Overview

- Part 1. Introduction to subsets and reference sets
  - Subsets, value sets and reference sets
  - Meeting subset requirements with reference sets
  - Enhancing subsets with reference sets

- Part 2. Subset creation principles
  - Purpose
  - Principles
    - Context
    - Consistency
    - Check-it-again
  - Demo
What are …
- Subsets?
- Value Sets?
- Reference Sets?

How are they connected?

What is a Subset?

- A set whose members are all contained in another set
- This is a general definition not specific to SNOMED CT
- Example

Vowels are a subset of the set of alphabet characters
What is a Value Set?

- A set of concept representations used to represent values in a particular data item
  - This definition is not specific to SNOMED CT
  - May include codes from different code systems, coded refinements or predefined text strings
- Example
  - A specification for a problem list might define a single value-set including:
    - SNOMED CT disorder concepts
    - SNOMED CT expressions that are subtypes of disorder
    - ICD-10 classification codes representing diseases

What is a Reference Set?

- A standard SNOMED CT release file format
- A set of references to SNOMED CT components
- A way to add optional data to referenced components

<table>
<thead>
<tr>
<th>Attribute</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>id</td>
<td>Identification and versioning and modularization information</td>
</tr>
<tr>
<td>effectiveTime</td>
<td>An identifier of the reference set</td>
</tr>
<tr>
<td>active</td>
<td>An identifier of a referenced component</td>
</tr>
<tr>
<td>moduleld</td>
<td>Optional additional data items to meet specific requirements</td>
</tr>
<tr>
<td>refsetId</td>
<td></td>
</tr>
<tr>
<td>referencedComponentId</td>
<td></td>
</tr>
</tbody>
</table>

… attribute-n>
Subsets, Value Sets and Reference Sets – Connections

Reference sets

Simple reference sets can represent a subset of concepts that can be all or part of a value set. Additional data about individual members of a subset can also represent other types of reference set.

Practical Uses of Reference Sets

Add features to the terminology
Manage terminology changes
Map to/from other code systems
Specify language preferences
List and prioritize content
Represent subsets
Meeting Subset Requirements with Reference Sets

Practical Requirements for Subsets

- Searches and data entry
  - Restricting searches to a set of concepts or descriptions
  - Specifying descriptions to appear in a list of options
  - Constraining data entry to a specified set of concepts
- Information model and communications
  - Specifying value sets for particular data items
- Data retrieval and analysis
  - Specifying query criteria
- Other uses
  - Subsets can be used for any purpose that requires selective inclusion or exclusion of specified sets of components
Requirements for Representing Subsets

▪ A simple list of identifiers can represent a subset of SNOMED CT components
  ▪ For example:
    ▪ 82272006
    ▪ 6142004
    ▪ 55604004
    ▪ … etc. …

In a SNOMED CT reference set, the list of component identifiers is represented by the
  • referencedComponentId column

Subset Represented by a Simple Reference Set

<table>
<thead>
<tr>
<th>Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>82272006</td>
<td>Common cold</td>
</tr>
<tr>
<td>6142004</td>
<td>Influenza</td>
</tr>
<tr>
<td>55604004</td>
<td>Avian influenza</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>id</th>
<th>effectiveTime</th>
<th>active</th>
<th>moduleId</th>
<th>refsetId</th>
<th>referencedComponentId</th>
</tr>
</thead>
<tbody>
<tr>
<td>0000d6608941-005b-510b-b60b-07891b210365</td>
<td>20140731</td>
<td>1</td>
<td>19999999103</td>
<td>49999999102</td>
<td>82272006</td>
</tr>
<tr>
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<td>6142004</td>
</tr>
<tr>
<td>00006ac9-3797-5ed4-b37e-84cb158c388d</td>
<td>20140731</td>
<td>1</td>
<td>19999999103</td>
<td>49999999102</td>
<td>55604004</td>
</tr>
</tbody>
</table>
Requirements for Representing Subsets

1. A simple list of identifiers can represent a subset of SNOMED CT components.
2. For practical use, a subset needs to be identified and named so it can be referred to unambiguously.

When a subset is represented by a reference set:
- The subset is identified by the refsetId column.
- The refsetId refers to a concept that has:
  - A description that names the reference set.
  - A relationship that refers to the reference set type.
  
  For example, [is a] simple type reference set.

Subset Represented by a Simple Reference Set

<table>
<thead>
<tr>
<th>id</th>
<th>effective Time</th>
<th>active</th>
<th>moduleId</th>
<th>refsetId</th>
<th>referenced ComponentId</th>
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<td>1</td>
<td>19999999103</td>
<td>49999999102</td>
<td>55604004</td>
</tr>
</tbody>
</table>

Infectious disease simple reference set (EXAMPLE ONLY)
Requirements for Representing Subsets

1. A simple list of identifiers can represent a subset of SNOMED CT components.
2. For practical use, a subset needs to be identified and named so it can be referred to unambiguously.
3. Subset membership may need to change with future releases of SNOMED CT or due to evolving requirements for inclusion of different content.

When a subset is represented by a reference set:
- The standard SNOMED CT approach to versioning and modularization allows full tracking of changes.
- This uses the following columns:
  - id, effectiveTime, active, moduleId

Subset Membership Changes

Represented by a Simple Reference Set

Changes in July 2016

<table>
<thead>
<tr>
<th>Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>82272006</td>
<td>Common cold</td>
</tr>
<tr>
<td>6142004</td>
<td>Influenza</td>
</tr>
<tr>
<td>55604004</td>
<td>Avian influenza</td>
</tr>
<tr>
<td>3928002</td>
<td>Zika fever</td>
</tr>
</tbody>
</table>

Reference Set

<table>
<thead>
<tr>
<th>Id</th>
<th>effectiveTime</th>
<th>active</th>
<th>moduleId</th>
<th>refsetId</th>
<th>referenced ComponentId</th>
</tr>
</thead>
<tbody>
<tr>
<td>0000d6608041-005b-5108-b608-07891b210365</td>
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<td>49999999102</td>
<td>6142004</td>
</tr>
<tr>
<td>00006ac8-2f97-5ed4-b37e-84cb156c388d</td>
<td>20160731</td>
<td>0</td>
<td>19999999103</td>
<td>49999999102</td>
<td>55604004</td>
</tr>
<tr>
<td>00006ac8-2f97-5ed4-b37e-84cb156c388d</td>
<td>20160731</td>
<td>1</td>
<td>19999999103</td>
<td>49999999102</td>
<td>3928002</td>
</tr>
</tbody>
</table>
Requirements for Subsets

1. A simple list of identifiers can represent a subset of SNOMED CT components
2. For practical use, a subset needs to be identified and named so it can be referred to unambiguously
3. Subset membership may need to change with future releases of SNOMED CT or due to evolving requirements for inclusion of different content
4. It may be useful to define the membership of a subset using rules rather than a list of identifiers
   • This is called an intensional subset definition

Intensional Subset Definitions:
Substrate and Expansion

Substrate
• The set to which an intensional subset definition is applied

Intensional definition
• A set of rules that defines whether a member of the substrate is included in the subset

Expansion
• The result of applying an intensional definition to a given substrate

Not a misspelling! Intensional is not the same as intentional
Intensional Subset Definitions: Substrate and Expansion (Example)

Substrate
▪ All the letters in the English alphabet

Intensional definition
▪ A rule that defines which letters are included in the subset

Expansion
▪ The result of applying the intensional definition to a given substrate

English Alphabet
Letters between L and S
M, N, O, P, Q, R

Extensional and Intensional Subset Definitions

▪ Extensional subset definitions
  ▪ Subset membership defined by enumeration
    ▪ Identifying each of the members individually

SNOMED CT supports extensional subset definitions:
▪ Simple type reference sets with members identified by component identifiers (referencedComponentId)

▪ Intensional subset definitions
  ▪ Subset membership is defined by a set of rules
    ▪ The rules are expressed as a query that computes the membership of a subset

SNOMED CT supports intensional subset definitions:
▪ SNOMED CT expression constraints can represent rules that determine which concepts are in a subset
Intensional Subset Definitions: Substrate and Expansion (SNOMED CT Example)

Substrate
• A specified version of a SNOMED CT edition

Intensional definition
• A SNOMED CT expression constraint
• For example subtypes of infectious disease

Expansion
• Concepts in the specified version of the edition that comply with the constraint

SNOMED CT International Edition 2019-07-31

< infectious disease

Concepts that are subtypes of infectious disease

Enhancing Subsets with Reference Sets
Requirements for Ordered Lists

- Requirements for ordered lists of descriptions
  - Presenting terms in an order that is rational or helpful for a particular purpose in user interface controls including:
    - Simple lists
    - Drop down lists
    - Popup menus

- Requirements for ordered lists of concepts
  - Presenting concepts in an order that is rational or helpful for a particular purpose irrespective of the term displayed
  - Examples
    - Body parts that have a natural order
      - Fingers, cranial nerves and vertebrae
    - Enumerated values
      - Mild, moderate, severe

Requirements for Prioritization

- Prioritization is similar to order but multiple components may have the same rank
- Requirements for prioritization of concepts
  - Making it easier to find concepts that are most commonly used in a particular specialty, department or data entry scenario
  - Highlighting concepts that are preferred options for a particular purpose without preventing access to a wider selection of concepts
- Priorities may be implemented in various ways
  - Highlight high priority items
  - Only show prioritized items initially with a display more option
Ordered Component Reference Sets

- An ordered component reference set has an additional integer column to specify either an order or a priority group for members of the subset.

Ordered Component Type Reference Set Representing Display Order or Priority

<table>
<thead>
<tr>
<th>id</th>
<th>effective Time</th>
<th>active</th>
<th>moduleId</th>
<th>refsetId</th>
<th>referenced ComponentId</th>
<th>order</th>
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<tbody>
<tr>
<td>21356012</td>
<td>20160731</td>
<td>1</td>
<td>19999999103</td>
<td>809999999103</td>
<td>127053016</td>
<td>1</td>
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<tr>
<td>136021011</td>
<td>20160731</td>
<td>1</td>
<td>19999999103</td>
<td>809999999103</td>
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<td>20160731</td>
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<td>809999999103</td>
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<td>4</td>
</tr>
<tr>
<td>127053016</td>
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<td>1</td>
<td>19999999103</td>
<td>809999999103</td>
<td>21356012</td>
<td>5</td>
</tr>
</tbody>
</table>

The order column is an integer that specifies:
- Display order
- Priority rank: 1 (first) highest rank

- Thumb
- Second finger
- Third finger
- Fourth finger
- Fifth finger

Fingers sorted A-Z

21356012  Fifth finger
136021011 Fourth finger
138873019 Second finger
108884010 Third finger
127053016 Thumb
Requirements for Adding Information to a Referenced Component

- Displaying a textual note or comment about a listed or selected concept
  - For example, a guidance note on requesting a particular procedure or service
- Marking descriptions with indications of whether the terms they contain are acceptable or preferred in a specified language or dialects
  - For example, distinguishing term usage between different languages, dialects, local or specialty groups
- Marking particular concepts with specific values to provide processable and/or displayable information
  - For example, marking inactive concepts with indicators of the reasons for inactivating them

Annotation and Attribute Value Reference Sets

- An **annotation reference set** has an additional text column to add notes to subset members
- An **attribute value reference set** has an additional concept id column to refer to structured data
### Annotation Type Reference Set
Adding Unstructured Information to a Concept

<table>
<thead>
<tr>
<th>Id</th>
<th>Term</th>
<th>ModuleId</th>
<th>Referenced ComponentId</th>
<th>Annotation</th>
</tr>
</thead>
<tbody>
<tr>
<td>82272006</td>
<td>Common cold</td>
<td>1</td>
<td>199999999103</td>
<td></td>
</tr>
<tr>
<td>6142004</td>
<td>Influenza</td>
<td>1</td>
<td>199999999106</td>
<td></td>
</tr>
<tr>
<td>55604004</td>
<td>Avian influenza</td>
<td>1</td>
<td>199999999106</td>
<td></td>
</tr>
<tr>
<td>3928002</td>
<td>Zika fever</td>
<td>1</td>
<td>199999999106</td>
<td></td>
</tr>
</tbody>
</table>

The **annotation** column is a string that contains a free text note about the referenced component.

#### Example Entries

- **20160731** 1 199999999103 199999999106 82272006
  - Trivial just treat symptoms.

- **20160731** 1 199999999103 199999999106 6142004
  - Risk in elderly. Advise immunization in future years.

- **20160731** 1 199999999103 199999999106 55604004
  - Report to public health is suspected. Admit if any respiratory distress.

- **20160731** 1 199999999103 199999999106 3928002
  - Indicate if pregnant or potentially pregnant. Note travel in affected area.

### Attribute Value Type Reference Set
Adding Structured Data to a Concept

<table>
<thead>
<tr>
<th>Id</th>
<th>Effective Time</th>
<th>Active</th>
<th>ModuleId</th>
<th>Referenced ComponentId</th>
<th>ValueId</th>
</tr>
</thead>
<tbody>
<tr>
<td>...</td>
<td>20160731</td>
<td>1</td>
<td>900000000000207008</td>
<td>1720006</td>
<td>900000000000482003</td>
</tr>
<tr>
<td>...</td>
<td>20160731</td>
<td>1</td>
<td>900000000000207008</td>
<td>4515009</td>
<td>900000000000484002</td>
</tr>
<tr>
<td>...</td>
<td>20160731</td>
<td>1</td>
<td>900000000000207008</td>
<td>4961001</td>
<td>900000000000485001</td>
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<td>...</td>
<td>20160731</td>
<td>1</td>
<td>900000000000207008</td>
<td>5381002</td>
<td>900000000000483008</td>
</tr>
</tbody>
</table>

The **valueId** column refers to a concept that represent the additional data applicable to the concept.

#### Example Entries

- **1720006** [Dextroposition of aorta] (Duplicate)
- **4515009** [Keratoderma punctata] (Ambiguous)
- **4961001** [Psychiatric function] (Erroneous)
- **5381002** [Enterobacter gergoviae] (Outdated)
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  - Purpose
  - Principles
    - Context
    - Consistency
    - Check-it-again
  - Demo

Purpose

- Ensure that the patient data captured using your subset, results in good data quality in clinical records
  - Good quality data capture
  - Good quality data retrieval
Purpose

- Support that the patient data captured using your subset, results in good data quality in clinical records
  - Good quality data capture
  - Good quality data retrieval
- Dependent on
  - Context
  - Consistency
  - Checking

Context Principles

- Ensure that the subset reflects the intended clinical data
- Ensure that the subset reflects the surrounding information model
- Choose a strategy for when no concept is available
Correct Clinical Content

- Ensure that the subset reflects the intended clinical data
  - Understand the clinical details of the data element in which your subset should work
  - Incorrect assumptions about the clinical workflow of a data entry form can lead to poor quality data
  - Avoid using concepts which are used to group specific clinical meanings

Correct Clinical Content – Example 1

- Hernia
- Hernia
- Edema
- Edema
Correct Clinical Content – Example 2

“Remember to exclude grouper concepts”

Information Model Aware

- Ensure that the subset reflects the surrounding information model
  - A subset often populates a specific data element in an information model
  - Take into account the surrounding data elements
  - Consider the relationship between interface model and storage model
Information Model Aware - Example

Fracture type subset

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>4399870009</td>
<td>Open fracture of bone (disorder)</td>
</tr>
<tr>
<td>3598170006</td>
<td>Closed fracture of hip (disorder)</td>
</tr>
<tr>
<td>16114001</td>
<td>Fracture of ankle (disorder)</td>
</tr>
</tbody>
</table>

Fracture body site subset

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>71341001</td>
<td>Bone structure of femur (body structure)</td>
</tr>
<tr>
<td>33696004</td>
<td>Bone structure of ankle (body structure)</td>
</tr>
</tbody>
</table>

Information Model Awareness - Example

Fracture type subset

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>4399870009</td>
<td>Open fracture of bone (disorder)</td>
</tr>
<tr>
<td>423125000</td>
<td>Closed fracture of bone (disorder)</td>
</tr>
<tr>
<td>443395009</td>
<td>Compression fracture (disorder)</td>
</tr>
</tbody>
</table>
### Information Model Awareness - Example

Fracture with location subset

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>71620000</td>
<td>Fracture of femur (disorder)</td>
</tr>
<tr>
<td>16114001</td>
<td>Fracture of ankle (disorder)</td>
</tr>
</tbody>
</table>

### Strategy for no Concept

- Choose a strategy for when no concept is available
  - Subsets often require clinical concepts not in your edition
  - Options
    - Split the meaning across multiple data elements
    - Allow postcoordinated expressions
    - Create a new extension concept
    - Use a more general concept

Choose up front!
Strategy for no Concept - Example

Body weight in the morning

1. Split the meaning across multiple data elements
   - Observation: coded text, 27113001 [Body weight]
   - Time frame: coded text, 73775008 [Morning]

2. Allow postcoordinated expressions
   27113001 [Body weight]:370134009 [Time aspect] = 73775008 [Morning]

3. Create a new extension concept
   - xxxx [Body weight in morning]

4. Use a more general concept:
   - 27113001 [Body weight]

Consistency Principles

- Be consistent in hierarchy use
- Avoid overlapping content
Consistent Hierarchy

- Be consistent in hierarchy use
- Typically include concepts from only one hierarchy
  - E.g. Cardiology diagnoses and cardiology procedures are both in a cardiology patient journal, but mixing is uncommon
- Exceptions do exist
  - E.g. Reason for admission subset

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>233604007</td>
<td>Pneumonia (disorder)</td>
</tr>
<tr>
<td>86591008</td>
<td>Fall from ladder (event)</td>
</tr>
<tr>
<td>177141003</td>
<td>Elective cesarean section (procedure)</td>
</tr>
</tbody>
</table>

Consistent Hierarchy - Example

- Similar sounding concepts

- Not term search only. Consider meaning and hierarchy
Consistent hierarchy - Using ECL

1. Define an intentional subset using ECL
2. Adapt it manually

Avoid Overlaps

- Difficult for user to select between overlapping concepts
  - Leads to inconsistent selections by different users
  - This can lead to poor data quality
- Overlapping meaning can be caused by
  - Including both a subtype and supertype concept
  - Including two concepts with a common ancestor that represent two different aspects of the same idea
- Sometimes it may be appropriate to include both a subtype and a supertype
  - Consideration should be given to make it clear to the users which one to chose in different situations
Avoid Overlaps - Example

Medical problems

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>195967001</td>
<td>Asthma</td>
</tr>
<tr>
<td>86049000</td>
<td>Cancer</td>
</tr>
<tr>
<td>46635009</td>
<td>Diabetes mellitus type 1</td>
</tr>
<tr>
<td>54329005</td>
<td>Acute myocardial infarction of anterior wall</td>
</tr>
<tr>
<td>57054005</td>
<td>Acute myocardial infarction</td>
</tr>
</tbody>
</table>

Avoid Overlaps - Example

- Two concepts with a common ancestor represent two different aspects of the same idea

Pain subset

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Name</th>
</tr>
</thead>
<tbody>
<tr>
<td>25064002</td>
<td>Headache (finding)</td>
</tr>
<tr>
<td>247373008</td>
<td>Ankle pain (finding)</td>
</tr>
<tr>
<td>274663001</td>
<td>Acute pain (finding)</td>
</tr>
<tr>
<td>55145008</td>
<td>Stabbing pain (finding)</td>
</tr>
</tbody>
</table>

- Only subtypes of pain, but each concept in the subset refers to different characteristics of the pain
Check-it-again principles

- Check inclusions and exclusions
- Avoid careless mistakes
- Use reviewers with multiple perspectives

Inclusions and Exclusions

- Check inclusions and exclusions
  - Check inclusions
    - Logical definitions and descriptions
    - Display terms should be valid synonyms
  - Check exclusions
    - Concepts missing from the subset
Check Definitions - Example

- Check that the concept belongs to the intended hierarchy
- Check explicit rules about the attribute relationships

Pain quality subset

<table>
<thead>
<tr>
<th>Concept Id</th>
<th>Term</th>
</tr>
</thead>
<tbody>
<tr>
<td>279093005</td>
<td>Cramping pain</td>
</tr>
<tr>
<td>8708008</td>
<td>Sharp pain</td>
</tr>
<tr>
<td>36349006</td>
<td>Burning pain</td>
</tr>
<tr>
<td>55145008</td>
<td>Stabbing pain</td>
</tr>
</tbody>
</table>

- Check expression constraint
  - $< |\text{Clinical finding}|:|\text{Finding site}| = ^*$

Check Descriptions - Example

- Especially for primitive concepts, we rely on the fully specified names
  - Choosing between primitive sibling concepts may be challenging

$<\text{165041004} \mid \text{Forced expired volume (observable entity)}$
Avoid Mistakes

- Avoid careless mistakes
  - Many subset design processes are still to some degree manual
  - They are prone to human mistakes
  - Careless mistake examples
    - Concept goes into the wrong subset
    - Digit goes missing
    - Inactive concept
  - Do not assume that this will never happen to you

Avoiding Mistakes - Solutions

- Create automatic tests
  - Check concept ids in your subset are active
  - Check each concept’s fully specified name and preferred term
- SNOMED-specific subset development tool
  - Speed up the process of finding appropriate concepts
  - Avoid copy-paste errors
  - Use existing subsets or ECL as starting point
  - Browse for additional concepts
  - Validation checks and review processes
Multiple Perspectives

▪ Use reviewers with multiple perspectives
  ▪ Ensure team has necessary capabilities
    ▪ Clinicians, information modelers, terminologists
  ▪ Ensure each subset is thoroughly reviewed
    ▪ To avoid consistency problems
    ▪ To avoid careless mistakes

Demonstration
Demonstration

- SNOMED International Refset Management Tool
  - [http://snomed.org/tools](http://snomed.org/tools)
  - [https://refset.ihtsdotools.org](https://refset.ihtsdotools.org)

- Refset Management Tool
  - Enables the management & creation of reference sets against the International Edition of SNOMED CT and Member extensions
  - Provide a directory of existing reference sets that can be searched and downloaded to be used by others

Extensional Refset Example

- Rare Diseases concepts
  7199000, 9014002, 13213009, 16631009, 22053006, 23238000, 24700007, 30188007, 31323000, 44785005, 51615001, 58606001, 62067003, 63702009, 65389002, 65880007, 74911008, 75053002, 76670001, 77128003, 80651009, 82275008, 86044005, 128241005, 190794006, 195353004, 230418006, 230791000, 234542004, 236403004, 239928004, 252246005, 387732009, 387759001, 396338004, 417759001, 417357006, 423590009, 699310000, 716997004

- Concept to add
  190905008 | Cystic fibrosis (disorder) |
Intensional Subset Example

Questions?