Using lexical and structural features for quality assurance of biomedical ontologies

Application to SNOMED CT

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Outline

◆ Motivation
◆ SNOMED CT
◆ Terminology QA approaches
  • Structural
  • Lexical
  • Hybrid (structural + lexical)
Motivation
Research and applications

Getting the foot out of the pelvis: modeling problems affecting use of SNOMED CT hierarchies in practical applications

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ABSTRACT

Objectives (a) To determine the extent and range of errors and issues in the Systematised Nomenclature of Medicine — Clinical Terms (SNOMED CT) hierarchies as they affect two practical projects. (b) To determine the origin of issues raised and propose methods to address them.

Methods The hierarchies for concepts in the Core Problem List Subset published by the Unified Medical Language System were examined for their appropriateness in two applications. Anomalies were traced to their source to determine whether they were simple local errors, systematic inferences propagated by SNOMED’s classification process, or the result of problems with SNOMED’s schemas. Conclusions were confirmed by showing that altering the root cause and reclassifying had the intended effects, and not others.

Main results Major problems were encountered, involving error or constraints, arising at various levels in the hierarchy. Where SNOMED codes are used in practice, such as in electronic medical records, when doctors apply SNOMED codes to a patient, they are stating that those codes and all their ancestors in the hierarchy apply to that patient. When researchers use codes in queries, they are querying for those codes and all of their descendants. When software interprets postcoordinated expressions, it depends on the hierarchies to give those expressions their correct meaning.

This paper reports attempts to use the SNOMED hierarchies in two practical applications:

► as a contributor to the ‘ontological component’ of the eleventh revision of the International Classification of Diseases (ICD-11);
► as part of the documentation tools for a commercial clinical information system.

By contrast with most previous studies, we are concerned here only with inferences that are incorrect or misleading clinically. We are not concerned with the use of SNOMED codes
Motivation

- Biomedical terminologies and ontologies are enabling resources for clinical decision support systems and data integration systems for translational research and health analytics
- Their quality has a direct impact on healthcare and biomedical research
- Quality assurance (QA) of biomedical terminologies remains an active field of research
SNOMED Clinical Terms
SNOMED CT Characteristics

- Developed by SNOMED International
  - Consortium of over 40 member countries

- Largest clinical terminology in the world
  - ~350,000 active concepts
  - ~1 million terms (“descriptions”)

- Major organizing principles
  - Logical definitions (incomplete: many primitives)
  - Built using description logics (\(\mathcal{EL}^{++}\))
SNOMED CT Example

Parents
- Operation on appendix (procedure)
- Partial excision of large intestine (procedure)

Appendectomy (procedure) ★
SCTID: 80146002
80146002 | Appendectomy (procedure) |
- Appendectomy
- Excision of appendix
- Appendectomy
- Appendectomy (procedure)

Procedure site - Direct → Appendix structure
Method → Excision - action

Children (8)
- Appendectomy with drainage (procedure)
- Emergency appendectomy (procedure)
- Excision of appendiceal stump (procedure)
- Excision of ruptured appendix by open approach (procedure)
- Incidental appendectomy (procedure)
- Interval appendectomy (procedure)
- Laparoscopic appendectomy (procedure)
- Non-emergency appendectomy (procedure)
SNOMED CT Challenges

◆ Legacy
  ● Many primitive concepts
  ● Not amenable to automatic DL classification

◆ Maintenance
  ● Human editors
  ● Error prone

◆ Quality assurance
  ● Difficult due to its size
  ● Ontology templates
    ▪ Difficult to apply retrospectively
Quality assurance approaches
Quality assurance approaches

◆ Three types of QA approaches applied to SNOMED CT by researchers

  ● Lexical
    ■ Based on the properties of terms, such as compositionality
  
  ● Structural
    ■ Based on the organizational structure of concepts
  
  ● Semantic
    ■ Rely on the logical definitions of concepts in description logic-based terminologies

◆ Hybrid approaches (structural + lexical)
Quality assurance approaches

Structural approaches
Lattices

- **Lattice**
  - Specific type of directed acyclic graph (DAG)
  - Any two nodes have a unique maximal common descendant, as well as a unique minimal common ancestor

- Bacterial meningitis
- Viral meningitis
- Infective meningitis
- CNS disease
- Infections disease
Lattice-based structural QA

- Non-lattice (approximation)
  - When two concepts have more than one ancestor in common

- Non-lattice subgraphs are often indicative of a problem in ontology construction
  - Missing hierarchical relation
  - Missing intermediary concept
Example of non-lattice subgraph

Example #1

Jiang et al., JAMIA, 2009

Partial hypophysectomy

Partial excision of pituitary gland by transfrontal approach

Transcranial hypophysectomy

Partial excision of pituitary gland by transphenoidal approach

Transphenoidal hypophysectomy

Upper bounds

Lower bounds
Example #1

Missing intermediary concept

- Partial hypophysectomy
  - Partial excision of pituitary gland by *transfrontal* approach
- Transcranial hypophysectomy
  - Partial excision of pituitary gland by *transsphenoidal* approach
- Transsphenoidal hypophysectomy
Example #2

Non-lattice subgraph in SNOMED CT

Acute respiratory disease

Chronic bronchitis

Acute exacerbation of chronic bronchitis

Chronic obstructive bronchitis

Zhang, AMIA, 2010; Zhang, ISWC, 2010
Example #2

Missing hierarchical relation

- Acute respiratory disease
- Chronic bronchitis
- Acute exacerbation of chronic bronchitis
- Acute exacerbation of chronic obstructive bronchitis
- Chronic obstructive bronchitis
Non-lattice subgraph in SNOMED CT

Example #2

- Acute respiratory disease
- Chronic bronchitis
- Acute exacerbation of chronic bronchitis
- Chronic obstructive bronchitis

而这组图展示了不同类型的呼吸系统疾病之间的非晶格子图关系。
Limitations of the structural approach

◆ Technically
  ● Computationally intensive (initially)

◆ Practically
  ● Limited precision
    ◆ Not all non-lattice subgraphs are indicative of an error
  ● Editorial guidelines in SNOMED CT
    ◆ Avoid systematic pre-coordination
  ● Trade-off between
    ◆ “Purity” of lattice representation
    ◆ Parsimony
Quality assurance approaches

Lexical approaches
QA based on lexical patterns

- Lexical differences among terms are often indicative of semantic relations among them
- Term compositionality

Simple implementation through bags of words

Acute exacerbation of chronic bronchitis

Acute exacerbation of chronic obstructive bronchitis
# Suggested missing hierarchical relations

<table>
<thead>
<tr>
<th>Child name</th>
<th>Parent name</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alveolar bone graft to mandible</td>
<td>Alveolar bone graft</td>
</tr>
<tr>
<td>Basal cell carcinoma of skin of lip</td>
<td>Carcinoma of lip</td>
</tr>
<tr>
<td>Carcinoma in situ of palate</td>
<td>Palate carcinoma</td>
</tr>
<tr>
<td>Chronic bacterial otitis externa</td>
<td>Chronic otitis externa</td>
</tr>
<tr>
<td>Congenital vascular anomaly of eyelid</td>
<td>Vascular anomaly of eyelid</td>
</tr>
<tr>
<td>Electrocoagulation of retina for repair of tear</td>
<td>Repair of retina</td>
</tr>
<tr>
<td>Hallucinogen intoxication delirium</td>
<td>Hallucinogen intoxication</td>
</tr>
<tr>
<td>Infection of preauricular sinus</td>
<td>Preauricular sinus</td>
</tr>
<tr>
<td>Pituitary stalk compression hyperprolactinemia</td>
<td>Pituitary stalk compression</td>
</tr>
<tr>
<td>Suture of tongue to lip for micrognathia</td>
<td>Suture of lip</td>
</tr>
</tbody>
</table>
Limitations of the lexical approach

- Limited precision
  - Many false positives when using a simple bag-of-words approach

- Limited recall
  - Many false negatives when using only the preferred terms

- Possible mitigation strategies
  - Add lexico-syntactic constraints to increase precision
  - Also use synonyms to increase recall
  - Overall: gain in recall does not compensate loss in precision
Quality assurance approaches

Combining structural and lexical approaches
Research and Applications

Mining non-lattice subgraphs for detecting missing hierarchical relations and concepts in SNOMED CT

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ABSTRACT

Objective: Quality assurance of large ontological systems such as SNOMED CT is an indispensable part of the terminology management lifecycle. We introduce a hybrid structural-lexical method for scalable and systematic discovery of missing hierarchical relations and concepts in SNOMED CT.

Material and Methods: All non-lattice subgraphs (the structural part) in SNOMED CT are exhaustively extracted
Objectives

- To combine lexical and structural QA approaches to automatically and precisely identifying missing hierarchical relations and missing concepts in SNOMED CT
- To suggest remediation for such inconsistencies

Materials: September 2015 version of SNOMED CT (U.S. edition)
Overview of the methods

◆ Identifying non-lattice pairs and subgraphs
◆ Analyzing non-lattice subgraphs with lexical patterns
  ● Containment
  ● Intersection
  ● Union
  ● Intersection-Union
◆ Evaluation
Identifying non-lattice pairs and subgraphs

◆ Hadoop-based technique
  ● 30 hours to analyze all pairs of SNOMED CT concepts

◆ Aggregation of non-lattice pairs with the same shared ancestors into non-lattice subgraphs
  ● Smaller subgraphs contained in larger subgraphs

◆ Select small non-lattice subgraphs (Size 4-6)
  ● Cognitively manageable

◆ 171,011 non-lattice subgraphs
  ● 70,250 small non-lattice subgraphs
    ■ 2046 exhibit one of the 4 lexical patterns
Lexical patterns (1) Containment

- The set of words for one concept in the upper (resp. lower) bounds is contained in the set of words for another concept in the upper (resp. lower) bounds
- Suggests a missing hierarchical relation between concepts in the upper (resp. lower) bounds
- 736 small non-lattice subgraphs with this pattern
Lexical patterns (1) Containment

Non-lattice subgraph

- Duodenal ulcer with perforation AND obstruction
- Chronic duodenal ulcer with perforation AND obstruction

Suggested remediation

- Obstruction of duodenum
- Chronic duodenal ulcer with perforation

Stated in terms of a non-lattice subgraph:

- Duodenal ulcer with perforation AND obstruction
- Chronic duodenal ulcer with perforation AND obstruction

Chronic duodenal ulcer with perforation AND obstruction
Lexical patterns (2) Intersection

- The intersection of sets of words for concepts in the lower bounds is equal to the set of words for some concept in the upper bounds.
- Suggests a *missing hierarchical relation* between concepts in the upper bounds.
- 1085 small non-lattice subgraphs with this pattern.
Lexical patterns (2) Intersection

Non-lattice subgraph

Disorder of colon

Irritable bowel syndrome

Irritable bowel syndrome variant of childhood

Irritable bowel syndrome with diarrhea

Suggested remediation

Disorder of colon

Irritable bowel syndrome

Irritable bowel syndrome variant of childhood

Irritable bowel syndrome with diarrhea

Irritable bowel syndrome variant of childhood \( \cap \) Irritable bowel syndrome with diarrhea
Lexical patterns (3) Union

- The union of the sets of words for concepts in the upper bounds is equal to the set of words for some concept in the lower bounds
- Suggests a missing hierarchical relation between concepts in the lower bounds
- 164 small non-lattice subgraphs with this pattern
Lexical patterns \((3)\) Union

Non-lattice subgraph

\[
\begin{align*}
\text{Epithelial neoplasm of skin} & \cup \text{Malignant neoplasm of skin} \\
\text{Malignant epithelial neoplasm of skin} & \cup \text{Squamous cell carcinoma of skin}
\end{align*}
\]

Suggested remediation

\[
\begin{align*}
\text{Epithelial neoplasm of skin} & \cup \text{Malignant neoplasm of skin} \\
\text{Malignant epithelial neoplasm of skin} & \cup \text{Squamous cell carcinoma of skin}
\end{align*}
\]

\[
\text{Malignant epithelial neoplasm of skin}
\]
Lexical patterns (4) Union-Intersection

- The union of the sets of words for concepts in the upper bounds is equal to the intersection of sets of words for concepts in the lower bounds.
- Suggests a *missing intermediary concept* between the upper bounds and the lower bounds.
- 61 small non-lattice subgraphs with this pattern.
**Lexical patterns (4) Union-Intersection**

**Non-lattice subgraph**
- Neoplasm of right upper lobe of lung
- Malignant neoplasm of upper lobe of lung
- Secondary malignant neoplasm of right upper lobe of lung
- Primary malignant neoplasm of right upper lobe of lung

**Suggested remediation**
- Neoplasm of right upper lobe of lung
- Malignant neoplasm of upper lobe of lung
- Secondary malignant neoplasm of right upper lobe of lung
- Primary malignant neoplasm of right upper lobe of lung

**Equation:**
\[
\text{Secondary malignant neoplasm of right upper lobe of lung} \cap \text{Primary malignant neoplasm of right upper lobe of lung} = \text{Malignant neoplasm of right upper lobe of lung}
\]
Evaluation

- 59 subgraphs independently reviewed by 2 experts after triaging
  - Differences resolved by discussion
- All contained errors – 61 errors
  - Missing hierarchical relation: 59
  - Missing intermediary concept: 2
- Lexical patterns
  - Containment: 34; Intersection: 14; Union: 8; U/I: 3
- Suggested remediation
  - Accepted for 53 subgraphs
  - Rejected for 6 subgraphs (deeper modeling issues)
Significance

- Most terminology QA techniques merely identify potential errors
- Our approach
  - Identified unreported errors
    - Confirmed by experts
  - Suggested appropriate remediation in many cases
- Should greatly facilitate error correction by the developers of SNOMED CT
- Scalable and applicable to other terminologies
Limitations and future work

◆ Suggested remediation (e.g., to add missing hierarchical relations) is based on the inferred concept hierarchy of SNOMED CT
  ● Does not address the root cause (e.g., incomplete/inaccurate logical definition)
  ● Root cause needs to be addressed by the SNOMED CT editors

◆ Only 4 lexical patterns considered
  ● Could be refined with additional patterns
Follow-up investigation

- **Additional lexical patterns**
  - Enrich bags of words with
    - Words from ancestors in the non-lattice subgraph
    - Pairs of hypernyms harvested from the subgraph

Cui, JBI, 2018