WHO – IHTSDO: SNOMED CT – ICD-11 coordination: Conditions vs. Situations

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Principles of ICD-SNOMED mapping within WHO IHTSDO JAG

- Goal: common ontological basis for both the (polyhierarchical) ICD-11 foundation component and SNOMED CT
- Each class (categories) in the ICD-11 foundation component will correspond to exactly one class in SNOMED CT. Exceptions: navigational classes, should be clearly kept distinct from ontological classes.
- The equivalence in meaning between these class pairs will be assured by common text definitions.
- The transitive closure of taxonomic (subclassOf) relations in ICD-11-FC is included in the transitive closure of subClassOf relations in SNOMED CT.
Summary Alan Rector’s paper (I) + discussion

- **ICD Foundational Layer**: contains much more knowledge than just ontology
  - **Ontological component**:
    - definitional / universal: “All x has some y”
    - What is shared with SNOMED CT is just the ontological “spine”, mainly the is-a hierarchies + axioms
    - text definitions
  - **Content model component**:
    - Non-ontological knowledge, e.g. Leukemia can be treated by Metotrexate, or
    - Those non-ontological pieces of knowledge are in the “content model”: probabilistic knowledge, default knowledge,
    - Refer to SNOMED CT concepts as value sets, but not in terms of DL logic
    - navigational classes
  - **Metadata component**
  - **Linguistic component**: Labeling information (linguistic), including multilingual issues
Summary Alan Rector’s paper (II)

• URIs: joint / separate

• Distribution form

• “Projection” (more an SW engineering issue)
  – Static version: no need to replicate
  – Dynamic version: replicate
Mapping principle

Edges correspond to subClassOf links. Each ICD class corresponds to exactly one SNOMED class (same letter).
SubClassOf - links contained in ICD but not SNOMED can be obtained by transitive closure.
Meaning of subClassOf (is-a)

Graph

Corresponding Venn diagram
Alignment of SNOMED CT and ICD11 requires that in both systems

1. The semantics of the subclass relation is shared
2. Classes to be aligned denote the same entities
Example 1

Fracture of Ulna
Fracture of Radius
Fracture of Radius and Ulna

subClassOf

Ulna
Radius
Is this True?

• FALSE, if X means “pathological entity”
• TRUE, if X means “situation with X” or “patient having X” (“additivity”)

**Example 2**

**Current Concept:**
- **Fully Specified Name:** Tetralogy of Fallot (disorder)
- **ConceptId:** 86299006

**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
Proper parts or taxonomic parents?

Example from Harold Solbrig

VSD  PVS  RVH  OA

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

Tetralogy of Fallot

Red Light  Yellow Light  Green Light

is-a  is-a  is-a

Traffic Light
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
‘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)’

‘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)’
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:
  
  – ‘$A_{\text{cond}}$ subClass of $B_{\text{cond}}$’ entails:  
    ‘$A_{\text{sit}}$ subClass of $B_{\text{sit}}$’
  
  – ‘$A_{\text{cond}}$ subClass of hasPart $B_{\text{cond}}$’ entails:  
    ‘$A_{\text{sit}}$ subClass of $B_{\text{sit}}$’
Review of 400 sample disorder concepts

- 4 experts: Kent Spackman, Alan Rector, Jean-Marie Rodrigues, Stefan Schulz

- Assessment of
  - FSN
  - Formal definitions
  - Children

Of a sample of disorder concepts

Schulz S, Rector A, Rodrigues JM, Spackman K. Competing Interpretations of Disorder Codes in SNOMED CT and ICD. Submitted to AMIA 2012
Table 1. Stratification of sample by distance from the root of the SNOMED CT hierarchy

<table>
<thead>
<tr>
<th>Distance from root</th>
<th>Number of concepts</th>
<th>Proportion</th>
<th>Number in Sample</th>
<th>Stratified distance</th>
<th>Proportion in Stratum</th>
<th>Concepts in Stratum</th>
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</thead>
<tbody>
<tr>
<td>0</td>
<td>1</td>
<td>0.0%</td>
<td>0</td>
<td>1</td>
<td></td>
<td></td>
</tr>
<tr>
<td>1</td>
<td>78</td>
<td>0.1%</td>
<td>0</td>
<td>1</td>
<td>14.2%</td>
<td>56</td>
</tr>
<tr>
<td>2</td>
<td>2127</td>
<td>3.3%</td>
<td>13</td>
<td>1</td>
<td>23.9%</td>
<td>96</td>
</tr>
<tr>
<td>3</td>
<td>7090</td>
<td>10.8%</td>
<td>43</td>
<td>1</td>
<td>26.9%</td>
<td>108</td>
</tr>
<tr>
<td>4</td>
<td>15657</td>
<td>23.9%</td>
<td>96</td>
<td>2</td>
<td>20.6%</td>
<td>82</td>
</tr>
<tr>
<td>5</td>
<td>17602</td>
<td>26.9%</td>
<td>108</td>
<td>3</td>
<td></td>
<td></td>
</tr>
<tr>
<td>6</td>
<td>13457</td>
<td>20.6%</td>
<td>82</td>
<td>4</td>
<td>9.8%</td>
<td>39</td>
</tr>
<tr>
<td>7</td>
<td>6392</td>
<td>3.5%</td>
<td>14</td>
<td>5</td>
<td>5</td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>2319</td>
<td>0.9%</td>
<td>4</td>
<td>5</td>
<td></td>
<td></td>
</tr>
<tr>
<td>9</td>
<td>577</td>
<td>0.9%</td>
<td>4</td>
<td>5</td>
<td>14.4%</td>
<td>58</td>
</tr>
<tr>
<td>10</td>
<td>92</td>
<td>0.1%</td>
<td>1</td>
<td>5</td>
<td></td>
<td></td>
</tr>
<tr>
<td>11</td>
<td>4</td>
<td>0.01%</td>
<td>0</td>
<td>5</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>C1: positive additivity of parent concepts</td>
<td>C2: Child concepts with additivity (count)</td>
<td>C2′: At least one child concept evident for Situation</td>
<td>C3: Evidence of Situation by fully specified name (ratio of positive ratings)</td>
<td>At least one positive rating at the concept level (C1 or C3), ratio of pos. ratings</td>
<td>At least one positive rating at concept + Child level (C1, C2′, C3), ratio of pos. ratings</td>
</tr>
<tr>
<td>------------------------------</td>
<td>------------------------------------------</td>
<td>------------------------------------------</td>
<td>-----------------------------------------------------</td>
<td>--------------------------------------------------------------------------</td>
<td>---------------------------------------------------------------------------------</td>
<td>-----------------------------------------------------------------------------------</td>
</tr>
<tr>
<td>Sample size</td>
<td>400</td>
<td>559</td>
<td>400</td>
<td>400</td>
<td>400</td>
<td>400</td>
</tr>
<tr>
<td>Number in sample</td>
<td>43</td>
<td>223</td>
<td>72</td>
<td>64</td>
<td>88</td>
<td>143</td>
</tr>
<tr>
<td>Ratio</td>
<td>10.8%</td>
<td>39.9%</td>
<td>18.0%</td>
<td>16.0%</td>
<td>22.0%</td>
<td>35.8%</td>
</tr>
<tr>
<td>Cohen’s Kappa for binary ratings</td>
<td>0.32</td>
<td>-</td>
<td>0.18</td>
<td>0.61</td>
<td>0.56</td>
<td>0.26</td>
</tr>
</tbody>
</table>
### Table 3. Influence of role groups

<table>
<thead>
<tr>
<th>Number of role groups (inherited plus asserted)</th>
<th>Number of concepts in the sample</th>
<th>Positive ratings at concept level (C1 or C3)</th>
<th>Percentage of positive ratings</th>
</tr>
</thead>
<tbody>
<tr>
<td>0</td>
<td>19</td>
<td>2</td>
<td>10.5%</td>
</tr>
<tr>
<td>1</td>
<td>194</td>
<td>39</td>
<td>20.1%</td>
</tr>
<tr>
<td>2</td>
<td>124</td>
<td>29</td>
<td>23.3%</td>
</tr>
<tr>
<td>3</td>
<td>52</td>
<td>15</td>
<td>28.8%</td>
</tr>
<tr>
<td>4 and more</td>
<td>11</td>
<td>3</td>
<td>27.3%</td>
</tr>
</tbody>
</table>

### Table 4. Influence of depth (stratified, see Table 1)

<table>
<thead>
<tr>
<th>Hierarchical level</th>
<th>Number of concepts in the sample</th>
<th>Positive ratings at concept level (C1 or C3)</th>
<th>Percentage of positive ratings</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>56</td>
<td>12</td>
<td>21.4%</td>
</tr>
<tr>
<td>2</td>
<td>96</td>
<td>21</td>
<td>21.8%</td>
</tr>
<tr>
<td>3</td>
<td>108</td>
<td>24</td>
<td>22.2%</td>
</tr>
<tr>
<td>4</td>
<td>82</td>
<td>19</td>
<td>23.1%</td>
</tr>
<tr>
<td>5</td>
<td>58</td>
<td>12</td>
<td>20.7%</td>
</tr>
<tr>
<td>Topology</td>
<td>Number of concepts in the sample</td>
<td>Positive ratings at concept level (C1 or C3)</td>
<td>Percentage of positive ratings</td>
</tr>
<tr>
<td>----------------------------------</td>
<td>----------------------------------</td>
<td>---------------------------------------------</td>
<td>--------------------------------</td>
</tr>
<tr>
<td>Terminal concepts (without children)</td>
<td>269</td>
<td>69</td>
<td>25.6%</td>
</tr>
<tr>
<td>Non-terminal concepts (with children)</td>
<td>131</td>
<td>19</td>
<td>14.5%</td>
</tr>
</tbody>
</table>
Results

• ~11% of disorder concepts represent situations rather than conditions
• For the rest, both interpretations are possible
• Agreement difficult – fuzzy boundary between what should be interpreted as a condition and what as a situation
Conclusions

• Redesigning the disorder hierarchy to exclude situation interpretation: huge effort, difficult decisions
• Leaving disorder code uncommitted: many existing subclass relations wrong
• Considering all disorder codes as denoting situation: consistent with current state of the disorder hierarchy, only rationale for concepts with single role groups
• If explicit reference to conditions: post-coordination