MEDINFO Panel II: Do we need a Common Ontology between ICD 11 and SNOMED CT to ensure seamless re-use and semantic interoperability?
Common ontology between the Foundation Component (FC) of ICD 11 and SNOMED CT

• Introduction: Stefan Schulz (2 min)

• Internal panelists Joint Advisory Group:
  • Kent Spackman IHTSDO (7 min)
  • Chris Chute WHO (7 min)
  • Jean marie Rodrigues (7 min)

• External panelists
  • Dipak Kalra EHR (7 min)
  • William Hogan Ontology (7 min)

• Panel Discussion (15 min)
• Questions from audience (15 min)
• Concluding remarks: Alan Rector (7 min)
Kent Spackman
Why Work Together?

- WHO classifications are used to capture snapshots of population health and to provide insight on trends.
- Often these aggregate data are derived from patient records that are increasingly held in electronic health records using SNOMED CT.
- We are working to make it easier to use them together.
Goals of linking SNOMED CT and ICD

- Record data once at the point of care
  - With fidelity to the clinical situation
  - At the clinical level of granularity and (un)certainty

- Re-use the coded EHR data for
  - Automated decision support
  - Mapping to classifications
    - Epidemiologic purposes
    - Health system management
    - Billing

- Avoid duplicate effort of double-coding
- Avoid (as much as possible) skewing of clinical data for billing purposes
Why work together?

- Avoidance of duplication of work and of public funding
  - IHTSDO (SNOMED) will require high-level nodes that aggregate more granular data
    - Use-cases include mutually exclusive, exhaustive,…
    - Sounds a lot like ICD
  - ICD-11 will require lower level terminology for aggregation logic definitions
    - Detailed terminological underpinning
    - Sounds a lot like SNOMED

Reference: Christopher G. Chute, MD DrPH
Why work together?

- Organizational benefits ['win-win-win']
  - WHO: Broaden scope of classification eg ICD 11 has potentially up to six views (linearizations) [use case specific classification]
  - IHTSDO: Enables the construction of these broadened scope views (linearizations) through SNOMED CT
  - Individual countries: Can achieve broadened and novel classification implementation and not disrupt patient care through the use of both standards

SNOMED & WHO Classifications are synergistic and not antagonistic
Why work together?

- Better national and international decision making
  - Linkages between SNOMED CT and ICD are international not local or national
  - Linkages between SNOMED CT and ICD are more mechanized [so transcription errors are avoided]
  - National classification data is produced in a more timely way direct from the patient record
  - Clear separation between *clinical* coding for individual patient care using SNOMED CT and *classification* coding for various statistical analyses using ICD
Terminology, classification and ontology

- Aren’t they all basically the same? NO
- Classification:
  - Exhaustive, mutually exclusive categories; double counting is bad; residual categories meaningful
  - Epidemiology, statistics & billing use cases (better than terminology or ontology alone)
- Terminology
  - Anything to be stated about the patient; double counting is good; residual categories meaningless
  - Electronic health record and decision support use cases (better than classification or ontology alone)
- Ontology
  - Formal definitions; no ambiguity; no unintentional vagueness
  - Defining meaning and cross-discipline interoperability (better than classification or terminology alone)
Do we need a common ontology?

- Re-phrased to avoid ambiguity of ‘ontology’:
  - Do we need a common representation of meaning?
  - Obviously, yes.
We need a common representation of meaning

- To achieve our goals, we need systems to interoperate by meaning
  - Not by surface forms (terms)
- To interoperate by meaning, we need clear definitions

- Clarify difficult and subtle meaning variations
  - anemia vs low hemoglobin vs genetic propensity to anemia
  - Pulmonary stenosis vs the class of all disorders with pulmonary stenosis as a necessary component
  - (an) aspirin vs (some) aspirin vs ASA molecule
Traditional Hierarchical System
ICD-10 and family

Mutually Exclusive
And Exhaustive
The ICD11 Foundation
a Semantic Network

Concept name
- Definition
  - Language translations
- Preferred string
  - Language translations
- Synonyms
  - Language translations
- Index Terms

Relationships
- Logical Definitions
- Etiology
- Genomic
- Location
  - Laterality
- Histology
- Severity
- Acuity

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Algorithmic Serialization of the Foundation Component into a *Linearization*

Mutually Exclusive And Exhaustive
Linearizations for multiple use-cases

Morbidity, Mortality, Quality, ...
Potential Future States (2007)

ICD-11

Ghost SNOMED

SNOMED

Ghost ICD
ICD11/SNOMED “Shared Layer” vs Common Ontology

- Classification ↔ Terminology dissonance
  - Focus on higher levels of abstraction

- Thesaurus ↔ Description Logic dissonance
  - Pragmatic hierarchies – parent-child
  - Formal logic where all is-a are always true

- Common Ontology is:
  - Based on Description Logics and “queries”
  - Provides a shared scaffolding for
    - The Foundation Layer of ICD 11 and SNOMED
    - Ignores residual categories of linearizations
Jean marie Rodrigues
SNOMED CT
Common Ontology Subset

ICD 11 Revision Architecture
Multi-layer system

Foundation Component
(Clinical knowledge:
signs, symptoms, causes, ...)

Ontology Component
(kinds / class definitions)

Mortality
Morbidty
Primary Care

Linearizations

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ICD 11 Revision Architecture: Linkage

Foundation Component
...
+
Links via queries

Ontology Component (kinds)

SELECT ?X WHERE
{?X subclassOf A
&
not (?X subclassOf B)}
Alignment of hierarchies

• (i) each class in the ICD-11 ontology core of the Foundation Component (FC) has to correspond to exactly one class in SNOMED CT

• (ii) the set of all parents (transitive closure of Is_a relations ) in Common Ontology must be included in the transitive closure of Is_a relations in SNOMED CT.

• (iii), the equivalence in meaning between aligned classes, as understood by users, will be assured by having common text definitions and descriptions, in addition to the formal axioms in description logic.
- Headers that aggregate entities of vocabulary, typically using plurals ("Diseases of...")

- Fine-grained parts of ICD, more specific than SNOMED CT

- Exclusion statements, only to be interpreted in linearizations

- Non Otherwise Specified

- Non-ontological content
  - (signs, symptoms, diagnostic criteria)
Hypertension in Prenatal period

SELECT ?CN WHERE (?CN SubclassOf Hypertension) MINUS (?CN SubclassOf Prenatal Disorder)
Objectives

• Common Ontology plus query on clinical SNOMED CT codes or on ICD 11 ontology based linearisations codes will give the same result

• Ability to reproduce exclusions by queries
Seamless re-use and Semantic Interoperability

Do we need a Common Ontology between ICD 11 and SNOMED CT to ensure seamless re-use and semantic interoperability?

• Dipak Kalra, EuroRec and UCL
  • on behalf of:
What are we trying to do, and why Heart Failure (HF)?

- Complex diagnostic, treatment and management issues
- Potential for effective self management
- Poor outcomes when badly managed but potential for good quality and improved expectancy, of life, when well managed
- General poor awareness in health care community and general population
- Care across range of domains
- Enormous capacity for miscommunication and diagnosis - dangerous
- Massive resource implications, recurrent hospitalisations, high health care contact - governments - commissioning
- Robust research basis and potential for further research

Source: John Cleland, Suzanna Hardman, SHN WP1
Mortality for patients hospitalised with HF

Inpatient Mortality 11.1%

- Cardiology ward 7.8%
- General medical 13.2%
- Other ward 17.4%

Source: John Cleland, Suzanna Hardman, SHN WP1
Is it heart failure? Key information needed for diagnosis

Suspected Heart Failure

Heart Failure Symptoms & Signs
- Orthopnoea / PND (improve/NA/FL, if NA - mild/mixed/severe/NA)
- Breathlessness at Rest (improve/NA, if NA - mild/mixed/severe/NA)
- Breathlessness when Walking (improve/NA, if NA - mild/mixed/severe/NA)
- Swelling of Ankles or Legs (improve/NA, if NA - mild/mixed/severe/NA)
- NYHA Class (I/II/III/IV/NA)
- Physical signs

Cardiac Biomarker
- NT-pro BNP - Value
- BNP - Value
- MR-pro ANP - Value

Cardiac Imaging
- Echo
- Other ECG CXR

Heart Failure Treatment
- Loop Diuretic
- Thiazide Diuretic
- ACE Inh.
- ARB
- Beta-Blocker
- MRA
- Warfarin

Left Ventricular Systolic Dysfunction
- Qualitative - (none/mild/mod/severe/NA) or LVEF value

Left Atrial Dilatation
- Qualitative - (none/mild/mod/severe/NA)
- Or LA dimension
- Or LA volume

Valve Disease
- Qualitative - (none/mild/mod/severe/NA)

Other Right/Left Heart Dysfunction
- Qualitative - (none/mild/mod/severe/NA)

Additional Information
- Age
- Sex
- Height / Weight
- Heart Rhythm - SR, SR with freq ES, AF, AFI, V.Pace, Haemoglobin
- Serum Creatinine

Current Diagnosis
- Heart Failure Excluded
- Heart Failure Confirmed
- Heart Failure Uncertain

Clinician completing HER (name and role)
Heart Failure Diagnosis Prompt: HeartBeat Disease Management Portal

Welcome to HeartBeat, Prof. Dipak Kaira. Your patient is Smith, John Henry (January 15, 1956).

Diagnosis Prompt

Fields marked * are mandatory

Criteria for Heart Failure

- Typical Symptoms:
- Shortness of Breath:
- Oedema:
- Other Symptoms:
- Signs of Heart Failure:
- Objective Evidence of Left Ventricular Dysfunction:
  - Systolic:
  - Describe:
- Other Cardiac Disease:
- Critical Investigations:
  - Outstanding:
- Symptoms Improved With:
- Treatment:
- Heart Failure Confirmed:

Should be confirmed only after taking an Echocardiogram, Cardiac Magnetic Resonance, Computed Tomography or Nuclear Imaging test.
Interoperable representations for diagnosis: archetype, OWL DL

\[ \text{shn:DiagnosisRecord} \text{ subClassOf } \text{shn:InformationItem} \text{ and } \\
\text{shn:hasInformationObjectAttribute} \text{ some } \text{shn:CertaintyAttribute} \text{C} \]
\[ \text{and } \text{btl:outcomeOf} \text{ some } \text{shn:DiagnosticProcedure} \text{D} \]
\[ \text{and } \text{btl:outcomeOf} \text{ some } (\text{shn:Interpreting} \text{ and } \text{btl:hasparticipant} \text{ some } \text{shn:Evidence}\text{E}) \]
\[ \text{and } \text{shn:isAboutSituation} \text{ only } \\
\text{(shn:ClinicalSituation}\text{X} \text{ and } \\
\text{and } \text{btl:temporallyRelatedTo} \text{ some } \text{shn:Time}\text{T} \]
\[ \text{and } \text{btl:causedBy} \text{ some } \text{shn:ClinicalSituation}\text{XY}) \]

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openEHR Heart Failure summary

HEART FAILURE CLINIC FIRST VISIT SUMMARY

Where not otherwise stated, elements are optional ([0..1]).

<table>
<thead>
<tr>
<th>Element</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>3. Presentation and symptoms</td>
<td></td>
</tr>
<tr>
<td>4. Physical Exam</td>
<td></td>
</tr>
<tr>
<td>5. Blood tests</td>
<td></td>
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<tr>
<td>6. Electrocardiography</td>
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<tr>
<td>7. Echocardiography</td>
<td></td>
</tr>
<tr>
<td>8. Other non-invasive Cardiac Imaging</td>
<td></td>
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<tr>
<td>9. Lung function</td>
<td></td>
</tr>
<tr>
<td>10. Invasive investigation</td>
<td></td>
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<tr>
<td>Assessment</td>
<td></td>
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<tr>
<td>Plan</td>
<td></td>
</tr>
</tbody>
</table>

Source: Ian McNicholl, openEHR, SHN WP4
William Hogan
The Good

• Division of labor

• Textual definitions

• Decoupling clinical data capture from administrative coding
The Good (continued)

• Classification is no longer “one size fits all”

• Machine-interpretable ICD exclusions

• Clearly separating information from what information is about
The Bad

- Shift from “ontology” to “representation of meaning”
  - What does it mean to “represent meaning”?
  - Can I take a picture or video of meaning?
  - Does my name designate a meaning or me?
  - The statement “brain located in skull” does not relate two meanings!

- Why not represent reality?
  - It is no concession to philosophical realism to say that medical records talk about real people, with real diseases
  - Nominalists believe people and diseases exist, too
  - From Ontology Summit 2013, endorsed by many (including ~3 of us in this room!)

  **Ontologies are human-intelligible and machine-interpretable representations of some portions and aspects of a domain**
The Bad (continued)

• Apparent maintenance of duplicate classes and identifiers
  • “each class…has to correspond to exactly one class…”
  • “aligned classes”
Questions

• How will classes of FC of ICD have description logic definitions?
  • Will there be anatomy, pathology, pathophysiology classes in FC/CO?
  • Will there be an upper ontology with standard relations?

• How will CO/FC/SNOMED CT be kept synchronized?
  • For example, after a change to a DL definition in SNOMED, how will CO get updated?
  • Or will the CO be the master, from which SNOMED and FC are derived?
  • Will FC classes have ICD11 identifiers?

• Is there a distinction between FC and CO as I am making here? What is the difference?