SNOMED CT and research

Why should the SNOMED community and the research community care?

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Outline

• Before SNOMED CT
• “The early days of SNOMED CT” – Aspects of SNOMED CT research in PubMed 2001-2006
• KR-MED 2008 – Representing and sharing knowledge using SNOMED
• My own research journey with SNOMED CT
• Final thoughts
Before SNOMED CT
SNOMED CT was born in a research lab...

• Representation of clinical data (using logical definitions; KRSS)

• Concurrency control (distributed editing and reconciliation)

• Configuration management (versioning)
Evaluation of a “Lexically Assign, Logically Refine” Strategy for Semi-automated Integration of Overlapping Terminologies

Robert H. Dolin, MD, Stanley M. Huff, MD, Roberto A. Rocha, MD, PhD, Kent A. Spackman, MD, PhD, Keith E. Campbell, MD, PhD

Abstract  Objective: To evaluate a “lexically assign, logically refine” (LALR) strategy for merging overlapping healthcare terminologies. This strategy combines description logic classification with lexical techniques that propose initial term definitions. The lexically suggested initial definitions are manually refined by domain experts to yield description logic definitions for each term in the overlapping terminologies of interest. Logic-based techniques are then used to merge defined terms.
Other early research efforts

• Language independent concept representation systems
• Description logics (GRAIL)
• Separation between
  • concept model
  • linguistic mechanisms
• Terminology server
“The early days of SNOMED CT”

Aspects of SNOMED CT research in PubMed 2001-2006
“SNOMED CT” in PubMed
The early days – Building it

Mapping between SNOMED RT and Clinical terms version 3: a key component of the SNOMED CT development process.
Wang AY, Barrett JW, Bentley T, Markwell D, Price C, Spackman KA, Stearns MQ.
PMID: 11825284

The SNOMED clinical terms development process: refinement and analysis of content.
Wang AY, Sable JH, Spackman KA.
PMID: 12463944

The SNOMED RT Procedure Model.
PMID: 11825170 Free PMC article.
The early days – Assessing its value [1/2]

Improved coding of the primary reason for visit to the emergency department using SNOMED.
McClay JC, Campbell J.
PMID: 12463874

An evaluation of the usefulness of two terminology models for integrating nursing diagnosis concepts into SNOMED Clinical Terms.
PMID: 12467792

Evaluation of SNOMED coverage of Veterans Health Administration terms.
Penz JF, Brown SH, Carter JS, Elkin PL, Nguyen VN, Sims SA, Lincoln MJ.
PMID: 15360871

Evaluation of the content coverage of SNOMED CT: ability of SNOMED clinical terms to represent clinical problem lists.
Elkin PL, Brown SH, Husser CS, Bauer BA, Wahner-Roedler D, Rosenbloom ST, Speroff T.
PMID: 16770974
The early days – Assessing its value [1/2]

Use of SNOMED CT to represent clinical research data: a semantic characterization of data items on case report forms in vasculitis research.
Richesson RL, Andrews JE, Krischer JP.
PMID: 16799121

Development and evaluation of methods for structured recording of heart murmur findings using SNOMED-CT post-coordination.
Green JM, Wilcke JR, Abbott J, Rees LP.
PMID: 16501179

Hunscher D, Boyd A, Green LA, Clauw DJ.
PMID: 17238580
Free PMC article.
The early days – Assessing its quality

Ontology-based error detection in SNOMED-CT.
Ceusters W, Smith B, Kumar A, Dhaen C.
PMID: 15360859

Reliability of SNOMED-CT coding by three physicians using two terminology browsers.
Chiang MF, Hwang JC, Yu AC, Casper DS, Cimino JJ, Starren JB.
PMID: 17238317 Free PMC article.

The semantics of procedures and diseases in SNOMED CT.
Schulz S, Hanser S, Hahn U, Rogers J.
PMID: 16964349

Comparing the representation of anatomy in the FMA and SNOMED CT.
Bodenreider O, Zhang S.
PMID: 17238300 Free PMC article.
The early days – Mapping/integration

**Terminological mapping for high throughput comparative biology of phenotypes.**
Lussier YA, Li J.
PMID: 14992504  Free PMC article.

**Integrating SNOMED CT into the UMLS: an exploration of different views of synonymy and quality of editing.**
Fung KW, Hole WT, Nelson SJ, Srinivasan S, Powell T, Roth L.
PMID: 15802483  Free PMC article.

**Standardized nursing language in the systematized nomenclature of medicine clinical terms: A cross-mapping validation method.**
Lu DF, Eichmann D, Konicek D, Park HT, Ucharattana P, Delaney C.
PMID: 16980782
The early days – Use as a knowledge source

Classifying diseases with respect to anatomy: a study in **SNOMED CT**.
Burgun A, Bodenreider O, Mougin F.
PMID: 16779008  Free PMC article.

Inter-patient distance metrics using **SNOMED CT** defining relationships.
Melton GB, Parsons S, Morrison FP, Rothschild AS, Markatou M, Hripcsak G.
PMID: 16554186  Free article.

Contribution to terminology internationalization by word alignment in parallel corpora.
Deléger L, Merkel M, Zweigenbaum P.
PMID: 17238328  Free PMC article.
The early days – Summary

• A few SNOMED CT papers at the very beginning ("building it")
• Adoption by the research community
  • Applied research – Coverage, utility
    • Mostly clinical communities
    • Pushing the envelope – e.g., clinical research
  • “Basic” research
    • Terminology research – New methods for quality assurance, mapping
    • Uses beyond terminology – Semantic similarity, knowledge source
• Sustained research: ~50 articles each year in PubMed
KR-MED 2008

Representing and sharing knowledge using SNOMED
KR-MED 2008

Representing and sharing knowledge using SNOMED

May 31 - June 2, 2008, Phoenix, Arizona, USA

Conference organized by the International Health Terminology Standards Development Organisation (IHTSDO®) and the Working Group on Formal (Elec.) Medical Knowledge Representation of the American Medical Informatics Association (AMIA).

Collocated with the 2008 AMIA Spring Congress.

The vision of a universal clinical terminology, covering a broad range of health-related domains and meeting the needs of all health professionals has stimulated numerous health informatics research activities in the last two decades.

SNOMED CT® (Systematized Nomenclature of Medicine-Clinical Terms) is emerging as a comprehensive, multilingual clinical healthcare terminology, since 2007 under a new international ownership. KR-MED 2008 will follow up the successful first SNOMED conference organized in Copenhagen in October 2006, and will address health policy makers, clinicians, nurses, system developers, informatics researchers, computer scientists, terminologists and translators. KR-MED 2008 is also the third event of the AMIA working group KR-MED. It will therefore challenge the current state and the planned roadmap of SNOMED CT development from a perspective of knowledge representation and formal ontologies.

A number of prominent invited speakers will provide an overview of current efforts and developments in the context of clinical terminologies and state-of-the-art ontology and terminology development.

Sponsors:
### May 31

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<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>1:30 pm</td>
<td><strong>Tutorials</strong></td>
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<tr>
<td></td>
<td>1. Terminological, Ontological and Knowledge Representation Aspects</td>
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<tr>
<td></td>
<td>Stefan Schulz, Kent Spackman</td>
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<td>2. Translating SNOMED CT</td>
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<td>Ulrich Andersen, Asta Hay</td>
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<td>3:00 pm</td>
<td><strong>Coffee Break</strong></td>
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<td>3:30 pm</td>
<td><strong>Tutorials (continued)</strong></td>
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<td>5:00 pm</td>
<td><strong>Opening</strong></td>
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<td>5:30 pm</td>
<td><strong>Invited Speech</strong></td>
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<td>Building medical ontologies using Description Logics: what does it buy us?</td>
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<td>Franz Baader, University of Dresden, Germany</td>
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<td>6:30 pm</td>
<td><strong>Reception</strong></td>
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### June 1st

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<tr>
<th>Time</th>
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<tr>
<td>8:30 am</td>
<td>Invited Speech</td>
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<td></td>
<td>Managing Clinical Terminology in a Post-Classification Era</td>
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<td>James R. Campbell, University of Nebraska</td>
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<td>9:30 am</td>
<td>Coffee Break</td>
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<tr>
<td>10:00 am</td>
<td>Scientific Session I (two parallel sessions)</td>
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<td></td>
<td><strong>Session I A - Formalization and Classification</strong></td>
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<td></td>
<td>1. Debugging SNOMED CT Using Axiom Pinpointing in the Description Logic EL+</td>
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<td>Boontaewee Suntisiravaraporn and Franz Baader</td>
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<td>2. Exploiting Fast Classification of SNOMED CT for Query and Integration of Health Data</td>
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<td>Michael Lawley</td>
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<td>3. Why do it the hard way? The Case for an Expressive Description Logic for SNOMED</td>
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<td>Alan Rector, Sebastian Brandt and Jay Kola</td>
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<td><strong>Session I B - Applications</strong></td>
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<td>1. Leveraging SNOMED CT with a General Purpose Terminology Server</td>
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<td>Robert Welda, Jack Bowle, Robert McClure and David Sporzel</td>
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<td>2. LinkBase® and SNOMED: some distinct features and impact on NLP</td>
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<td>Marla van Gurp, Marnix Holvoet and Mariana Casella dos Santos</td>
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<td>3. SNOMED CT: Browsing the Browsers</td>
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<td>Jeremy Rogers and Olivier Bodenreider</td>
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<td>3:30 pm</td>
<td>Scientific Session II (two parallel sessions)</td>
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<td><strong>Session II A - Mapping and comparing</strong></td>
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<td>1. Comparing SNOMED CT and the NCI Thesaurus through Semantic Web Technologies</td>
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<td></td>
<td>Olivier Bodenreider</td>
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<td>2. Exploratory Reverse Mapping of ICD-10-CA to SNOMED CT</td>
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<td></td>
<td>Dennis Lee and Francis Lau</td>
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<td><strong>Session II B - SNOMED CT and Information Models</strong></td>
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<td>1. Interoperability of Data Models and Terminology Models: Issues with using the SNOMED CT terminology</td>
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<td>Rahil Qamar Siddiqui, Jay Kola and Alan Rector</td>
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<td>2. Essential SNOMED: Simplifying SNOMED-CT and supporting Integration with Health Information Models</td>
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<td>Peter Maclsaac, Donald Walker, Rachel Richeson, Heather Grain, Peter Elkin and Jon Patrick</td>
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June 2nd

8:00 am  **Scientific Session III (two parallel sessions)**

**Session III A - Representation**

1. Comparing the Effects of Two Semantic Terminology Models on Classification of Clinical Notes: A Study of Heart Murmur Findings
   Guoqian Jiang and Christopher G Chute

   Howard Goldberg, Vipul Kashyap and Kent Spackman

3. Representing clinical information using SNOMED Clinical Terms with different structural information models
   David Markwell, Laura Sato and Edward Cheetham

**Session III B - Mapping and Subsetting**

1. Strategies for Updating Terminology Mappings and Subsets
   John Mapoles, Corey Smith, Jane Cook and Brian Levy

2. Using SNOMED CT as a Mediation Terminology: Mapping Issues, Lessons Learned, and Next Steps Toward Achieving Semantic Interoperability
   Sarah Maulden, Patty Greim, Omar Boulhaddou, Pradnya Warnekar, Laura Megas, Fola Parrish and Michael Lincoln

3. Using SNOMED CT For Translational Genomics Data Integration
   Joel Dudley, David P. Chen and Atul Butte

10:00 am  **Scientific Session IV (two parallel sessions)**

**Session IV A**

1. A Methodology for Encoding Problem Lists with SNOMED CT in General Practice
   Francis Lau, Raymond Slinkus and Dennis Lee

2. Post-Co-Ordination in the Mapping of Interface Terms of a Clinical Wound Documentation System to SNOMED CT
   Martin Boeker, Stefan Schulz and Thilo Schuler

3. Using SNOMED CT Concepts for PAIRS
   Modern Rao

**Session IV B - State of Affairs In SNOMED CT Member Countries (panel)**

11:30 am  **Invited Speech**

SNOMED CT Adoption: Lessons and Challenges
   Howard S. Goldberg, Partners Healthcare

12:30 pm  **Closing Remarks**
KR-MED 2008 – Summary

• The medical informatics research community was very engaged and driving the agenda (Sponsored by AMIA)
• Even attracted the computer science community
• The only KR-MED entirely dedicated to SNOMED CT
  • Previous KR-MED had some SNOMED CT papers
  • Later on, ICBO has been dominated by OBO, with less clinical ontology and occasional SNOMED CT papers
    • Until ICBO 2019, where SNOMED International was a sponsor
My own research journey with SNOMED CT
“SNOMED CT” in PubMed – Bodenreider edition
Review/education

Biomedical ontologies in action: role in knowledge management, data integration and decision support.
Bodenreider O.
Yearb Med Inform. 2008;67-79.
PMID: 18660879  Free PMC article.

Recent Developments in Clinical Terminologies - SNOMED CT, LOINC, and RxNorm.
Bodenreider O, Cornet R, Vreeman DJ.
PMID: 30157516  Free PMC article.
Quality assurance

Investigating subsumption in **SNOMED CT**: an exploration into large description logic-based biomedical terminologies.
Bodenreider O, Smith B, Kumar A, Burgun A.
PMID: 17241777  Free PMC article.

Using SPARQL to Test for Lattices: application to quality assurance in biomedical ontologies.
Zhang GQ, Bodenreider O.
PMID: 25699294

Auditing **SNOMED CT** hierarchical relations based on lexical features of concepts in non-lattice subgraphs.
Cui L, Bodenreider O, Shi J, Zhang GQ.
PMID: 29274386  Free PMC article.
“before it was cool”/trial balloons [1/2]

Comparing the representation of anatomy in the FMA and SNOMED CT.
Bodenreider O, Zhang S.
PMID: 17238300 Free PMC article.

Issues in mapping LOINC laboratory tests to SNOMED CT.
Bodenreider O.
PMID: 18999311 Free PMC article.

Using SNOMED CT in combination with MedDRA for reporting signal detection and adverse drug reactions reporting.
Bodenreider O.
PMID: 20351820 Free PMC article.
“before it was cool”/trial balloons [2/2]

Coverage of rare disease names in standard terminologies and implications for patients, providers, and research.
Fung KW, Richesson R, Bodenreider O.
PMID: 25954361  Free PMC article.

Interoperability between phenotypes in research and healthcare terminologies--Investigating partial mappings between HPO and SNOMED CT.
Dhombres F, Bodenreider O.
PMID: 26865946  Free PMC article.
My own journey – Summary

• Interesting collaborations
• Lots of fun investigations
  • Cool methods
  • Applied at scale
• Privileged to investigate issues before they became mainstream
• Minor impact on SNOMED CT overall
  • Not integrated in the SNOMED CT development process (e.g., QA)
Final thoughts

*Why should the SNOMED community and the research community care?*
Different communities

• Two active, but separate communities with different goals and cultures
  • SNOMED CT community of practice
    • Results-driven, pragmatic
    • On-time, at-scale
    • Long-term
    • SNOMED CT Expo
  • Research community (biomedical informatics)
    • Methods-driven
    • Toy examples, limited scale
    • Short-term (e.g., grant)
    • Scientific conferences
How the research community perceives SNOMED CT

• Pros
  • Rich and voluminous material
  • Interesting problems for driving research

• Cons
  • Constraints imposed by the license
    • Cannot distribute materials freely
  • Perceived lack of interest from the SNOMED CT community of practice
  • Lack of a process for engaging the research community*
  • More interested in working with other communities of practice or SDOs

*until recently
How the SNOMED CT community perceives research*

• Pros
  • Potential source of innovation and experimentation

• Cons
  • Not always involved clinically / disconnected from practice
  • Often not engaged in the long term
  • Distraction; not essential to business
  • Mild hostility: Some researchers have been openly critical of SNOMED CT

*until recently
Suggestions for moving forward? (researcher’s perspective)

• SNOMED CT Research Webinar series – Thanks, Suzy!
• SNOMED International convening/sponsoring research workshops
• Research license
• Organize research efforts (e.g., for clinical analytics)
  • E.g., testing SNOMED CT at scale in clinical data warehouses
• Provide support for evaluation
  • Access to SNOMED CT experts
  • In-kind contribution to research efforts