SNOMED Clinical Terms Fundamentals

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Outline of presentation

- What is SNOMED CT?
- What is it for?
  - EHR, statements and expressions
- What kinds of things are represented?
- How is it organized?
- How does it fit into/with the Electronic Health Record?
- What additional components & essential materials should I know about?
  - Mapping, subsets, extensions, definitions, documentation, etc
- How can I suggest changes or improvements?
  - working groups, communication, governance
What is SNOMED CT?

- What is it?
- What does it do?
- Where did it come from?
- What is it made up of?
- How big is it?
What is SNOMED CT?

What is SNOMED Clinical Terms?

- **Name:**
  - Systematized Nomenclature of Medicine – Clinical Terms

- **Description:**
  - A work of clinical terminology

- **Main purpose:**
  - Coded representation of meanings used in health information
What is a clinical terminology?

- Terminology (ordinarily):
  - A structured collection of terms

- A clinical terminology
  - SNOMED CT is a terminology
    - consisting of terms used in health & health care
    - attached to concept codes with multiple terms per code
    - structured according to logic-based representation of meanings
Codes organized in a directed acyclic graph

- Each code is represented by a node in the graph
- Each relationship is an arrow
- There are no cycles
- Codes may have >1 outgoing arrow
  - if only 1 outgoing, you have a tree
  - but C.S. trees are upside down

What is SNOMED CT?
A compact disc with data files

What is SNOMED CT?

<table>
<thead>
<tr>
<th>CONCEPT ID</th>
<th>FULLY SPECIFIED NAME</th>
<th>CTV3 ID</th>
<th>SNOMED ID</th>
<th>ISPRI</th>
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<td>S922.</td>
<td>DD-3317D</td>
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<td>S93..</td>
<td>R-F5944</td>
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What does it do?

- Facetiously: Nothing, it just sits there.
  - Until incorporated into software systems
Organisational background and history

- International Health Terminology Standards Development Organisation (IHTSDO)
  - A not-for-profit organisation incorporated in Denmark
  - Member Nations provide the resources for coordinated development and release of terminology products
  - Owns and governs SNOMED CT and antecedent works

What is SNOMED CT?

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Where did it come from?

- **College of American Pathologists**
  - SNOMED 2 (1979)* Most widely adopted version in pathology systems worldwide
  - SNOMED 3 ‘International’ (1993)
- **United Kingdom – National Health Service**
  - Read Codes ‘4-byte’ (1984)
  - Read Codes 2 ‘5-byte’ (1988)* Still the most widely used codes in GP systems in the UK
  - Clinical Terms version 3 ‘CTV3’ (‘Read Codes’) (1999)
- **A true confluence**
  - All codes in SNOMED RT and CTV3 are included in SCT

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What is SNOMED CT?

What is it made of?

- **Components**
- **So-called “core” components:**
  - Concept codes
  - Descriptions (terms)
  - Relationships
- **Other components:**
  - Reference Sets (RefSets), RefSet Members
  - CrossMap Sets, Cross Maps, Cross Map Targets
Concept Codes

- One code per meaning, one meaning per code
  - Strings of digits, length 6 to 18 (most commonly 8 or 9 digits)
    - 22298006 means “myocardial infarction (MI)”
    - 399211009 means “past history of MI”
  - Meaningful, but without embedded meaning within the code

- Codes vs Concepts vs Real things
  - Concepts are in people’s heads
  - Codes are in the terminology
  - The codes refer to real things in the real world
A term string is a sequence of readable characters
  E.g. “immunosuppression”
A “description” is a term attached to a concept
These are two different “descriptions” that have the same term string:
  - immunosuppression → immunosuppressive therapy (procedure)
    - Description ID = 507152014
  - Immunosuppression → immunosuppression (finding)
    - Description ID = 63394015
What is SNOMED CT?

Relationships

- **Can be of several types:**
  - **Definitional:** necessarily true about the concept
  - **Qualifiers:** may be added to specialize the concept
  - **Historical:** provides a pointer to current concepts from retired
  - **Additional:** allows non-definitional information to be distributed
How big is it?

- 283,000 Active concept codes vs. 295,542 July 2011
- 732,000 Active terms (descriptions) vs. 769,428 July 2011
- 923,000 Active defining relationships vs.

  - If you spent 1 minute examining each description,
  - Working 40 hrs/week (2400 minutes/week), it would take
    - 305 weeks (~6 years) to examine all the active descriptions

- **Scale** is a major issue in developing, using and maintaining it
IHTSDO Structure and Governance

**GENERAL ASSEMBLY**

- Harmonization Boards
- Management Board
- Affiliate Forum

- Content Committee
  - Working Groups

- Technical Committee
  - Working Groups

- Research & Innovation Committee
  - Research Teams

- Quality Assurance Committee
  - Working Groups

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What is SNOMED CT?

Requirements, benefits, users and uses

- **What does it do?** facetiously: Nothing, it just sits there.
  - Until incorporated into software systems

- **Really:** It enables semantic interoperability, when implemented in an electronic health record
  - Supports implementation of electronic health records
  - Decision support systems
    - makes them systematically maintainable, sharable

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Terminology enables decision support

- Influenza vaccination reminder
- decision support program criterion:
  - chronic cardiorespiratory disorders
- patient record:
  - mild persistent asthma
Purpose of the terminology

- To provide a consistent way of indexing, storing, retrieving and aggregating clinical data from structured, computerised clinical records

- In order to support clinical care
  - Recording statement about health and health care of an individual patient
  - Retrieving those statements according to their meaning
    - At various levels of abstraction
    - For clinicians, patient, researchers, organisations and other computer systems
To represent health information
- Recorded by clinicians
- At the level of detail they prefer

To retrieve and analyse health information
- Retrieving those statements according to their meaning
  - At various levels of abstraction
  - For clinicians, patient, researchers, organizations and other computer systems
Who uses it?

- Users:
  - Clinicians
    - The end users of EHRs
  - System developers & vendors / suppliers
  - System implementers
    - Hospitals, clinics, laboratories, etc
  - Policy makers (government, professions, etc)
  - Researchers
What are the uses?

- **Representation of health information**
  - Indexing & retrieval of health information generally
  - Recording health & care of individuals
    - with fidelity to the clinical situation
  - Record retrieval & analysis based on meaning
    - Important for decision support applications

- **More specific examples**
  - Public health reporting – infectious diseases, cancer, biosurveillance
  - Reminders and alerts for preventive care
Expected benefits

- Reduction of errors
  - Elimination of errors of omission via “reminders”
  - Elimination of errors of commission via “alerts”
- Monitoring and responding to trends & problems in the health of populations
- Expanding knowledge of diseases, treatments and outcomes
Building blocks

- **Concepts**
  - The anchors for meaning

- **Descriptions**
  - Terms (strings of readable characters) used to express the meanings of the concepts in human language

- **Relationships**
  - Concept-to-concept links used to express information in computer processable language
    - First purpose: formal logical meanings
    - Other purposes: tracking retired concepts, representing facts that may vary, and supporting post-coordination
What kinds of things have codes?

- Organizing the world into types or classes is the work of “ontology”
- SNOMED focuses on classes that are useful in health & health care
  - Situations with explicit context
  - Procedures
  - Findings & disorders
  - Events
  - Body structures, anatomical or morphologically abnormal
  - Things that contribute to illness:
    - Organisms, substances, forces, objects, social context,
  - Other things important for health
Situations with explicit context

- Statements with “context” are those that express something about: who, whether, and/or when
  - Who:
    - is it about the subject of the record, or someone else?
  - Whether:
    - for findings, is the finding present, absent, or unknown?
    - for procedures, was it done, not done, or planned, …
  - When:
    - was the statement about the present, past, or perhaps future?
Situation examples

- Family history of diabetes mellitus
  - Who: a family member of the subject of record
- Tetanus booster given
  - Whether: yes, the procedure was done
- Past history of pelvic fracture
  - When: sometime in the past
Findings & Disorders

- **Findings:**
  - fever
  - low platelet count
  - rash
  - normal blood pressure
  - knee jerk reflex 2+/4+

- **Disorders:**
  - sickle cell disease
  - Fanconi’s anemia
  - heart disease
Procedures

- Any type of action done intentionally as part of the process of delivering health care
  - Patient education
  - Surgical procedure
  - Cholecystectomy
  - X-ray of left wrist
  - Discharge from nursing home
  - Family counseling
What kinds of things are represented?

Events

- Occurrences, things that happen (not necessarily unintentional)
  - exposure to toxin
  - death
  - environmental event
  - homicide
  - travel
Observables

- Qualities, properties and other observable entities
- “Incomplete findings”, that is, findings without their values
  - blood pressure
  - age
  - respired oxygen concentration
  - ability to walk (“whether able to walk”, not “able to walk”)
  - histologic grade
  - lesion size
What kinds of things are represented?

“Value hierarchies”

- Anatomy
- Morphology
- Drugs
- Substances
- Devices
- Organisms
- Physical objects
- Physical forces
- Social context
What kinds of things are represented?

**Miscellaneous**

- Staging, scales, & other qualifier values
- Record artifacts
- “Special” concepts
  - Inactive
  - Navigational
- Linkage concepts
  - Attributes
How are the codes organized?

1) Directed acyclic graph
   - logical subsumption relationships, with a single root
2) Attributes with values
   - Necessarily true “existential restrictions”
3) Description logic definitions of each concept code
   - Structured combinations of isa’s and attribute-value relationships
DAG (Directed Acyclic Graph)

- Called the “is a hierarchy”
  - Represents logical subsumption
  - A isa B means all instances of A are also instances of B

How is it organized?
Relationships: isa examples

- Lung disease
- Pneumonia
- Infectious disease
- Infectious pneumonia
- Viral pneumonia
- Virus

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Attribute-value relationships

- attribute – value
  - Logical “existential restriction”
  - A rel B means that for every instance of A, there is at least one relationship “rel” with a value that is an instance of B
Attribute example: causative agent

- Lung disease
- Pneumonia
- Infectious disease
- Infectious pneumonia
- Viral pneumonia
- Virus

How is it organized?

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How is it organized?

Description logic definitions

- **Viral pneumonia**
  - Is-a infectious pneumonia,
  - Causative agent = virus
Combining isa and attribute relationships

- Lung disease
- Infectious disease
  - Infectious pneumonia
  - Viral pneumonia
  - Virus
  - Pneumonia

How is it organized?

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Where do the codes go in a record?

- Statements in EHR’s
  - Electronic health record is made up of a series of statements
- Codes are the values for fields/slots in the information model
  - Codes from the terminology fill in some or all of the statement body
  - Information model determines the fields/slots available
- Coordination required to avoid gaps & overlaps between
  - terminology model
  - Information model
Additional components & features

- History tracking
- Cross Maps
- Subsets & Reference Sets
- Extensions
History Tracking

- Each component is permanent
  - But sometimes we correct errors. Then what?
- Components may be marked inactive
  - A component status field is included for each component
  - Additional two fields: release date, and change type
- Historical references link inactive components to current (active) ones
- Application maintenance can use the history tracking mechanisms to:
  - Update applications with new releases
  - Properly conduct retrievals on data containing inactive codes
Cross Maps

- Cross mapping involves linking SNOMED CT to other terminologies
- Each cross map has a direction
  - Either from SNOMED to the other, or vice versa.
- Archetypal crossmap is from SNOMED to ICD (9 or 10 or variant)
- Usual use case for ICD:
  - I have a record. It needs to be assigned the right code.
  - NOS and NEC are meaningful and necessary
- Usual use case for SNOMED:
  - I have a patient. I can document all that is relevant, and my EHR will attach codes to much of it (not all).
  - NOS and NEC are meaningless
Reference Sets (RefSets)

- Formerly called “subsets”
- Define groups of SNOMED components to be used for a particular purpose
- Types of RefSets
  - Simple
  - Group
  - Tagged
  - Language
  - Navigation
  - Aggregation
  - Prioritized
Navigational RefSet Example

How a GP might like to navigate to Influenza A virus from “virus”:

- Virus
  - Influenza A virus

Six levels deep if you try to navigate the is-a hierarchy:

1. Virus
2. RNA virus
   - Enveloped ssRNA virus without a DNA step in life-cycle
   - Enveloped ssRNA virus without a DNA step with multiple-stranded negative-sense genome
3. Family Orthomyxoviridae
4. Genus Influenzavirus A
5. Influenza A virus
Making SNOMED Usable

- Requires design and selection of usable components
- Requires hiding some of the complexity from the users
- Requires software that enables the users to accomplish their goals
Extensions

- SNOMED CT Identifiers
  - Called “SCTIDs”
- Allow for a part of the code to identify a Namespace
  - A namespace is controlled by an organization other than IHTSDO
- Extensions should add content that is not required in the international release
  - Realm-specific content:
    - Otero County (Colorado) jail cell number
    - leave granted under the Mental Health Act 1983 (England and Wales)
There is a wealth of documentation available, often overlooked:

- SNOMED CT User Guide
- SNOMED CT Technical Reference Guide
- SNOMED CT Technical Implementation Guide
  - http://www.ihtsdo.org/fileadmin/user_upload/doc/
- Abstract Logical Models & Representational Forms
- Transforming Expressions to Normal Forms
- Reference Sets – Technical Specification
- SNOMED Interchange Format
Distribution files

- SNOMED CT is distributed in three “core” distribution files
  - Concepts (one row per conceptID)
  - Descriptions (one row per descriptionID)
  - Relationships (one row per relationshipID)

- The international release consists of a common set of these core files
- Each national release centre may also provide extensions to each of these files
Browsers

A browser generally
- Displays the components of the terminology
- Allows searching
- Allows navigation along the hierarchies
- Some may have more specialized functions

There are numerous freely available browsers
- Some examples include:
  - CliniClue (www.cliniclue.com)
  - SNOB (snob.eggbird.eu)
Classifiers

- A description logic classifier can:
  - Compare two expressions for subsumption or equivalence
  - Structure the is-a hierarchy
  - Identify expressions (including definitions) that match a query

- A few well-known DL classifiers include:
  - Apelon’s Ontylog
  - FaCT++
  - CEL
  - Racer and RacerPro
  - Pellet
Influencing SNOMED & making improvements

- Governance & meetings
- www.ihtsdo.org
- Collaborative web site
- Working groups
  - Project groups
  - Special interest groups
Meetings

- **In person**
  - General Assembly meets twice per year
  - Pattern has been for committees, working groups and Management Board to meet at same venue

- **Teleconferences**
  - Each committee & WG has its own schedule
Working groups

- Two types: Project Groups & Special Interest Groups

- Project Groups:
  - Focused on a particular task and project plan
  - Duration limited
  - Open to participation
  - Resourced according to the project needs

- Special Interest Groups:
  - Focused on a particular interest, community, or topic area
  - May be ongoing
  - Open to participation
  - Reliant largely on voluntary participation
Working Groups

- For a complete list of current working groups, see:
  - www.ihtsdo.org/about-ihtsdo/governance-and-advisory/working-groups/

**Current SIGs include:**
- Anesthesia
- Concept Model
- Education
- IHTSDO Workbench Developer’s Implementation
- International Family Practice/GP
- International Pathology & Laboratory Medicine
- Mapping
- Pharmacy
- Translation

**Current Project Groups include:**
- Anatomy Model
- Collaborative Editing Roadmap
- Event, Condition and Episode Model
- Family/GP RefSet & ICPC Mapping
- Machine & Human Readable Concept Model
- Mapping SNOMED to ICD-10
- Migration
- Observable & Infectious Disease Model
- Request Submission
- Substance Hierarchy Redesign
- Translation Quality Assessment
Influencing SNOMED & making improvements

Web site & collaborative site

- www.ihtsdo.org
  - Calendar
  - Official announcements
  - Contact information
- Collaborative site (https://csfe.aceworkspace.net/sf/sfmain/do/home)
  - Working group discussions
  - Agendas & minutes of committee & WG meetings
  - Special interest group collaborative sites
  - Access freely available, registration required to join, email: collabnet@ihtsdo.org
Questions?