The Role of Ontologies for Sustainable, Semantically Interoperable and Trustworthy EHR Solutions

TermInfo Draft Standard for Trial Use (DSTU): Managing overlap between SNOMED CT and HL7-RIM

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Outline

• Health information standards
• Typology of representation artifacts
• Semantic overlap between representation artifacts
• TermInfo Draft Standard for Trial Use
• Outlook
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<tr>
<th>Standards</th>
<th>Typology</th>
<th>Overlap</th>
<th>TermInfo</th>
<th>Outlook</th>
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<td><strong>Two Health Information Standards</strong></td>
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<tr>
<th></th>
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<tr>
<td><strong>Characterization</strong></td>
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<td>Terminology Standard for healthcare</td>
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<td>Ontology-inspired Terminology</td>
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<td>Model of meaning</td>
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<td><strong>Represents</strong></td>
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<td>Ann Arbor, Michigan, USA</td>
<td>International Healthcare Terminology Standards Development Organisation Copenhagen, Denmark</td>
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<td><strong>Participation</strong></td>
<td>HL7 local organizations in over 30 countries</td>
<td>Member states: Australia, Canada, Cyprus, Denmark, Lithuania, New Zealand, Singapore, Spain, Sweden, The Netherlands, United Kingdom, United States</td>
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From metaphysics...

Ontology
- theory of reality

Semantics
- theory of meaning of (human language) designations

Epistemology
- theory of knowledge
...to an ideal world of representation artifacts

Ontologies
- theories that attempt to give precise mathematical formulations of the properties and relations of certain entities.
  (Stanford Encyclopedia of Philosophy)

Terminologies
- Set of terms representing the system of concepts of a particular subject field.
  (ISO 1087)

Data models / Information models
- artefacts in which information is recorded

A. Rector, SemanticHealth D6.1
Examples

### Ontologies

- **Formal descriptions**
  - MRSA *subtype-of* SA
  - SA *subtype-of* Staphylococcus
  - SA *implies* bearer-of *some* MR quality

- **Textual descriptions**
  - “MRSA is defined as SA for which methicillin has no toxic effect”

### Terminologies

- **Concept 1**
  - Synonyms:
    - SA
    - *Staphylococcus aureus*
    - *Staph. aur.*

- **Concept 2**
  - Synonyms:
    - MRSA
    - Methicillin-resistant SA
    - Methicillin-resistant *Staphylococcus aureus*

### Data models / Information models

- Methicillin resistance:
  - Clinically confirmed
  - Confirmed by antibiogram
  - Suspected
  - None
  - Unknown
but in the real, chaotic world...
we have to deal with “living” representational artifacts far from being ideal
that combine terminology, ontology and information model elements
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Problem: Semantic Overlap

Terminologies / Ontologies stray the terrain of information models

- Historically: solution developers tended to work with a single representation standard
- Multiple approaches to encode the same information
- Risk of arbitrary design decisions when used together
- Lack of clarity with regard to satisfying unmet representational need
Examples of “epistemic intrusion”

- **SNOMED CT**: “Suspected autism”
- **SNOMED CT**: “Biopsy planned”
- **SNOMED CT**: “Take at regular intervals”
- **ICD 10**: “Tuberculosis of lung, confirmed histologically”
- **ICD-O**: “Basal cell tumor, uncertain whether benign or malignant”
- **ICD-9-CM**: “Replacement of unspecified heart valve”
- **NCI Thesaurus**: “Unknown If Ever Smoked”
- **NCI Thesaurus**: “Absent Adverse Event”
Solutions

- Establish a clear boundary between information models and ontologies: desirable but unfeasible for legacy systems

- Develop rules for managing ambiguities
  - HL7 TermInfo

*https://svn.connectingforhealth.nhs.uk/svn/public/nhscontentmodels/TRUNK/ref/HL7/TermInfo.htm*
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TermInfo Draft Standard for Trial Use (DSTU): History

- 2004 onwards, Growing interest in use of SNOMED Clinical Terms (SNOMED CT) in the HL7 community
- HL7 Vocabulary Technical Committee (supported by SNOMED International and NASA) launched the 'TermInfo Project’ with the following missions:
  - General: investigate interfacing between HL7 information models and terminologies or code systems.
  - Specific: A guide on use of SNOMED CT within the HL7 V3
- Outcome September 2007:
  - Guide to Use of SNOMED CT in HL7 Version 3’ accepted as a Draft Standard for Trial Use (DSTU)

Using SNOMED CT in HL7 v3 DSTU

1.3 Scope

The primary scope of this implementation guide is to provide guidance for the use of SNOMED CT in the HL7 V3 Clinical Statement pattern. The intent is to guide implementers in the construction of instances based on models derived from that pattern. These include models covering the representation of clinical information from the perspective of various HL7 domains including Structured Documents (CDA release 2), Patient Care, Orders and Observations and models using the Clinical Statement CMETATM.
Structure of **SNOMED CT** in **HL7 v3 DSTU**

1. Introduction and Scope

2. **Guidance on Overlaps between RIM and SNOMED CT Semantics**
   3. Common Patterns
   4. Normal Forms
   5. SNOMED CT vocabulary domain constraints
   6. Glossary

Appendix A  General Options for Dealing with Potential Overlaps
Appendix B  References
Appendix C  Revision changes
Appendix D  SNOMED CT Open Issues
Appendix E  Detailed aspects of issues with a vocabulary specification formalism
Section 2: Guidance on overlaps between RIM and SNOMED CT Semantics

- Detailed walk-through of RIM attributes vs. SNOMED CT properties:
  - Act.classCode
  - Act.code and Observation.value
  - Act.moodCode
  - Act.statusCode
  - Procedure.targetSiteCode and Observation.targetSiteCode
  - Procedure.approachSiteCode and SubstanceAdministration.approachSiteCode
  - Procedure.methodCode and Observation.methodCode
  - Act.priorityCode
  - Act.negationInd
  - Act.uncertaintyCode
  - Representation of Units
  - Dates and Times
HL7 moodCode ↔ SNOMED CT finding and procedure context

And so on...
### Using SNOMED CT in HL7 v3 DSTU

**Guidance on overlaps between RIM and SNOMED CT Semantics**

Each subsection in HL7 v3 DSTU Section 2: divided into:

1. Potential overlap
2. Rules and guidance
3. Rationale

Two examples
Example 1: `Procedure.targetSiteCode` and `Observation.targetSiteCode`

- **Potential Overlap:**
  - Complete overlap
    - HL-7 `targetSiteCodes` are defined as “the anatomical site or system that is the focus of the procedure / observation.”
    - SNOMED CT finding and procedure concepts have a defining attribute that specifies the site: e.g. `Appendicitis – Finding Site – Appendix structure`

- **Rules and Guidance**
  - omit `targetSiteCode` attribute from:
    - any `Act` class clone in which SNOMED CT is the only permitted code system for the `Act.code` attribute.
    - any `Observation` class clone in which SNOMED CT is the only permitted code system for the `Observation.value` attribute...`

- **Rationale**
  - Argues case for SNOMED CT attribute preference
  - Precision of available attributes; relationship grouping
  - The site of an action or event is clearly of ontological nature

- **Potential overlap**
  - The values in *ActMood* vocabulary partially overlap with SNOMED CT representations of *Finding context* and *Procedure context*
    - *Finding context* relevant to instances of HL7 Observation classes expressed in "event", "goal", "expectation" and "risk" moods.
    - *Procedure context* relevant to (i) instances of various HL7 Act classes including Procedure, SubstanceAdministration and Supply, (ii) instances of the HL7 Observation class except in "intent" moods (including "request" and other subtype of "intent").

- **Rules and guidance**
  - The moodCode SHALL be present in all Act class instances
  - Rules for valued moodCode / SNOMED CT associations:
    - ‘...IF moodCode <> INT (or subtype), THEN code attribute of Observation class MAY be populated by the following SNOMED CT expression patterns...’
      - Defaults described by default correspondence tables
      - Allowable patterns described by constraint tables
    - ‘If both are present then they must be kept in step’
**Example 2: Act.MoodCode**

- Mood Code = SNOMED CT context default and constraint tables

<table>
<thead>
<tr>
<th>moodCode</th>
<th>Mood Name</th>
<th>SNOMED CT Finding context</th>
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<td>EVN</td>
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<tr>
<td>GOL</td>
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<td>EXPEC</td>
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Finding constraints

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<th>moodCode</th>
<th>Mood name</th>
<th>SNOMED CT Finding context</th>
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<tr>
<td>EVN</td>
<td>Event</td>
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### Next Steps - DSTU

- Encourage use and testing
  - Marketing effort

- Encourage and support submission and timely resolution of issues encountered in use
  - **HL7 DSTU** issue reporting mechanism (pending re-publication)
    - [http://www.hl7.org/dstucomments/index.cfm](http://www.hl7.org/dstucomments/index.cfm)
  - **HL7** Project Homebase mechanism
    - [http://hl7projects.hl7.nscee.edu/](http://hl7projects.hl7.nscee.edu/)

- Encourage list membership and submission of issues
  - [http://www.hl7.org/special/committees/list_sub.cfm?list=hl7TermInfo](http://www.hl7.org/special/committees/list_sub.cfm?list=hl7TermInfo)
  - Conference call debate and resolution
  - Establish close ties with e.g. **IHTSDO** expertise for timely resolution/interim suggestions
  - Advancement through **IHTSDO** standards approval processes
HL7 and IHTSDO Sign Agreement
Up front coordination will bring significant improvements in interoperability and patient safety

Chicago, IL., U.S. and Copenhagen, Denmark – April 5, 2009 – Health Level Seven® Inc. (HL7®), the leading authority for global healthcare IT standards, and the International Health Terminology Standards Development Organisation (IHTSDO®), the leading provider of standardized clinical terminology, today announced a collaboration agreement that will foster interoperability and lead to improvements in patient safety by eliminating gaps and overlaps between the HL7 and IHTSDO standards.
The ontology / epistemology boundary is crossed by both standards SNOMED CT and HL7v3. Consequence: overlap!

DSTU produced to assist in the co-implementation of SNOMED CT and HL7v3

Provides guidance on:
- Representation overlap management
- Sensible integration of the standards

Provides mechanism for issues resolution and gap management where both standards used

Does not claim perfection and does need systematic testing

Recommendation: decrease of SNOMED CT / HL7v3 overlaps by collaborative development of both standards, assigning representational responsibility based on reproducible boundary rules—informed by ontological/epistemological considerations, balanced with consideration of real-world practical considerations.