Ontology-based Convergence of Medical Terminologies: SNOMED CT and ICD-11

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WHO – IHTSDO Joint Advisory Group (JAG) for the harmonisation between ICD-10, ICD-11 and SNOMED CT
ICD-11 – SNOMED CT Harmonization

• Background:
  – ICD: disease classification maintained by WHO (World Health Organization) ICD-11 revision process ongoing
  – SNOMED CT: ontology-based clinical terminology maintained by IHTSDO (International Health Terminology Standards Development Organisation)
  – Size: SNOMED CT >> ICD,
  – Coverage: ICD only diseases, SNOMED CT: all EHR content

• Institutional agreement between WHO and IHTSDO:
  – Goal: common ontological basis for both the ICD-11 foundation component (FC) and SNOMED CT
  – Practical implications:
    • Each class in the ICD-11 foundation component will correspond to exactly one class in SNOMED CT.
    • The transitive closure of taxonomic (subclass) relations in ICD-11-FC is included in the transitive closure of these relations in SNOMED CT.
ICD - SNOMED CT Mapping principle

- Taxonomies are main construction principle for both terminologies
- Edges correspond to subclass links. Each ICD class corresponds to exactly one SNOMED class (same letter).
- Subclass links contained in ICD but not SNOMED must be obtained by transitive closure.

ICD 11 Foundation Component (multihierarchica)

SNOMED CT
Two Principles of ontology-based mapping of SNOMED CT and ICD-11

1. The semantics of the subclass relation is shared

2. Classes to be aligned denote the same types of entities
Meaning of subClassOf

\[ \text{subClassOf} \ (X, Y) = \text{def} \ \forall i: i \in X \rightarrow i \in Y \]
Two Principles of ontology-based alignment of SNOMED CT and ICD-11

1. The semantics of the subclass relation is shared √

2. Classes to be aligned denote the same types of entities?
Is this correct?

Fracture of Ulna
Fracture of Radius
Fracture of Radius and Ulna

SNOMED CT Example
• **No**, if “clinical condition”: the combined fracture is composed by the two single fractures, not a subtype

• **Yes**, if “clinical situation”: “situation with X” or “patient having X”
Clinical condition view

Fractured radius

Fractured ulna

No overlap / inclusion of classes

Fractured radius + ulna
Clinical situation view

Clinical situation with fractured ulna

Clinical situation with fractured radius
Current axiomatization in SNOMED CT

‘Fracture of radius (disorder)’ equivalentTo

‘Fracture of forearm (disorder)’ and ‘Injury of radius (disorder)’ and

Group some (‘Associated morphology’ some ‘Fracture (morphologic abnormality)’) and

‘Finding site’ some ‘Bone structure of radius (body structure)’)

‘Fracture of ulna (disorder)’ equivalentTo

‘Fracture of forearm (disorder)’ and ‘Injury of ulna (disorder)’ and

Group some (‘Associated morphology’ some ‘Fracture (morphologic abnormality)’) and

‘Finding site’ some ‘Bone structure of ulna (body structure)’)

‘Fracture of radius AND ulna (disorder)’ equivalentTo

‘Fracture of radius (disorder)’ and ‘Fracture of ulna (disorder)’ and

Group some (‘Associated morphology’ some ‘Fracture (morphologic abnormality)’) and

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Current axiomatization in SNOMED CT

Inferred taxonomic links

- ‘Fracture of radius (disorder)’ equivalentTo
  ‘Fracture of forearm (disorder)’ and ‘Injury of radius (disorder)’ and
  Group some ‘Associated morphology’ some ‘Fracture (morphologic abnormality)’ and
  ‘Finding site’ some ‘Bone structure of radius (body structure)’

- ‘Fracture of ulna (disorder)’ equivalentTo
  ‘Fracture of forearm (disorder)’ and ‘Injury of ulna (disorder)’ and
  Group some ‘Associated morphology’ some ‘Fracture (morphologic abnormality)’ and
  ‘Finding site’ some ‘Bone structure of ulna (body structure)’

subClassOf

- ‘Fracture of radius AND ulna (disorder)’ equivalentTo
  ‘Fracture of radius (disorder)’ and ‘Fracture of ulna (disorder)’ and
  Group some ‘Associated morphology’ some ‘Fracture (morphologic abnormality)’ and
  ‘Finding site’ some ‘Bone structure of radius (body structure)’ and
  Group some ‘Associated morphology’ some ‘Fracture (morphologic abnormality)’ and
  ‘Finding site’ some ‘Bone structure of ulna (body structure)’
Facts / Hypotheses

• “Problematic” subclass links between SNOMED CT classes result from formal (description logics) definitions

• It can be shown:

\[ A_{\text{cond}} \text{ subClassOf } B_{\text{cond}} \]
\[ A_{\text{sit}} \text{ subClassOf } B_{\text{sit}} \]

\[ A_{\text{cond}} \text{ subClassOf hasPart } B_{\text{cond}} \]
\[ A_{\text{sit}} \text{ subClassOf } B_{\text{sit}} \]

→ More subClassOf relations between \textit{situation} classes
Review of 400 sample SNOMED CT disorder concepts

• Four experts:
  K. Spackman, A. Rector, J.-M. Rodrigues, S. Schulz

• Assessment of a sample of 400 SNOMED disorder concepts
  – Fully specified names
  – Formal definitions
  – Parent classes
  – Child classes

• Evidence for “clinical situation” reading

Schulz S, Rector A, Rodrigues JM, Spackman K. Competing Interpretations of Disorder Codes in SNOMED CT and ICD. Submitted to AMIA 2012
• \(\sim 11\%\) of disorder evidence that they represent *situations* and not *conditions* (such as *Fracture of radius and ulna*)
• For the rest, both interpretations are possible
• Agreement difficult – fuzzy boundary between what should be interpreted as a *clinical condition* and what as a *clinical situation*.
Possible actions

1. Redesigning the SNOMED CT disorder hierarchy to exclude interpretation of clinical situations: huge effort, difficult boundary decisions

2. Leaving disorder classes uncommitted: should support condition interpretation: many existing subclass relations wrong

3. Considering all SNOMED CT disorder codes as denoting clinical situations:
   – more robust
   – consistent with current state of the disorder hierarchy
   – agreement with ICD view on the meaning of the code
   – compatible with clinical use cases
Foundations of ontology-based alignment of SNOMED CT and ICD-11

1. The semantics of the subclass relation is shared

\[ \text{subClassOf} (X, Y) = \text{def} \forall i: i \in X \rightarrow i \in Y \]

2. Classes to be aligned denote the same types of entities

SNOMED CT disorder codes and ICD-11 classes denote:

Clinical Situations
**Current Concept:**

<table>
<thead>
<tr>
<th>Fully Specified Name:</th>
<th>Tetralogy of Fallot (disorder)</th>
</tr>
</thead>
<tbody>
<tr>
<td>ConceptId:</td>
<td>86299006</td>
</tr>
</tbody>
</table>

**Defining Relationships:**

- **Is a** Congenital abnormality of ventricles and ventricular septum (disorder)
- **Is a** Overriding aorta (disorder)
- **Is a** Pulmonic valve stenosis (disorder)
- **Is a** Right ventricular hypertrophy (disorder)
- **Is a** Ventricular septal defect (disorder)
Example 2

Extension of “Pulmonic Valve Stenosis” includes extension of “Tetralogy of Fallot”: FALSE
Example 2

Extension of “Situation with Pulmonic Valve Stenosis” includes extension of “Situation with Tetralogy of Fallot”: TRUE
Two diverging interpretations of disorder terms in SNOMED CT and ICD:

• They denote patient-borne **Conditions** such as body processes, states, dispositions, or (patho-) anatomical structures, which are reportable in the context of medical records

• They denote Clinical **Situations**, which are defined as phases of a patient’s life, during which he/she is bearer of (some combination of) pathological conditions.
Situations, conditions and role groups

‘Fracture of radius AND ulna (disorder)’ equivalentTo

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‘Finding site’ some ‘Bone structure of ulna (body structure)’)

Facts / Hypotheses

• Most SNOMED CT disorder concepts contain role groups

• The role group link can be interpreted as a relation that links a situation with a condition

• It can be shown:
  – ‘A_cond subClass of B_cond’ entails: ‘A_sit subClass of B_sit’
  – ‘A_cond subClass of hasPart B_cond’ entails: ‘A_sit subClass of B_sit’
Proper parts or taxonomic parents?

Example from Harold Solbrig

Tetralogy of Fallot

Traffic Light

VSD  PVS  RVH  OA

Red Light  Yellow Light  Green Light

is-a  is-a  is-a  is-a

is-a  is-a  is-a
Proper parts or taxonomic parents?

Example from Harold Solbrig