Terminology Track –
An Introduction to SNOMED CT

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## Requirements for Meaningful Health Records

<table>
<thead>
<tr>
<th>Making health records electronic</th>
<th>... but it is only a partial solution; the real challenge is ...</th>
</tr>
</thead>
<tbody>
<tr>
<td>A significant step forward</td>
<td>Improves communication</td>
</tr>
<tr>
<td></td>
<td>Increases availability of relevant information</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Making health records meaningful</th>
<th>SNOMED CT represents clinical information meaningfully as part of a well-designed EHR</th>
</tr>
</thead>
<tbody>
<tr>
<td>Identifying significant facts in oceans of data</td>
<td>Enabling effective meaning-based retrieval</td>
</tr>
<tr>
<td>Linking the EHR to authoritative clinical knowledge</td>
<td></td>
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</table>

SNOMED CT represents clinical information meaningfully as part of a well-designed EHR.
Benefits of SNOMED CT Design

• Logical definitions
  • Common framework for consistent retrieval and processing
  • Defining relationships between concepts
  • Retrieval criteria based on the meaning of any related concept
• Optional post-coordination
  • Combining codes to add detail and specificity
  • Increases scope without 'combinatorial explosion' of codes
• Updates and versioning
  • Regular updates to International Release (six-monthly)
  • Support for incremental updates
  • Full historical view of all previous versions of SNOMED CT
• Comprehensive clinical scope
  • Reduces need to support multiple code systems
  • Common framework for consistent retrieval and processing
Introduction to SNOMED CT Components
Concepts, Descriptions and Relationships
SNOMED CT: Overview of the Logical Design

• Content components
  • Concepts
  • Descriptions
  • Relationships

• Localization mechanisms
  • Reference sets
  • Extensions

• Concept model
  • How relationships represent the computable meaning of each concept

• Expression model
  • How SNOMED CT can be used to represent meaningful information in clinical records, knowledge resources, etc.
Concepts

• Concepts are the central components of SNOMED CT

A SNOMED CT Concept is a clinical idea associated with a unique identifier
  • The meaning is specified by an association with a term known as the *fully specified name*
  • The link between the identifier and the meaning of that clinical idea is permanent and unchangeable
Descriptions

- All concepts have:
  - At least one *fully specified name*;
  - At least one *synonym*

- **Fully Specified Name (FSN)**
  - A phrase that unambiguously describes the concept
  - Contains a hierarchy tag (semantic tag) in brackets after the phrase to indicate the type of concept

- **Synonym**
  - A word or phrase commonly used by clinicians to refer to a concept
  - Used at user interface for search, selection and display

- Other description types
  - A *text definition* which provides descriptive text about the concept

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**Example:**

- **FSN**: appendectomy (procedure)
- **Synonym**: appendectomy, appendicectomy, excision of appendix
- **Concept ID**: 80146002
Descriptions

appendectomy (procedure)
appendectomy
appendicectomy
excision of appendix
appendektomi
appendektomi
appendektomi
apendicectomía
resección del apéndice ileocecal
apendicectomía (procedimiento)
Terms Do Not Need to be Unique

• The same term can be a synonym of more than one concept
  • In these cases there is more than one description containing the same term and each description refers to a different concept
  • The fully specified name can be checked to disambiguate terms that are associated with more than one concept

Example
• The term “fundus” is a short synonym which according to context can refer to one of four different body structures

<table>
<thead>
<tr>
<th>Synonym</th>
<th>Fully specified name</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fundus</td>
<td>Gastric fundus structure (body structure)</td>
</tr>
<tr>
<td>Fundus</td>
<td>Structure of fundus of eye (body structure)</td>
</tr>
<tr>
<td>Fundus</td>
<td>Structure of fundus uteri (body structure)</td>
</tr>
<tr>
<td>Fundus</td>
<td>Structure of fundus of gallbladder (body structure)</td>
</tr>
</tbody>
</table>
Concepts and Relationships

• Each concept is associated with other concepts by a set of relationships
• The relationships express defining characteristics of a concept

- Appendectomy
- Procedure
- Appendix

procedure site
is a subtype of
Subtype Hierarchy Relationships – IS A

- Subtype relationships
  - Create a hierarchy linking each concept to more general concepts
- Enable retrieval of specific concepts in response to general queries
Attribute Relationships

• Attribute relationships provide additional defining information about concepts

Examples
• Linking disorder concepts to sites, causative agents and morphological abnormalities
• Linking procedure concepts to sites and methods
All the Defining Relationships of Appendectomy

- Procedure site: Direct
  - Operation on appendix
- Method
  - Partial excision of large intestine
  - Appendectomy

- Is a subtype of:
  - Appendectomy
  - Appendix structure
  - Excision - action
Sets... all types of them

- **Reference Sets**
  - A SNOMED CT thing
  - A refset consists of a set of references to SNOMED CT components, like concepts, descriptions or relationships and is a published/released artefact

- **Value sets**
  - In this context, a FHIR thing
  - A uniquely identifiable set of concept representations from any coding system/terminology

- **Subsets**
  - General term
  - A set is a subset if all of its members are all contained in another set.

Within SNOMED CT, both value sets and subsets can be represented by refsets.
FHIR Terminology Services with SNOMED CT
FHIR Terminology Services Resources & Operations

**Naming System**
Defines the identifiers of a code or identifier system
- e.g. 2.16.840.1.113883.6.96 http://snomed.info/sct

**ConceptMap**
Mappings between code system concepts (in source and target value set contexts)
- e.g. 263204007 | Fracture of shaft of ulna | (SNOMED CT)
- 552.209A “Unspecified fracture of shaft of unspecified ulna, initial encounter for closed fracture” (ICD-10-CM)

**Code System**
Defines a set of concepts with a coherent meaning
- Code
- Display
- Definition
- e.g. SNOMED CT

**Value Set**
A selection of a set of codes for use in a particular context
- e.g. “SNOMED CT fracture codes”

**Element Definition**
Data element, binding characteristics and value set reference
- e.g. Condition.code

**Element (Instance)**
Coded Data Type
- code/
- Coding/
- CodeableConcept
- e.g. 263204007 | Fracture of shaft of ulna |
Terminology Services Operations - CodeSystem

- CodeSystem
  - **validate-code**: “Validate that a coded value is in the code system”
  - **subsumes**: “Test the subsumption relationship between code/Coding A and code/Coding B”
  - **lookup**: “Given a code/system, or a Coding, get additional details about the concept, including definition, status, designations, and properties”

- Examples:
  - look up SNOMED CT code 427623005
    ```
    ```
  - look up SNOMED CT code 427623005, showing the Normal form
    ```
    {{url}}/fhir/CodeSystem/$lookup?system=http://snomed.info/sct&code=427623005&property=normalForm&property=sufficientlyDefined&_format=json
    ```
Terminology Services Operations - ConceptMap

- **$translate:** “Translate a code from one value set to another, based on the existing value set and concept maps resources, and/or other additional knowledge available to the server”

- **Examples:**
  - list the ICD-10 target codes of any map records for SCTID 195967001
    ```
    ```
  
  - list the code which is the “SAME AS” the inactivated concept 134811001
    ```
    ```
**Terminology Services Operations - ValueSet**

- **$expand**: “The definition of a value set is used to create a simple collection of codes suitable for use for data entry or validation”.
- The following are parameters (on the url) useful for SNOMED CT:

<table>
<thead>
<tr>
<th>Parameter</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>fhir_vs</td>
<td>Implicit valueset of all concept ids in the edition/version. If the base URI is <a href="http://snomed.info/sct">http://snomed.info/sct</a>, this means all possible SNOMED CT concepts</td>
</tr>
<tr>
<td>fhir_vs=isa/[sctid]</td>
<td>all concept ids that are subsumed by the specified concept.</td>
</tr>
<tr>
<td>fhir_vs=refset</td>
<td>all concept ids that represent the reference sets that are explicitly defined in the specified SNOMED CT edition</td>
</tr>
<tr>
<td>fhir_vs=refset/[sctid]</td>
<td>all concept ids in the specified reference set</td>
</tr>
<tr>
<td>filter</td>
<td>a text filter that is applied to restrict the codes that are returned (this is useful in a UI context). The interpretation of this is delegated to the server in order to allow to determine the most optimal search approach for the context</td>
</tr>
<tr>
<td>displayLanguage</td>
<td>specifies the language using ISO code to be used for description in the expansions if the language is available in the edition referenced (by module id)</td>
</tr>
</tbody>
</table>
Operations – ValueSet $expand examples

- list the descendants of the SNOMED CT code 27624003
  ```
  ```

- list the descendants of the SNOMED CT code 27624003... in Spanish
  ```
  ```

- list the concepts with a description containing “heart attack”
  ```
  ```

- list the concepts within a given ValueSet/refset
  ```
  ```
Terminology Services Resources – ValueSet

● Creating a ValueSet resource

An HTTP PUT request eg:
curl -i --request PUT 
"https://snowstorm-alpha.ihtsdotools.org/fhir/ValueSet/GPS" --header "Content-Type: application/fhir+json" -d @gps.json

Update using HTTP PUT with the same id

Delete using HTTP Delete & id
Harness the Power of SNOMED CT

• With the *Expression Constraint Language (ECL)*

• SNOMED CT is a semantically rich ontology, concepts are organized into hierarchies. This can help find more general or more specific variations of a medical concept.

• Concepts also contain other attributes which allow making selections which cut across hierarchies.

• E.g. selecting disorder or procedure concepts using the location where they occur in the body.
Expression Constraint Language – Data Input

- Modern applications use advanced input fields like typeahead/incremental search when there are a large number of options.

- In a medical application ECL can be combined with a text search to limit typeahead matches to the relevant area of the hierarchy.

Typeahead field constrained by ECL to match Assessment Scales in SNOMED rather than Disorders. This makes finding the relevant concept easier!

Assessment Used:
- Asth|
- Asthma control test (assessment scale)
- Asthma control questionnaire (assessment scale)
Current Encounter

Date/Time
Wed Nov 20 2019 22:36:06 GMT+0100 (Central European Standard Time)

Reason for Encounter

heart

Diagnosis Note

Procedure

Laterality

Encounter Notes

Save
Expression Constraint Language – Data Analysis

When analyzing patient data ECL can be used to find the relevant concepts to match against patient records:

○ Find patients with any type of Diabetes:

<< 73211009 | Diabetes mellitus |

○ Find patients with any infectious disorder of the lung:

< 40733004 | Infectious disease | :
363698007 | Finding site | = << 39607008 | Lung structure |

○ Find patients with any behavior finding in the Nursing Health Issues Reference Set:

^ 733991000 | Nursing Health Issues Reference Set|
AND < 844005 | Behavior finding (finding)|
Expression Constraint Language – Syntax

- `memberOf = ^`
- `wildCard = *`
- `descendantOf = <`
- `descendantOrSelfOf = <<`
- `childOf = <!`
- `ancestorOf = >`
- `ancestorOrSelfOf = >>`
- `parentOf = >!`
- `conjunction = AND`
- `disjunction = OR`
- `exclusion = MINUS`
- `to = ..`

- `reverseFlag = R`
- `expressionComparisonOperator = = / !=`
- `numericComparisonOperator = = / != / <= / < / >= / >`
- `stringComparisonOperator = = / !=`
- `numericValue = [-/+] (decimalValue / integerValue)`

For more information see the ECL guide - [http://snomed.org/ecl](http://snomed.org/ecl)
Terminology Services Operations – Implicit ValueSets

- **ValueSet**
  - **$expand**: "The definition of a value set is used to create a simple collection of codes suitable for use for data entry or validation"

  ```
  fhir_vs=ecl/[ecl]  all concept ids that match the supplied (URI-encoded) expression constraint
  ```

- **Examples**:
  - list the descendants of chronic disease
    
    (ecl = < 27624003)
    ```
    {{url}}/fhir/ValueSet/$expand?url=http://snomed.info/sct?fhir_vs=ecl/%3C27624003
    ```
  
  - list any behavior finding in the Nursing Health Issues Reference Set
    
    (ecl = ^ 733991000 AND < 844005)
    ```
    {{url}}/fhir/ValueSet/$expand?url=http://snomed.info/sct?fhir_vs=ecl/%5E733991000%20AND%20%3C844005&_format=json
    ```
Terminology Services Resources – ValueSet again

- Creating a ValueSet resource containing an ECL query
  
  ```json
  "compose": {
    "include": [
      {
        "filter": [
          {
            "property": "constraint",
            "op": "=",
            "value": "< 763158003 |Medicinal product (product)| : << 762951001 |Has ingredient (attribute)| = 255641001 |Caffeine (substance)|, 732946004 |Has presentation strength denominator value (attribute)| = *"  }
        ]
      },
      {
        "system": "http://snomed.info/sct"
      }
    ]
  }
  ```
  
- Expand a pre-defined ECL query value set
  - `{{url}}/fhir/ValueSet/GPS/$expand`

- Expand and filter a pre-defined ECL query value set
  - `{{url}}/fhir/ValueSet/GPS/$expand?filter=heart`
Advanced SNOMED Retrieval
Differences between SNOMED CT and Classifications

- Classifications like ICD-9 and ICD-10
  - Valuable for statistical reporting
  - Limited value in an individual patient EHR

- SNOMED CT
  - Rich semantic structure adds meaning to the EHR
  - Adequate detail for clinical recording
  - Broad scope of coverage

- SNOMED CT maps to Classifications
  - Existing maps to ICD-9-CM and ICD-10
  - Enhanced rule-based mapping to ICD-10
  - Maps to ICD-10 are used by NLM for mapping to ICD-10-CM
Does patient have respiratory disorder?
Yes: code starts with “J”

Does patient have an infection?
No: code does not start with “A”

Does the disorder affect the lung?
Unknown: no easy way to tell this

Is the disorder caused by a virus?
Unknown: cannot identify specific virus
Supporting clinical queries – SNOMED CT

Does patient have a respiratory disorder?
- Yes: subtype of respiratory disorder

Does patient have an infection?
- Yes: subtype of infectious disease

Does the disorder affect the lung?
- Yes: finding site is lung structure

Is the disorder caused by a virus?
- Yes: causative agent is virus

Data entry

75570004 Viral pneumonia
The final furlong
More Terminology this week

Let’s Build – SNOMED CT

Next session here, 10:15

Using LOINC with FHIR with Daniel

Richard, 11:55 Today

$closure operation with Rob H

Elon, 9:00am Tomorrow

Terminology Analytics with John

Richard, 11:55 Tomorrow
Links to Further Information

• A summary of SNOMED CT components is provided in the SNOMED CT Starter Guide
  • [http://snomed.org/sg](http://snomed.org/sg)

• Working with SNOMED CT in FHIR
  • [http://build.fhir.org/snomedct.html](http://build.fhir.org/snomedct.html)

• SNOMED International’s open source, FHIR enabled SNOMED CT terminology server, Snowstorm
  • [https://github.com/IHTSDO/snowstorm](https://github.com/IHTSDO/snowstorm)

• Review other examples of concepts, descriptions and relationships by using an online browser
  • [SNOMED International’s SNOMED CT Browser](https://confluence.ihtsdotools.org/display/FHIR)

• SNOMED on FHIR working group
  • [https://confluence.ihtsdotools.org/display/FHIR](https://confluence.ihtsdotools.org/display/FHIR)
Questions?

Hands On Session Next
http://snomed.org/fhir-devdays

pwi@snomed.org