SNOMED CT Overview

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SNOMED Clinical Terms®
Overview

- SNOMED CT – background, history and purpose
- Requirement, benefits, users and uses
- Structure of SNOMED CT
- Important materials for working with SNOMED
- Influencing SNOMED and making improvements
- Adopting SNOMED CT in the EHR
What is SNOMED CT?

- **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 organised 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
A compact disc with data files

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<th>CTV3ID</th>
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Organisational background & 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
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
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
Terms & descriptions

- 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
Relationships

- Can be of several types:
  - Definitional: necessarily true about the concept
  - Qualifiers: may be added to specialise 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
- 732,000 Active terms (descriptions)
- 923,000 Active defining relationships

- 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

GENERAL ASSEMBLY

Harmonisation Boards

Management Board

Content Committee

Technical Committee

Research & Innovation Committee

Quality Assurance Committee

Research Teams

Working Groups

Working Groups

Working Groups

Working Groups

National E-Health Transition Authority
IHTSDO Enterprise Model:
Organisational Relationships

International release available to all Member Nations

Cooperation between & among countries

Principles of fairness, openness, transparency

Local/National Health Entities
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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
Terminology enables decision support

- Influenza vaccination reminder

- Decision support program criterion:
  - chronic cardiorespiratory disorders

- Patient record:
  - mild persistent asthma
Purpose of the terminology (1)

- 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 statements about health and health care of an individual patient
  - Retrieving those statements according to their meaning
    - At various levels of abstraction
    - For clinicians, patients, researchers, organisations and other computer systems
Purpose of the terminology (2)

- To represent health information
  - Recorded by clinicians
  - At the level of detail they prefer
    - Not forced into arbitrary categories
  - In coordination with a known information model

- To retrieve and analyse health information
  - According to its meaning, not merely its surface form
  - To enable:
    - Decision support for individual cases
    - Population-based aggregation and analysis
Supporting delivery of healthcare to individuals

- **Assisting structured data entry**
  - Interface Terminology features

- **Facilitating decision support**
  - Selective retrieval
  - Reference Terminology features

- **Enabling communication**
  - Meeting the processing needs of recipient systems
  - Simplifying specification of communication requirements

- **Involving the patient**
  - Possible future role for “consumer friendly terms”
Supporting delivery of healthcare to populations

- Identifying health needs
- Auditing the quality of service
- Supporting research
- Managing services using primary clinical data
  - Minimising need for specific administrative data entry by clinicians
Purpose of the terminology (3)

- **It is for building applications capable of:**
  - Recording statements about the health and health care of individuals
    - In a way that permits retrieval according to the **meaning** of the statements, rather than just the words used
  - Retrieving individual cases and groups of cases
    - To enable more automated and sophisticated decision support, epidemiology, and research
Purpose of the terminology (4)

- Semantic interoperability
  - The ability of computer systems to interchange data and to interpret and use the data according to its meaning, rather than just its surface form

- If all you need to do is send text for a person to read, a fax machine suffices (assuming the recipient speaks the language)

- But computers cannot safely and consistently interpret natural language, even if they have all the individual words, phrases or sentences
Expected benefits of semantic interoperability

- **Reduction of errors**
  - Elimination of errors of omission via reminders
  - Elimination of errors of commission via alerts

- **Management of costs**
  - Elimination of redundant testing and investigation

- **Monitoring and responding to trends & problems in the health of populations**

- **Expanding knowledge of diseases, treatments and outcomes**
Requirements to achieve these benefits:

- Automation of systems that deal with health information requires clinical data that:
  - Is recorded at the appropriate level of detail
    - not forced to be either too general or too specific
  - Is consistent over time and across boundaries
  - Can be transmitted without loss of meaning
  - Can be aggregated at more general levels, and along multiple different perspectives
  - Can be interpreted by automated systems
Differences between terminologies & classifications

- SNOMED CT is a clinical terminology
- ICD-10 is a classification
- They have different purposes, use cases, structures and functions
Typical classification use case

- **Actor**: health care records department personnel
- **Available information**: the clinical record as documented during the patient encounter
- **Timing**: after the patient has left
- **Goal**: identify a single primary discharge diagnosis for purposes of morbidity / mortality statistics OR for reimbursement purposes
- **Code system**:
  - Mono-hierarchical, with inclusion and exclusion criteria to avoid overlap
  - Not Otherwise Specified categories to code cases where the clinical record is not specific enough for the code system
  - Not Elsewhere Classified categories to code cases where the clinical record is more specific than the code system
Typical terminology use case

- **Actor**: health care provider
- **Available information**: whatever the provider can observe, test, or otherwise obtain
- **Timing**: while the patient is present
- **Goal**: document information about the patient and the encounter according to professional standards and to meet information needs of the health system
- **Code system**:
  - Multi-hierarchical terminology
  - Clinician can be as general or specific as they wish
  - Specificity is served by allowing multiple codes; combinations of codes; and, where necessary, free text entries (uncoded)
  - NOS and NEC are meaningless
What is SNOMED Clinical Terms?

- **A Coding Scheme**
  - Codes (identifiers) for concepts

- **A Classification**
  - Subtype multi-axial classification - yes
  - **Not** a statistical classification
    - Can be mapped to statistical classifications

- **A Reference Terminology?**
  - Yes, to the extent that its structure adds “reference features”
    - logic-based subsumption & retrieval

- **An Interface Terminology?**
  - Yes, to the extent that it supports “interface features” such as:
    - synonyms – including multi-dialect/multi-lingual support
    - subsets
    - navigation hierarchies
    - search support resources
Value of reference features

- Represent a large range of possible meanings in a consistent manner
- Specify the relationships between different concepts
  - Tuberculous pneumonia
    - Is a kind of lung infection
    - Is a kind of tuberculosis
    - Caused by mycobacterium tuberculosis
    - Site lung
    - Morphology inflammation
- Provide a semantic foundation
  - Facilitate reliable retrieval and aggregation
  - Independent of initial purpose of collection
Value of interface features

- Assist data entry
- Find concepts
  - Text searches
  - Navigation hierarchies
  - Qualifier options
- Express concepts with preferred or familiar terms
  - Preferred terms, Synonyms
- Define what concepts mean
  - Fully specified names, text definitions
- Provide consistency
  - Subsets of concepts common to a speciality or community
Misconceptions (1)

- You can solve interoperability if you just . . .
  - Get everyone to say it the same way
  - Make a huge list of everything people have said
  - Make a common data dictionary for fields that can be filled out (like “zip code” etc)
  - Aggregate multiple small lists from each clinical specialty
  - Use special-purpose natural language processing or Google-style indexing and search
Misconceptions (2)

- Achieving interoperability requires ordinary practitioners to . . .
  - Code everything
  - Use all of the terminology available
  - Document more than they really know
  - Document findings with more certainty than they actually have
Words alone are insufficient

1. There are national, regional and local variations in meaning of words and phrases (even within the same language)
2. Multiple meanings with the same "preferred name"
3. Combining words results in a meaning entirely different from the sum of the parts
4. Ambiguous shorthand and abbreviations are common
5. The same phrase means different things to different specialists
6. The same word or phrase means different things depending on what you are doing at the time
7. Significant differences in meaning are often obscured through use of the same word
8. Successful communication relies on making ontological distinctions that are ignored by common phrasing
9. Formal definitions are often at variance with common clinical usage
10. A general name often takes on a more specific meaning
11. A manifestation is often used to name the disorder in which it occurs
What SNOMED is not (1)

- SNOMED is not an attempt to
  - standardise the *language* of health care providers
  - get everyone to speak the same language

- “We are not the language police”

- Language is very changeable, fluid, and context-laden

- Clinicians (and people in general) determine what words mean by how they use them.

- SNOMED attempts to properly reflect the meanings given to words and phrases by people.
What SNOMED is not (2)

- SNOMED is not an attempt to independently create standard meanings for health professions or scientists
  - It follows existing published standards
  - It seeks to encourage scientific and professional groups to come to consensus and publish standards
- For example:
  - ISBT (International Society of Blood Transfusion) publishes terminology for Human Red Blood Cell Surface Antigens
  - SNOMED CT attempts to properly reflect and integrate the standard terminology so that it is usable with all others
What SNOMED is not (3)

- SNOMED is not a complete knowledge base
  - It represents *terminological knowledge* only
    - Definitional, always necessarily true of each instance
  - It does not represent *assertional knowledge*
    - Uncertain, variable from case to case

- Example: Appendicitis
  - Terminological knowledge:
    - Inflammation located in the appendix
  - Assertional knowledge:
    - associated with anorexia, nausea, abdominal pain initially central but moving to RLQ, rebound tenderness over McBurney’s point, and increased WBC
Who uses it?

- Users:
  - Clinicians
    - The end users of EHRs
  - Software designers & builders / System developers
  - Vendor / Suppliers of software systems
  - System implementers
    - Hospitals, clinics, laboratories, etc
  - The health care professions
  - Governments and other policymakers
  - 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
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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 by suggesting valid qualifiers
Codes, concepts, classes, instances

- Code:
  - In general, any sequence of characters used to represent something in a coding system
  - SNOMED Clinical Terms Identifier (SCTID):
    - A sequence of 6 to 18 digits that identifies a component

- Concept:
  - In general, an idea which has meaning. Through its meaning, a person can identify specific instances of the concept

- Class:
  - An abstract category of things sharing common features

- Instance:
  - A particular real member of a class
What kinds of things have codes?

- Organising 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 anaemia
- 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
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
“Value hierarchies”

- Anatomy
- Morphology
- Drugs
- Substances
- Devices

- Organisms
- Physical objects
- Physical forces
- Social context
Miscellaneous

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

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
Relationships: isa examples

- Lung disease
- Pneumonia
- Infectious disease
- Infectious pneumonia
- Viral pneumonia
- Infectious agent
- Virus
Attribute-value relationships

- attribute – value
  - Logical “existential restriction”
  - $A \mathsf{rel} B$ 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

Infectious disease

Causative agent

Causative agent

Causative agent
Viral pneumonia

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

- Lung disease
- Pneumonia
- Infectious disease
- Infectious pneumonia
- Viral pneumonia
- Infectious agent
- Virus
Where do the codes go in a record?

- **Statements in EHRs**
  - 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 maintainers 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
- Prioritised
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:

- Virus
  - 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
  - Family Orthomyxoviridae
  - Genus Influenzavirus A
    - Influenza A virus
Extensions

- **SNOMED CT Identifiers**
  - Called “SCTIDs”

- **Allow for a part of the code to identify a Namespace**
  - A namespace is controlled by an organisation other than IHTSDO

- **Extensions should add content that is not required in the international release**
  - **Realm-specific content:**
    - Multnomah County (Oregon) jail cell number
    - Leave granted under the Mental Health Act 1983 (England and Wales)
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Documentation

- There is a wealth of documentation available, often overlooked:
  - SNOMED CT User Guide
  - SNOMED CT Technical Reference Guide
  - SNOMED CT Technical Implementation Guide
  - Abstract Logical Models & Representational Forms
  - Transforming Expressions to Normal Forms
  - Reference Sets – Technical Specification
  - SNOMED Interchange Format

See www.ihtsdo.org/our-standards/technical-documents/
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 specialised functions

There are numerous freely available browsers

Some examples include:

- CliniClue
- SNOB
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
Terminology servers

■ Part of a modular software architecture

□ In general, the architecture modularises components so that terminology services are separate from application programs and patient data

□ Access to the terminology is achieved via an API
“Desiderata” for a clinical terminology
(Jim Cimino)

1. Content Completeness
2. Concept Orientation
3. Concept permanence
4. Identifiers with no embedded semantics
5. Polyhierarchy
6. Formal Definitions
7. Reject NEC
8. Multiple granularities
9. Multiple consistent views
10. Context representation
11. Evolve gracefully
12. Recognise redundancy

SNOMED Additional “Desiderata”
13. Understandable, Reproducible and Useful (URU)
14. User friendly terms – interface terminology
15. Component persistence and history tracking
16. Implementable in software applications
Is SNOMED CT ready?

- YES

- Based on the “desiderata” SNOMED CT is already superior to:
  - Any version of the Read Codes
  - Any other known clinical terminology

- More importantly, it is the focus of continuing international efforts to refine, extend and enhance every aspect of its quality

- SNOMED CT is not perfect but it has no equals
SNOMED CT Overview

- SNOMED CT – background, history and purpose
- Requirement, benefits, users and uses
- Structure of SNOMED CT
- Important materials for working with SNOMED
- Influencing SNOMED and making improvements
- Adopting SNOMED CT in the EHR
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 (1)

Two types:

- **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 (2)

- For a list of current working groups, see:
  - www.ihtsdo.org/about-us/working-groups/

Current SIGs include:
- Anesthesia
- Concept Model
- Education
- Mapping
- Nursing
- Primary Care
- Pathology and Laboratory Medicine
- Pharmacy
- Translation

Current Project Groups include:
- Collaborative working
- Education
- Machine & Human Readable Concept Model
- Mapping SNOMED to ICD-10
- Mapping standard processes
- Pharmacy content and model
- Pharmacy naming and editorial rules
- Request submission
- Substance hierarchy redesign
- Translation standard processes
Web site & collaborative site

- www.ihtsdo.org
  - Calendar
  - Official announcements
  - Contact information

- Collaborative site (currently: thecap.seework.com)
  - Working group discussions
  - Agendas & minutes of committee & WG meetings
  - Special interest group collaborative sites
  - Access freely available, registration required: support@ihtsdo.org
Are there any SNOMED implementations? NO (not per se), but ...

- There are EHR implementations in which SNOMED is used

**Major tasks:**

- Identifying the set of SNOMED codes and their associated display names to be used for each EHR function and/or field
- Linking or mapping from any legacy codes to SNOMED (for backwards compatibility with existing data)
- Integrating into reimbursement, billing and/or reporting functions
  - Usually requires establishing and maintaining a mapping from SNOMED to reimbursement codes
- Education of system maintenance staff & users
- Integration with decision support