Tutorial: SNOMED CT Concept Model

Yongsheng Gao, Romin Khazai
Senior Terminologist, IHTSDO
SNOMED CT Implementation Showcase 2014, Amsterdam, the Netherlands
Content of tutorial

- Introduction to concept model
  - Concept model
    - Top level categories
  - Attributes
    - IS A – Subtype relationship
    - Defining attributes and values
  - Domains and Ranges
  - Primitive vs. Fully defined
  - Description Logic and auto-classification
  - Stated view vs. Inferred view, Normalised form
- Expression and syntax
  - Pre - coordinated expression
  - Post - coordinated expression
  - Compositional grammar
  - OWL
- Clinical finding/disorder concept model
- Procedure concept model
- Situation with explicit context concept model
Audience and Objectives

- **Audience**
  - All standards and terminology leaders, implementers and users

- **Objective**
  - To understand the importance and the key aspects of concept model for SNOMED CT content development and advanced implementation.
Concept model

- The model for specifying **logical definition** of concepts in SNOMED CT

- The concept model is based on formal description logic

- The editorial rules for the permitted attributes and values
Top level categories

- Clinical finding
- Procedure
- **Situation with explicit context**
- Observable entity
- **Pharmaceutical/biologic product**
- Physical object
- Staging and scales

- Body structure
- Organism
- Substance
- Specimen

- Physical force
- Event
- Environment or geographical location
- Social context

- Qualifier value
- Record artifact
- Special concept
- SNOMED CT Model Component
Attribute - IS A - subtype relationship

- All hierarchies are based on true subtype relationship specified explicitly by IS A attribute
  - Not member of
  - Singular vs. plural
  - Not part of

Joint pain, Pain in multiple joints, ankle pain
SEP model for anatomy:
  - Entire hand, entire thumb
  - Hand structure, thumb structure

- Poly-hierarchical structure - concept can have more than one parent
Poly-hierarchical structure of SNOMED CT

A screenshot of inferred IS A relationships of MRI guided biopsy in OWL Viz plugin of Protégé

SNOMED CT is a directed acyclic graph (DAG) in mathematics and computer science.
Defining attributes and values

IHTSDO standard diagram for representing concept definition
Defining attributes and values

IHTSDO standard diagram for representing concept definition
Role group

- Role grouping had been introduced in SNOMED CT to have clear semantics and correct inferences for complex concepts which involve more than one site, or more than one morphology.

- The attribute-value pairs are logically associated with each other by grouping them together (nesting) to indicate that certain roles must go together, e.g. which site goes with which morphology.

- Role group can be interpreted as **has-part** to take conditions or procedures expressed by expressions as values.

-Spackman KA, Dionne R, Mays E, Weis J. Role grouping as an extension to the description logic of ONTYLOG, motivated by entity modeling in SNOMED. AMIA Annual Symposium Proceedings; Washington, DC; 2002. pp. 712-716

Role group example

263063009 Fracture dislocation of joint (disorder)

108367008 Dislocation of joint (disorder)

125605004 Fracture of bone (disorder)

116676008 Associated morphology (attribute) -> 87642003 Dislocation (morphologic abnormality)

363698007 Finding site (attribute) -> 39352004 Joint structure (body structure)

363698007 Finding site (attribute) -> 272673000 Bone structure (body structure)

116676008 Associated morphology (attribute) -> 72704001 Fracture (morphologic abnormality)
Domain and Range

Specify constraints for concept model
▪ Domain - an attribute can be applied to
  ▪ Domain: Clinical finding
    ▪ Attributes: finding site, associated morphology
  ▪ Domain: Procedure
    ▪ Attributes: method, procedure site

▪ Range - an attribute can take values from
  ▪ Attribute: finding site
    ▪ Range: Anatomical or acquired body structure (<<)
  ▪ Attribute: associated morphology
    ▪ Range: Morphologically abnormal structure (<<)
  ▪ Attribute: method
    ▪ Range: Action (<<)
Primitive vs. Fully defined

▪ **Primitive**
  ▪ Concepts are defined by necessary conditions only
    ▪ **Disease**
      – Diabetes mellitus
        » Is a disorder of endocrine system
        » Is a disorder of glucose metabolism
    ▪ **Procedure**
      – Percutaneous transluminal angioplasty (procedure)
        » Is a Transluminal angioplasty
        » Is a Catheter procedure
        » Surgical repair procedure by device

▪ **Fully defined**
  ▪ Concepts are defined by necessary and sufficient conditions
    ▪ Fracture of bone
    ▪ MRI guided biopsy
Description Logic and auto-classification

- Description logic
  - A family of knowledge representation formalisms that define meaning of terms
  - Extensions of AL (attribute language) distinguished by the implemented constructs

- Benefits for using DL for terminology
  - Formal logic based semantics
  - Auto-classification provides inference for equivalence
  - Auto-classification provides inference for subsumption relationships
  - Consistency checking
Description logic - OWL 2 EL profile

- Designed for large bio-health ontologies
- Less expressive but more efficient for computing very large amount data
- SNOMED CT concept model follows EL profile
- Axioms/constructs in EL profile
  - Existential quantification, SOME
  - Intersection of classes, Conjunction, AND
  - Class inclusion – IS A
  - Domain restrictions
  - Range restrictions
  - Class equivalence
  - Object property inclusion – sub-attribute
Description logic - OWL 2 EL profile

- Constructs are not implemented in SNOMED CT
  - Class disjointness
  - Property equivalence
  - Transitive object properties
  - ...

- Constructs are not supported by EL profile
  - Universal quantification, ALL, ONLY
  - Disjunction, OR
  - Class negation, NOT
  - Inverse object properties
Incomplete list of DL reasoners

- **CEL** - http://lat.inf.tu-dresden.de/systems/cel/
- **FaCT++** - http://owl.cs.manchester.ac.uk/tools/fact/
- **HermiT** - http://hermit-reasoner.com
- **Pellet** - http://clarkparsia.com/pellet
- **Racer** - https://github.com/ha-mo-we/Racer
Expressions

- **Pre-coordinated expression**
  - A concept is represented by a single code
    - 73211009 |Diabetes mellitus|
    - 169069000 |Computed tomography of chest|
    - 7246002 |Kidney biopsy|
    - 12676007 |Fracture of radius|

- **Post-coordinated expression**
  - A concept is represented by combination of codes

Expression with role group refinement represents the same concept 12676007.

- 64572001 |Disease|:
  - {116676008 |Associated morphology| = 72704001 |Fracture|,
    363698007 |Finding site| = 62413002 |Bone structure of radius|}
Syntax: Compositional grammar

- **Simple expression**
  73211009 |Diabetes mellitus| or 73211009

- **Multiple focus concepts**
  217724009 |accident caused by blizzard| + 297186008 |motorcycle accident|

- **Expression with refinement**
  182201002 |hip joint|:
    272741003 |laterality| = 24028007 |right|

- **Expression with nested refinement**
  397956004 |prosthetic arthroplasty of the hip|:
    363704007 |procedure site| =
    (24136001 |hip joint structure|:
     272741003 |laterality| = 7771000 |left|)

- **Expression with role group refinement**
The OWL Web Ontology Language is a standard from W3C
SNOMED CT in OWL can be generated by Perl scripts that has been included in the international release

<table>
<thead>
<tr>
<th>DL Syntax</th>
<th>OWL Constructs</th>
<th>Manchester OWL Syntax</th>
</tr>
</thead>
<tbody>
<tr>
<td>∃ R.C</td>
<td>someValuesFrom</td>
<td>R some C</td>
</tr>
<tr>
<td>C ⊓ D</td>
<td>intersectionOf</td>
<td>C and D</td>
</tr>
<tr>
<td>∀ R.C</td>
<td>allValueFrom</td>
<td>R only C</td>
</tr>
<tr>
<td>¬ C</td>
<td>complementOf</td>
<td>not C</td>
</tr>
</tbody>
</table>
Stated/inferred views, normalised form

- **Stated view**
  - Attributes and values of a concept definition are stated by a modeler
  - Distributed in “stated relationship table” in release

- **Inferred view**
  - Attributes and values of concept definition are generated by description logic reasoner
    - Includes relationships inferred from the stated view
    - Redundant relationships removed
  - The relationship table in release is based on inferred view

- **Normalised form**
  - Only presents proximal primitive super-concepts and non-redundant defining relationships
  - Suitable for comparing expressions
<table>
<thead>
<tr>
<th>Stated view</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fracture</td>
</tr>
<tr>
<td>and</td>
</tr>
<tr>
<td>RoleGroup some (Finding site some femur and Associated morphology some fracture)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Inferred view</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fracture of lower limb and RoleGroup some (Finding site some femur and Associated morphology some fracture)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Normal form</th>
</tr>
</thead>
<tbody>
<tr>
<td>Disease and RoleGroup some (Finding site some femur and Associated morphology some fracture)</td>
</tr>
</tbody>
</table>
Stated view - fracture of femur
Normal form - Fracture of femur

- Disease
- Fracture of bone
- Fracture of lower limb
- Bone structure of lower limb
- Bone structure
- Femur structure

Finding site: Fracture of femur

Finding site: Fracture of bone

Finding site: Fracture of lower limb
Inferred view - Fracture of femur

Disease is a Finding site
Fracture of bone is a Finding site
Bone structure is a Finding site
Fracture of bone is a Fracture of lower limb
Fracture of lower limb is a Bone structure of lower limb
Fracture of femur is a Finding site
Finding site is a Bone structure

Transitive reduction vs. Transitive closure
# Clinical finding/disorder

<table>
<thead>
<tr>
<th>Attributes</th>
<th>Range of allowable values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Finding site</td>
<td>Anatomical or acquired body structure (head, kidney, artery, bone)</td>
</tr>
<tr>
<td>Associated morphology</td>
<td>Morphologically abnormal structure (fracture, stenosis, inflammation)</td>
</tr>
<tr>
<td>Associated with</td>
<td>Clinical finding, Procedure, Event …</td>
</tr>
<tr>
<td>Due to</td>
<td>Clinical finding, Event</td>
</tr>
<tr>
<td>After</td>
<td>Clinical finding, Procedure</td>
</tr>
<tr>
<td>Causative agent</td>
<td>Organism, Substance, Physical object, Physical force, Pharmaceutical/biologic product</td>
</tr>
<tr>
<td>Pathological process</td>
<td>Infectious process, Hypersensitivity process, Autoimmune</td>
</tr>
<tr>
<td>Clinical course</td>
<td>Courses (chronic, acute)</td>
</tr>
<tr>
<td>Occurrence</td>
<td>Periods of life (congenital, fetal period, childhood, adulthood)</td>
</tr>
<tr>
<td>Severity</td>
<td>Severities (mild, moderate, severe)</td>
</tr>
</tbody>
</table>
Pulmonary infection due to mycobacteria
Penicillamine nephropathy

- 236521003 Penicillamine nephropathy (disorder)
- 236519008 Chronic drug-induced renal disease (disorder)
- 363698007 Finding site (attribute) -> 64033007 Kidney structure (body structure)
- 246075003 Causative agent (attribute) -> 387235007 Penicillamine (substance)
## Procedure

<table>
<thead>
<tr>
<th>Attributes</th>
<th>Range of allowable values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Procedure site</td>
<td>Anatomical or acquired body structure</td>
</tr>
<tr>
<td>Procedure site - direct</td>
<td>Anatomical or acquired body structure</td>
</tr>
<tr>
<td>Procedure site - indirect</td>
<td>Anatomical or acquired body structure</td>
</tr>
<tr>
<td>Method</td>
<td>Action (insertion, imaging action, evaluation)</td>
</tr>
<tr>
<td>Procedure morphology</td>
<td>Morphologically abnormal structure</td>
</tr>
<tr>
<td>Direct morphology</td>
<td>Morphologically abnormal structure</td>
</tr>
<tr>
<td>Indirect morphology</td>
<td>Morphologically abnormal structure</td>
</tr>
<tr>
<td>Procedure device</td>
<td>Device</td>
</tr>
<tr>
<td>Direct device</td>
<td>Device</td>
</tr>
<tr>
<td>Indirect device</td>
<td>Device</td>
</tr>
<tr>
<td>Using device</td>
<td>Device</td>
</tr>
<tr>
<td>Using access device</td>
<td>Device</td>
</tr>
</tbody>
</table>
Intubation of stomach
Endoscopic biopsy
<table>
<thead>
<tr>
<th>Attributes</th>
<th>Range of allowable values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Using substance</td>
<td>Substance (e.g. contrast media)</td>
</tr>
<tr>
<td>Direct substance</td>
<td>Substance, Pharmaceutical/biologic product</td>
</tr>
<tr>
<td>Has intent</td>
<td>Intents (e.g. guidance, diagnostic, therapeutic; Imaging guided procedure)</td>
</tr>
<tr>
<td>Access</td>
<td>Surgical access values (e.g. )</td>
</tr>
<tr>
<td>Surgical approach</td>
<td>Procedural approach ()</td>
</tr>
<tr>
<td>Route of administration</td>
<td>Route of administration value (e.g. )</td>
</tr>
<tr>
<td>Has focus</td>
<td>Clinical finding</td>
</tr>
<tr>
<td>Priority</td>
<td>Priorities (e.g. )</td>
</tr>
<tr>
<td>Revision status</td>
<td>Primary operation, Revision – value, Part of multistage procedure</td>
</tr>
<tr>
<td>Recipient category</td>
<td>Person, Family, Community, Donor for medical or surgical procedure, Group</td>
</tr>
<tr>
<td>Using energy</td>
<td>Physical force (e.g. )</td>
</tr>
</tbody>
</table>
CT of Knee with contrast

- 702501008: Computed tomography of knee with contrast (procedure)
- 702502001: Computed tomography of lower limb with contrast (procedure)
- 241587004: Computed tomography arthrogram of knee (procedure)
- 446025003: Arthrography using contrast (procedure)
- 260686004: Method (attribute)
- 312251004: Computed tomography imaging - action (qualifier value)
- 424361007: Using substance (attribute)
- 385420005: Contrast media (substance)
- 405813007: Procedure site - Direct (attribute)
- 49076000: Knee joint structure (body structure)
## Situation with explicit context

<table>
<thead>
<tr>
<th>Attributes</th>
<th>Range of allowable values</th>
</tr>
</thead>
<tbody>
<tr>
<td>Subject relationship context</td>
<td>Person</td>
</tr>
<tr>
<td>Temporal context</td>
<td>Temporal context value</td>
</tr>
<tr>
<td>Associated finding</td>
<td>Clinical finding, Event, Observable entity</td>
</tr>
<tr>
<td>Finding context</td>
<td>Finding context value</td>
</tr>
<tr>
<td>Associated procedure</td>
<td>Procedure, Observable entity</td>
</tr>
<tr>
<td>Procedure context</td>
<td>Context values for actions</td>
</tr>
</tbody>
</table>
Subject context values and examples

- Subject of record (person)
- Person in family of subject (person)
  - Grandparent of subject (person)
  - Parent of subject (person)
    - Mother of subject (person)
    - Father of subject (person)
  - Spouse of subject (person)
    - Wife of subject (person)
    - Husband of subject (person)
  - Sibling of subject (person)
  - Child of subject (person)

- Examples:
  - Wife pregnant (situation)
  - Father smokes (situation)
  - Family history of neurological disorder (situation)
Family history of neurological disorder

297239000
Family history of neurological disorder (situation)

281666001
Family history of disorder (situation)

408731000
Temporal context (attribute)

410511007
Current or past (actual) (qualifier value)

246090004
Associated finding (attribute)

118940003
Disorder of nervous system (disorder)

408729009
Finding context (attribute)

410515003
Known present (qualifier value)

408732007
Subject relationship context (attribute)

444148008
Person in family of subject (person)
Temporal context values and examples

- In the past (qualifier value)
  - Past - time unspecified (qualifier value)
  - Past - time specified (qualifier value)
    - All times past (qualifier value)
- Current or specified time (qualifier value)
  - Specified time (qualifier value)
  - Current (qualifier value)
- Current - time specified (qualifier value)
- Current - time unspecified (qualifier value)

Example:
  - History of fracture (situation)
Finding context values and examples

- **Known (qualifier value)**
  - Known present (qualifier value)
  - Known possible (qualifier value)
    - Suspected (qualifier value)
    - NOT suspected (qualifier value)
  - Known absent (qualifier value)

- **Unknown (qualifier value)**

- **Example:**
  - Suspected diabetes mellitus (situation)
  - Sickle cell disease not suspected (situation)
  - No family history diabetes (situation)
Suspected clinical finding

473127005
Suspected diabetes mellitus (situation)

41769001
Disease suspected (situation)

408731000
Temporal context (attribute)

408732007
Subject relationship context (attribute)

408729009
Finding context (attribute)

246090004
Associated finding (attribute)

410512000
Current or specified time (qualifier value)

73211009
Diabetes mellitus (disorder)

410604004
Subject of record (person)

415684004
Suspected (qualifier value)
Clinical finding not suspected
Procedure context values

- Contraindicated (qualifier value)
- Indicated (qualifier value)
- Not indicated (qualifier value)
- Not done (qualifier value)
- Post-starting action status (qualifier value)
  - In progress (qualifier value)
    - Suspended (qualifier value)
    - Started (qualifier value)
  - Ended (qualifier value)
    - Discontinued (qualifier value)
    - Done (qualifier value)
- Pre-starting action status (qualifier value)
  - Not to be done (qualifier value)
    - Refused (qualifier value)
    - Canceled (qualifier value)
  - Organized (qualifier value)
  - To be done (qualifier value)
  - Under consideration (qualifier value)
  - Planned (qualifier value)
Procedure contraindicated

416704001 Dual X-ray absorptiometry scan contraindicated (situation)

444208005 Radiographic imaging procedure with explicit context (situation)

18932001 Procedure contraindicated (situation)

408730004 Procedure context (attribute)

410536001 Contraindicated (qualifier value)

408731000 Temporal context (attribute)

410511007 Current or past (actual) (qualifier value)

363589002 Associated procedure (attribute)

241686001 Dual energy X-ray photon absorptiometry (procedure)

408732007 Subject relationship context (attribute)

410604004 Subject of record (person)
Procedure declined

408567009 Computed tomography scan brain declined (situation)

168499009 Radiographic imaging procedure refused (situation)

363589002 Associated procedure (attribute) → 34227000 Computerized axial tomography of brain (procedure)

408731000 Temporal context (attribute) → 410511007 Current or past (actual) (qualifier value)

408730004 Procedure context (attribute) → 443390004 Refused (qualifier value)

408732007 Subject relationship context (attribute) → 410604004 Subject of record (person)
Procedure not done

164997006
Hypersensitivity skin test not done (situation)

165008002
Allergy testing not done (situation)

408731000
Temporal context (attribute)

410511007
Current or past (actual) (qualifier value)

363589002
Associated procedure (attribute)

268377001
Hypersensitivity skin testing (procedure)

408730004
Procedure context (attribute)

385660001
Not done (qualifier value)

408732007
Subject relationship context (attribute)

410604004
Subject of record (person)
Procedure to be done

698306007
Awaiting transplantation of kidney (situation)

129125009
Procedure with explicit context (situation)

408731000
Temporal context (attribute)

408732007
Subject relationship context (attribute)

408730004
Procedure context (attribute)

363589002
Associated procedure (attribute)

410512000
Current or specified time (qualifier value)

410604004
Subject of record (person)

385643006
To be done (qualifier value)

70536003
Transplant of kidney (procedure)
Procedure done

164996002
Hypersensitivity skin test done (situation)

165007007
Allergy testing done (situation)

408731000
Temporal context (attribute)

408730004
Procedure context (attribute)

36359002
Associated procedure (attribute)

408732007
Subject relationship context (attribute)

410512000
Current or specified time (qualifier value)

385658003
Done (qualifier value)

268377001
Hypersensitivity skin testing (procedure)

410604004
Subject of record (person)
QUESTIONS & DISCUSSION
Contact information

- Yongsheng Gao       yga@ihtsdo.org
- Romin Khazai       rkh@ihtsdo.org