across the i2b2 community is keeping these terminologies up-to-date and loading them into i2b2 for timely analysis of EHR data. We have developed tool kits for rapid deployment of SNOMED CT metadata and will be extending the work to RXNORM/NDC and LOINC.

Objectives

Our primary objective is to keep our i2b2 ontology build current with standards development organization terminology releases. SNOMED CT is an ontological model that relies on polyhierarchies, which are challenging to deploy in a clinical research data warehouse reliant upon relational database structures. By building an architecture using queriable transitive closure tables that can be loaded directly from SDO terminology releases, we create an i2b2 metadata table capable of supporting advanced querying of patient data.

Methodology

We developed an approach to rapidly respond to the problem using transitive closure tables as a component of i2b2 metadata. Transitive closure tables include every concept in a hierarchy in a relationship with all of its hierarchical descendants. Transitive closure tables support i2b2 folder searching and also the build of all other CRC tables. We created a SQL procedure to produce those tables quickly as needed whenever we receive a new release of SNOMED CT. We are now working to implement this tooling and architecture for RXNORM and LOINC.

SNOMED CT Transitive Closure Build

Transitive closure tables are built from the terminology by traversing and listing all the IS_A relationships which relate concepts within the ontology. Each SUPERTYPE_SCTID reference is accompanied by one row for each unique SUBTYPE_SCTID which is a subtype of that concept. The GENERATION counts the minimum number of relationship traversals between the two concepts in the SNOMED CT ontology.

Domain Description: Contains SNOMED CT reference codes and terms with lists of dependent (subtype) concepts for use in query and creating SNOMED CT value sets.

SNOMED CT Table Specification

Field Name	Data Tuna	Value Cot	Definition and Comments	Course
Field Name	Data Type (SQL)	Value Set	Definition and Comments	Source
SUPERTYPE_SCTID	code (biginteger)	SNOMEDCT	The SNOMED CT concept identifier of the node which is the supertype (parent) in this relationship between SNOMED CT concepts. Format 19N.) IHTSDO
			The context free reference code for the SNOMED CT concept that is the supertype concept in this relationship, of the format 'SNOMEDCT:NNNNNN'. This is the reference as it is stored in the i2b2 table OBSERVATION_FACT.	
SUPERTYPE_CNCPT	char(40)	SNOMEDCT		IHTSDO
			The Fully specified name of the SNOMED CT parent concept	
SUPERTYPE TERM	char(2048)	n/a		IHTSDO
SUBTYPE SCTID	code (biginteger)	SNOMEDCT	The SNOMED CT concept identifier of the node which is the subtype (child, grandchild, great-grandchild) in this relationship between SNOMED CT concepts. Format 19 N.	IHTSDO
		SNOVED ST	The context free reference code for the SNOMED CT concept that is the subtype concept in this relationship, of the format 'SNOMEDCT:NNNNNN'. This is the reference as it is stored in the i2b2 table OBSERVATION_FACT and used for searching by i2b2.	uuten o
CONCEPT_CD	char(40)	SNOMED CT		IHTSDO
			The Fully specified name of the SNOMED CT child concept.	
SUBTYPE_TERM	char(2048)	n/a		IHTSDO
			How many IS_A relationships separate the two concepts within SNOMED CT. (The shortest path length from the SUPERTYPE concept to the SUBTYPE concept in the SNOMED CT hierarchy)	
GENERATION	integer	n/a	as a second of the second of t	IHTSDO

SNOMEDCT_TRC to i2b2 Metadata

Transitive closure tables contain all necessary data to construct the SNOMED CT hierarchy. A second SQL procedure processes the table into an i2b2 metadata table strictly adherent to i2b2 formatting, but employing folder searching using equivalence searching into the transitive closure table.

Pneumococcal Pneumonia Exemplar

ERTYPE_SCTID SUPERTYPE_CNCPT	SUPERTYPE_TERM	SUBTYPE_SCTID SUBTYPE_CNCPT	SUBTYPE_TERM GENERA
	Pneumococcal		Pneumococcal
SNOMEDCT:23360700	pneumonia		pneumonia
2336070000	(disorder)	233607000 SNOMEDCT:233607000	(disorder)
			Bronchopneumoni
			a caused by
	Pneumococcal		Streptococcus
SNOMEDCT:23360700	pneumonia	10625711000119 SNOMEDCT:1062571100011	pneumoniae
2336070000	(disorder)	100 9105	(disorder)
			Pneumococcal
			pneumonia
			associated with
			acquired
	Pneumococcal		immunodeficiency
SNOMEDCT:23360700	pneumonia		syndrome
2336070000	(disorder)	420787001 SNOMEDCT:420787001	(disorder)
	Pneumococcal		Pneumococcal
SNOMEDCT:23360700	pneumonia		lobar pneumonia
2336070000	(disorder)	266350000 SNOMEDCT: 266350000	(disorder)

Conclusions

Developing methods for implementing rapidly evolving medical ontologies as i2b2 metadata requires tool kits and procedures supporting efficient processing of complex data into useful i2b2 artifacts. In light of the recent invasion of COVID-19 on the world health scene, the ability to respond and deploy changed terminology content is imperative to support the mission of i2b2 as instrument of interoperable research in a changing world.

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