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Using lexical and structural features for quality assurance of biomedical ontologies

Application to SNOMED CT



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References

- ◆ Cui L, Zhu W, Tao S, Case JT, Bodenreider O, Zhang GQ. Mining non-lattice subgraphs for detecting missing hierarchical relations and concepts in SNOMED CT. *J Am Med Inform Assoc*. 2017;24(4):788-798. PMID: 28339775

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Research and Applications

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Research and Applications

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

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SNOMED Clinical Terms

SNOMED
International

Leading healthcare
terminology, worldwide

SNOMED CT Characteristics

- ◆ Developed by SNOMED International
 - Consortium of 30 member countries
- ◆ Largest clinical terminology in the world
 - ~320,000 active concepts
 - ~ 1 million terms (“descriptions”)
- ◆ Major organizing principles
 - Logical definitions (incomplete: many primitives)
 - Built using description logics (EL++)



SNOMED CT Top level

- ▼ ● SNOMED CT Concept
 - ▶ ● Body structure (body structure)
 - ▶ ● Clinical finding (finding)
 - ▶ ● Environment or geographical location (environment / location)
 - ▶ ● Event (event)
 - ▶ ● Observable entity (observable entity)
 - ▶ ● Organism (organism)
 - ▶ ● Pharmaceutical / biologic product (product)
 - ▶ ● Physical force (physical force)
 - ▶ ● Physical object (physical object)
 - ▶ ● Procedure (procedure)
 - ▶ ● Qualifier value (qualifier value)
 - ▶ ● Record artifact (record artifact)
 - ▶ ● Situation with explicit context (situation)
 - ▶ ● SNOMED CT Model Component (metadata)
 - ▶ ● Social context (social concept)
 - ▶ ● Special concept (special concept)
 - ▶ ● Specimen (specimen)
 - ▶ ● Staging and scales (staging scale)
 - ▶ ● Substance (substance)

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
Appendicectomy
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

- Developed by many human editors
- Error prone

◆ Quality assurance

- Difficult due to its size
- Ontology design patterns (“concept model”)
 - Difficult to apply retrospectively



Quality assurance approaches

Quality assurance approaches

- ◆ Three types of QA approaches applied to SNOMED CT by researchers
 - Lexical
 - Structural
 - Semantic

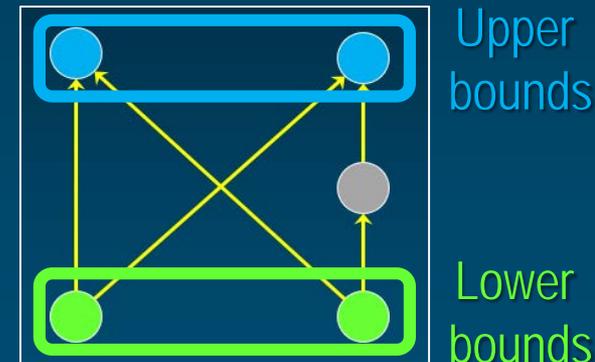
Lattice-based structural QA

◆ Lattice

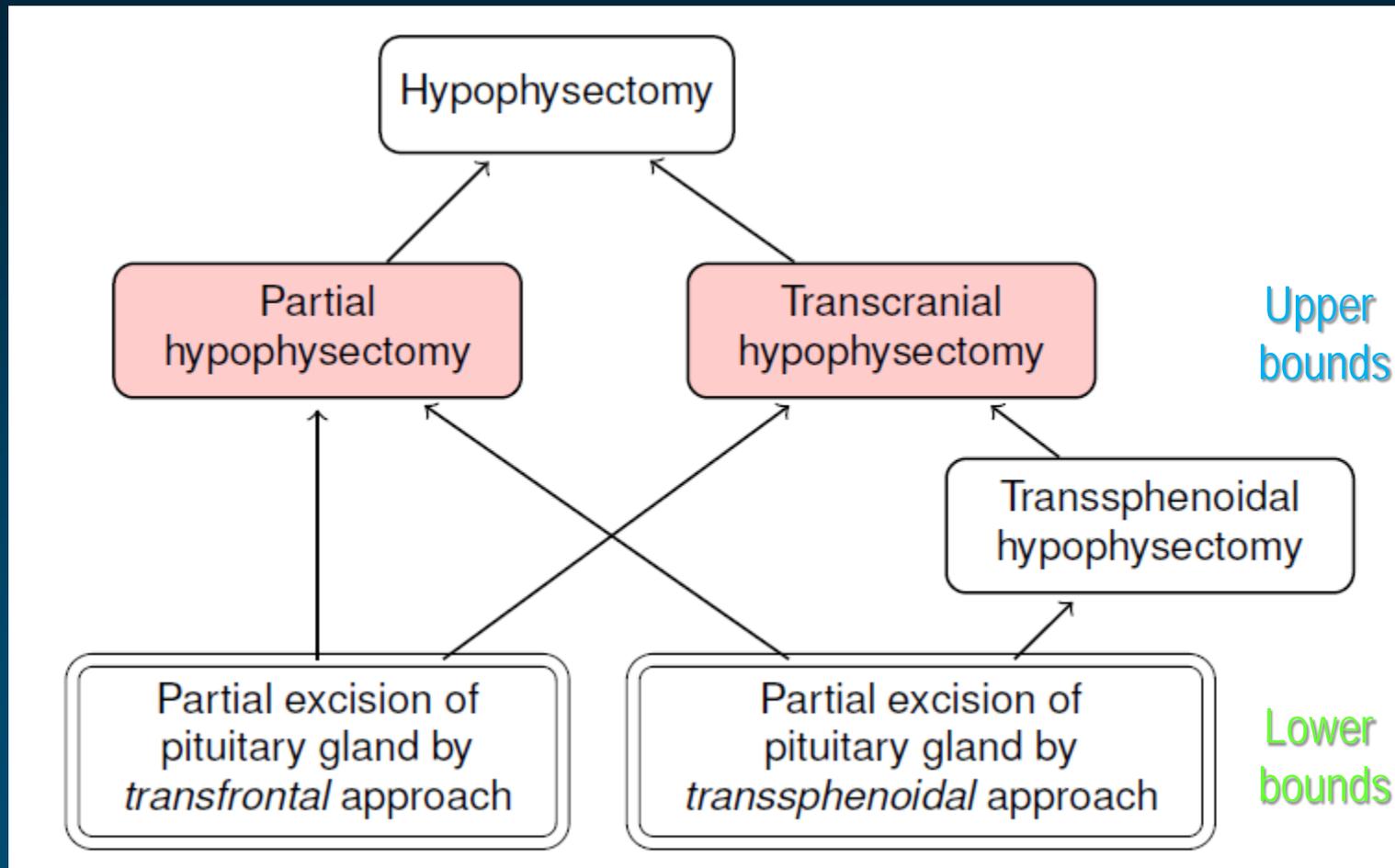
- Specific type of directed acyclic graph (DAG)
- Any two nodes have a unique maximal common descendant, as well as a unique minimal common ancestor

◆ Non-lattice fragments are often indicative of a problem in ontology construction

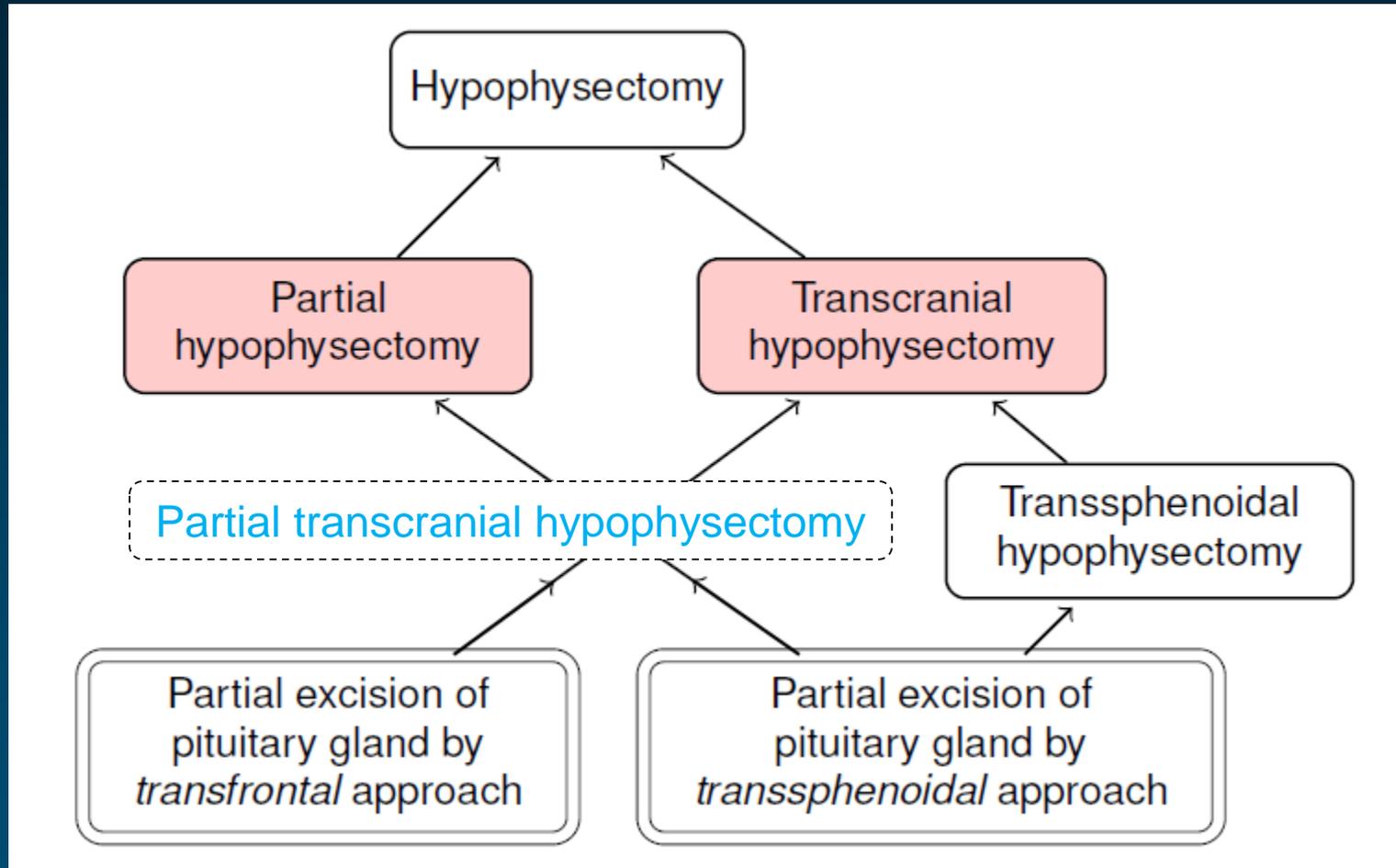
- Missing hierarchical relation
- Missing intermediary concept



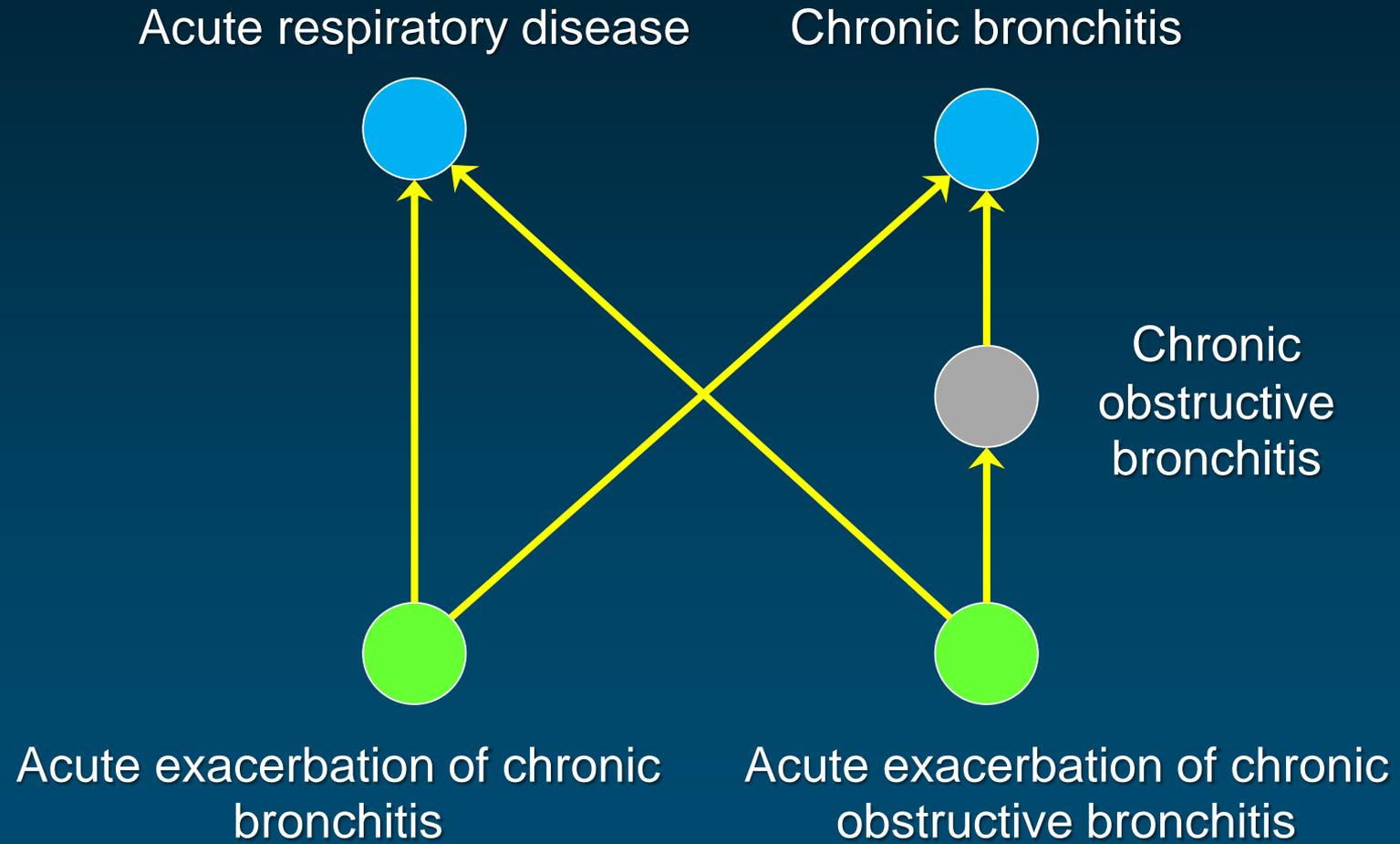
Example of non-lattice fragment



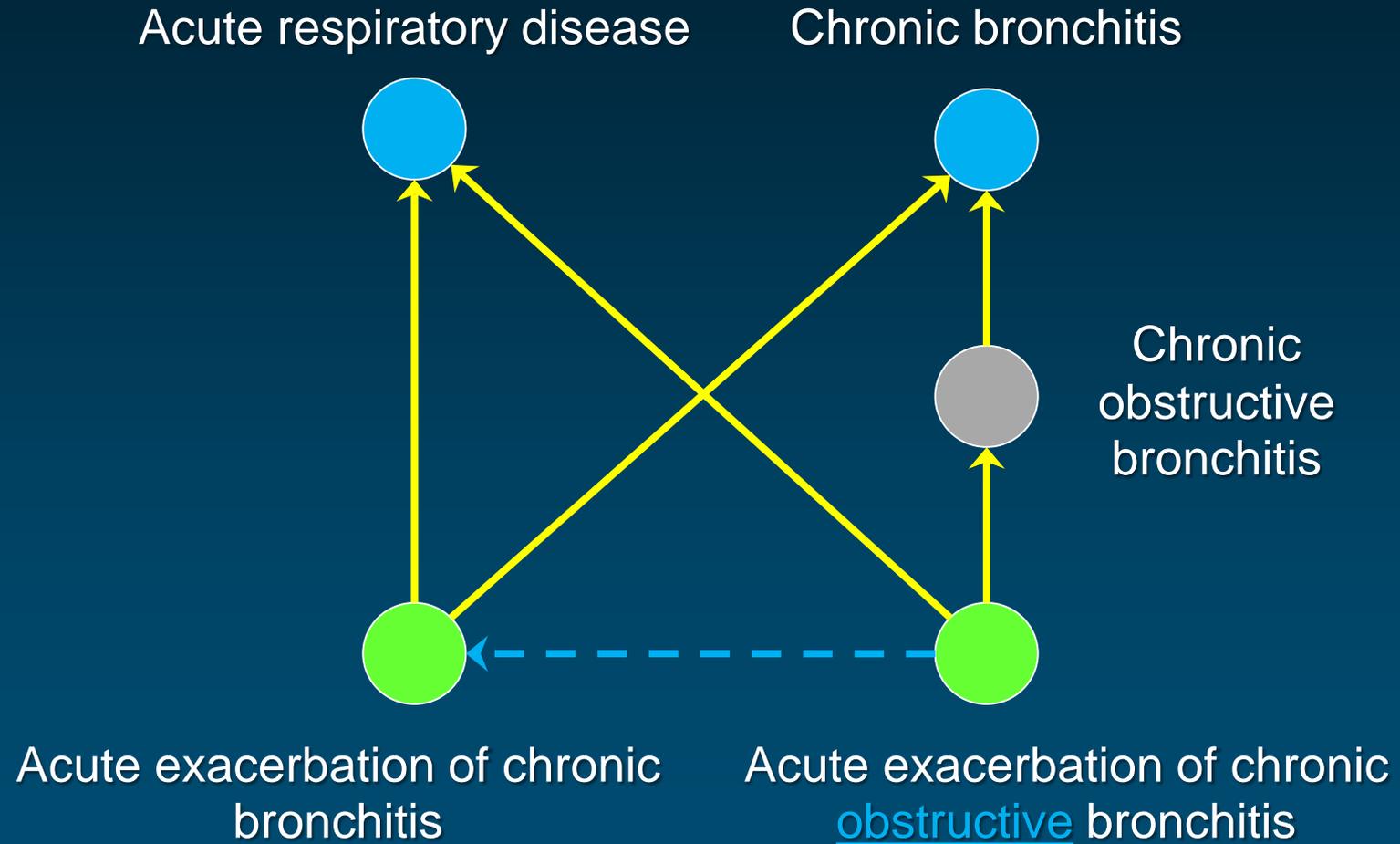
Missing intermediary concept



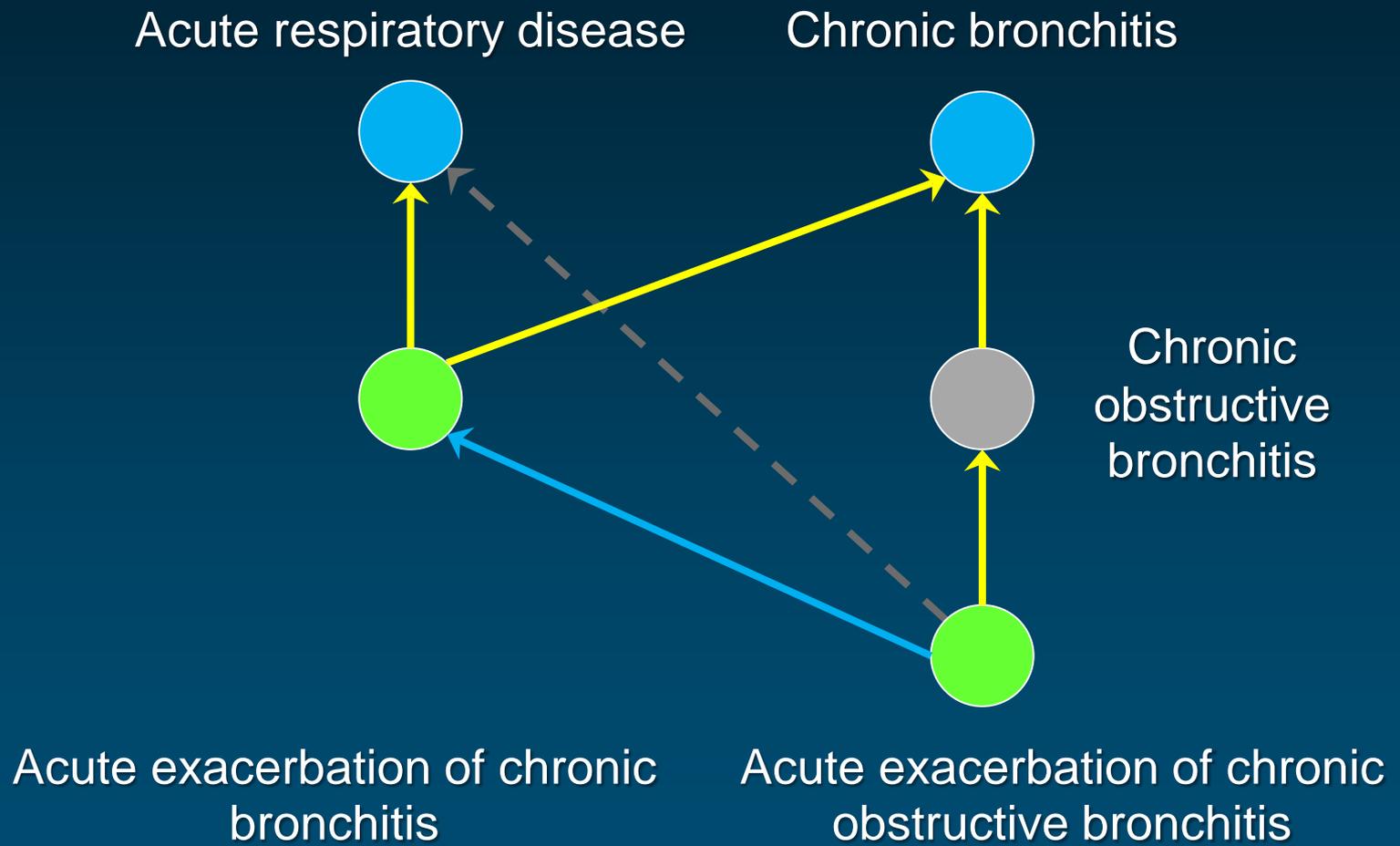
Non-lattice fragment in SNOMED CT



Missing hierarchical relation

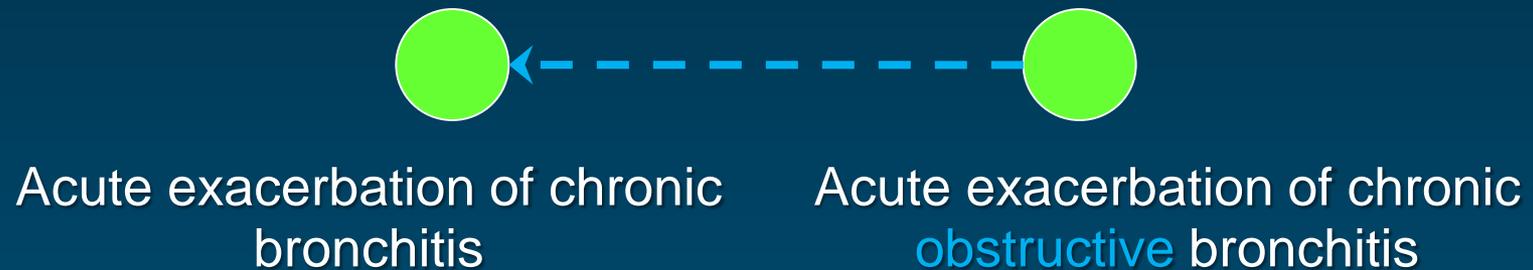


~~Non-lattice~~ fragment in SNOMED CT



QA based on lexical patterns

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



Suggested missing hierarchical relations

Child name	Parent name
Alveolar bone graft <u>to mandible</u>	Alveolar bone graft
<u>Basal cell carcinoma</u> <u>of skin</u> of lip	Carcinoma of lip
Carcinoma <u>in situ of</u> palate	Palate carcinoma
Chronic <u>bacterial</u> otitis externa	Chronic otitis externa
<u>Congenital</u> vascular anomaly of eyelid	Vascular anomaly of eyelid
<u>Electrocoagulation</u> of retina <u>for</u> repair <u>of</u> tear	Repair of retina

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)



Methods & Results

Overview

- ◆ Identifying non-lattice pairs and subgraphs
- ◆ Identifying lexical patterns indicative of missing concepts and relations
- ◆ Analyzing non-lattice subgraphs with lexical patterns
- ◆ 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
- ◆ 631,006 non-lattice pairs
- ◆ 171,011 non-lattice subgraphs
 - Focus on small subgraphs



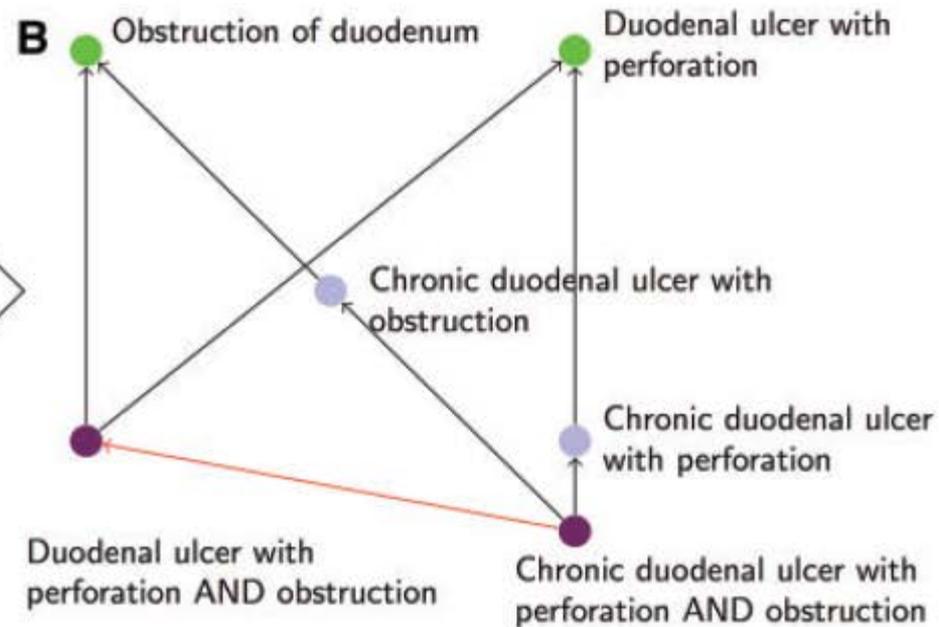
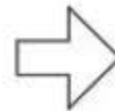
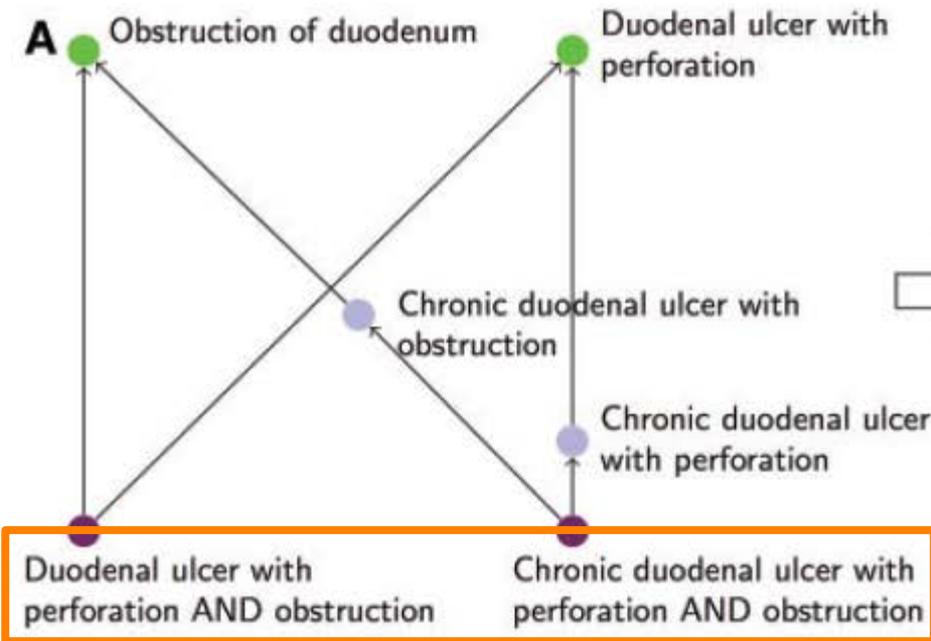
Lexical patterns (1) Containment

- ◆ 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

Suggested remediation



Duodenal ulcer with perforation AND obstruction \supset Chronic duodenal ulcer with perforation AND obstruction

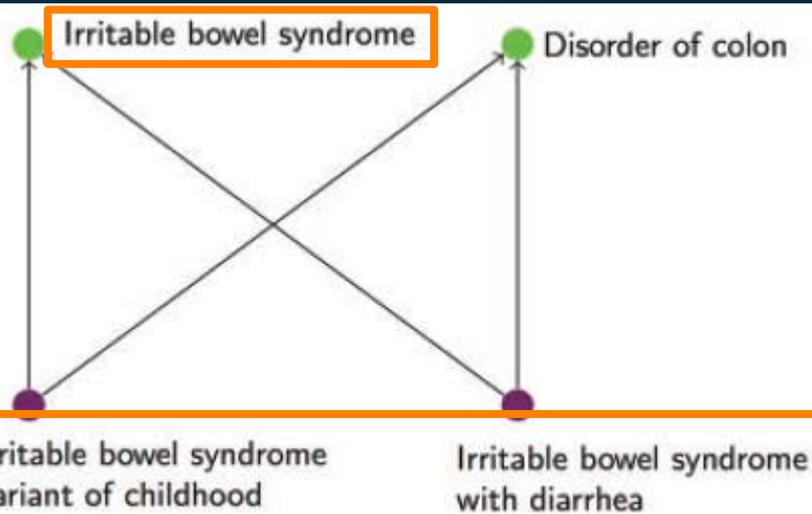
Lexical patterns (2) Intersection

- ◆ 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

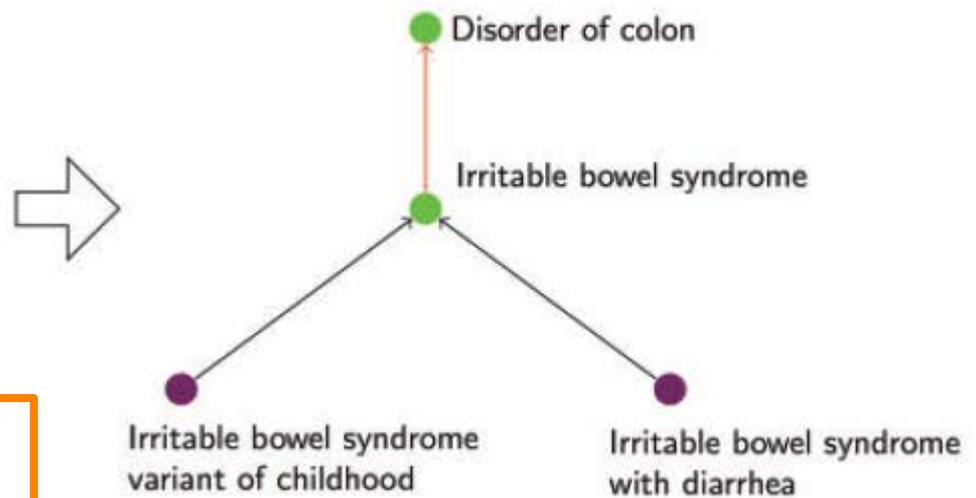


Lexical patterns (2) Intersection

Non-lattice subgraph



Suggested remediation



Irritable bowel syndrome

Irritable bowel syndrome
variant of childhood



Irritable bowel syndrome
with diarrhea

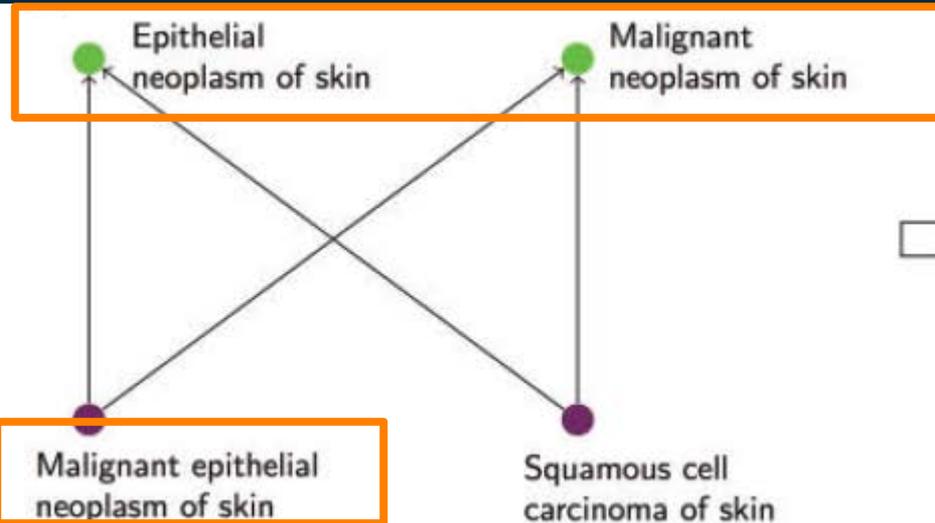
Lexical patterns (3) Union

- ◆ 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

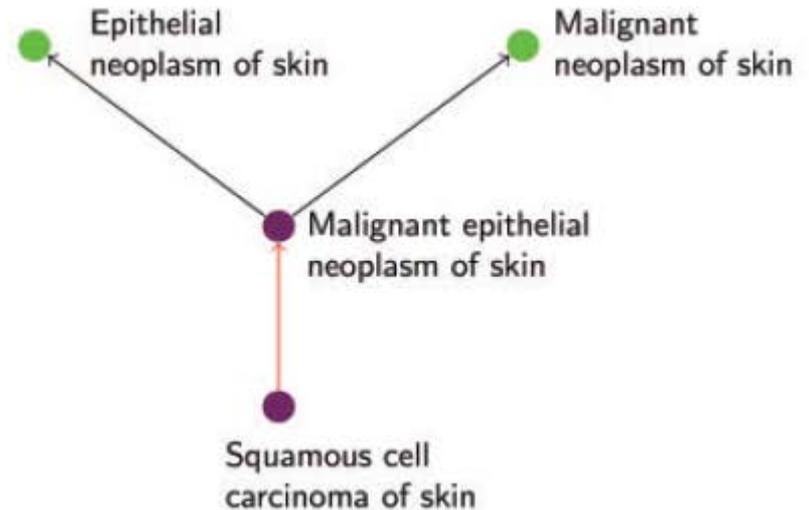


Lexical patterns (3) Union

Non-lattice subgraph



Suggested remediation



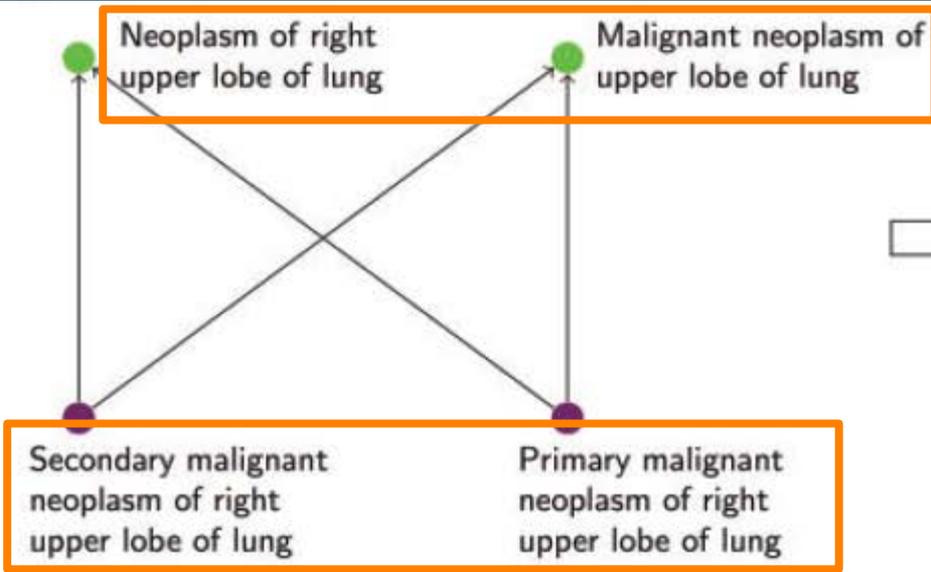
Epithelial neoplasm of skin \cup *Malignant* neoplasm of skin
Malignant epithelial
neoplasm of skin

Lexical patterns (4) Union-Intersection

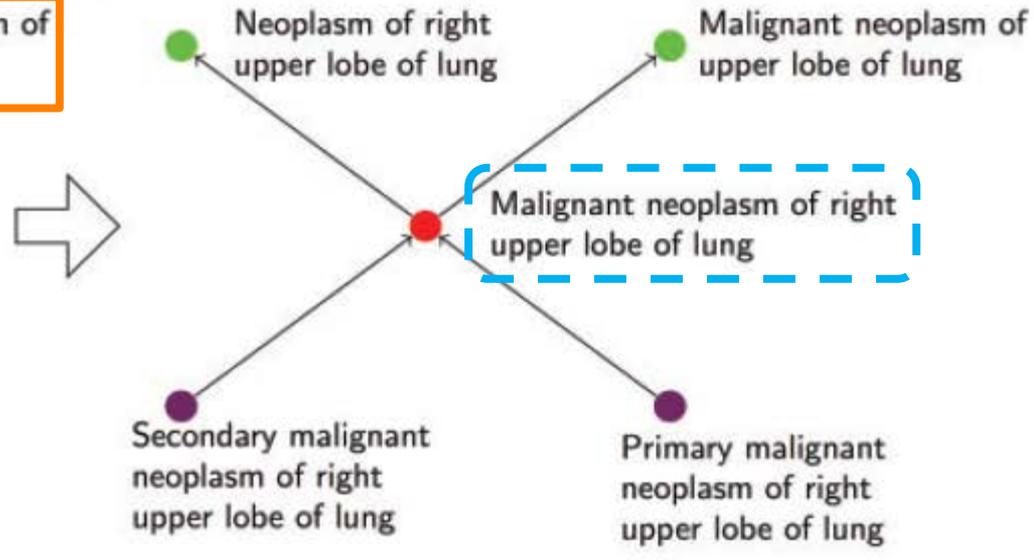
- ◆ The union of the sets of words for concepts in the upper bounds is equal to the intersection of sets of words for concepts in the lower bounds
- ◆ Suggests a *missing intermediary concept* between the upper bounds and the lower bounds
- ◆ 61 small non-lattice subgraphs with this pattern

Lexical patterns (4) Union-Intersection

Non-lattice subgraph



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

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)

Discussion

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

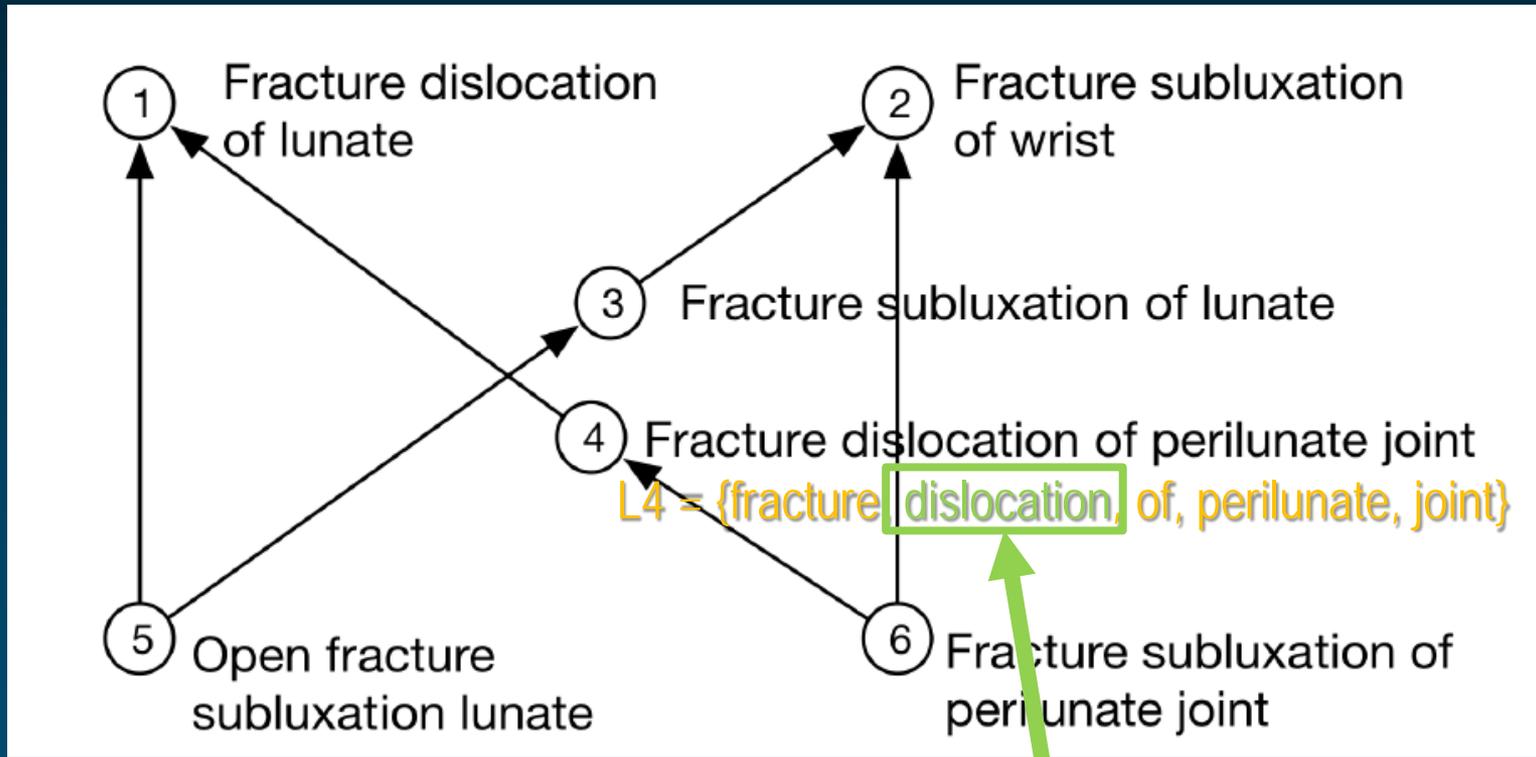
- ◆ Suggested remediation (e.g., to add missing hierarchical relations) is based on the inferred concept hierarchy of SNOMED CT
 - Does not address the root cause (e.g., incomplete/inaccurate logical definition)
 - Root cause needs to be addressed by the SNOMED CT editors
- ◆ Only 4 lexical patterns considered
 - Could be refined with additional patterns
- ◆ Only used the preferred terms
 - Could also use synonyms



Current work

- ◆ Maximize the chances for word set inclusion among concepts within the non-lattice subgraph (beyond the words of the terms themselves)
- ◆ Enrich the lexical definitions with
 - Words from ancestor concepts (within the non-lattice subgraph)
 - Additional hypernyms extracted from hierarchically related concepts (within the non-lattice subgraph)

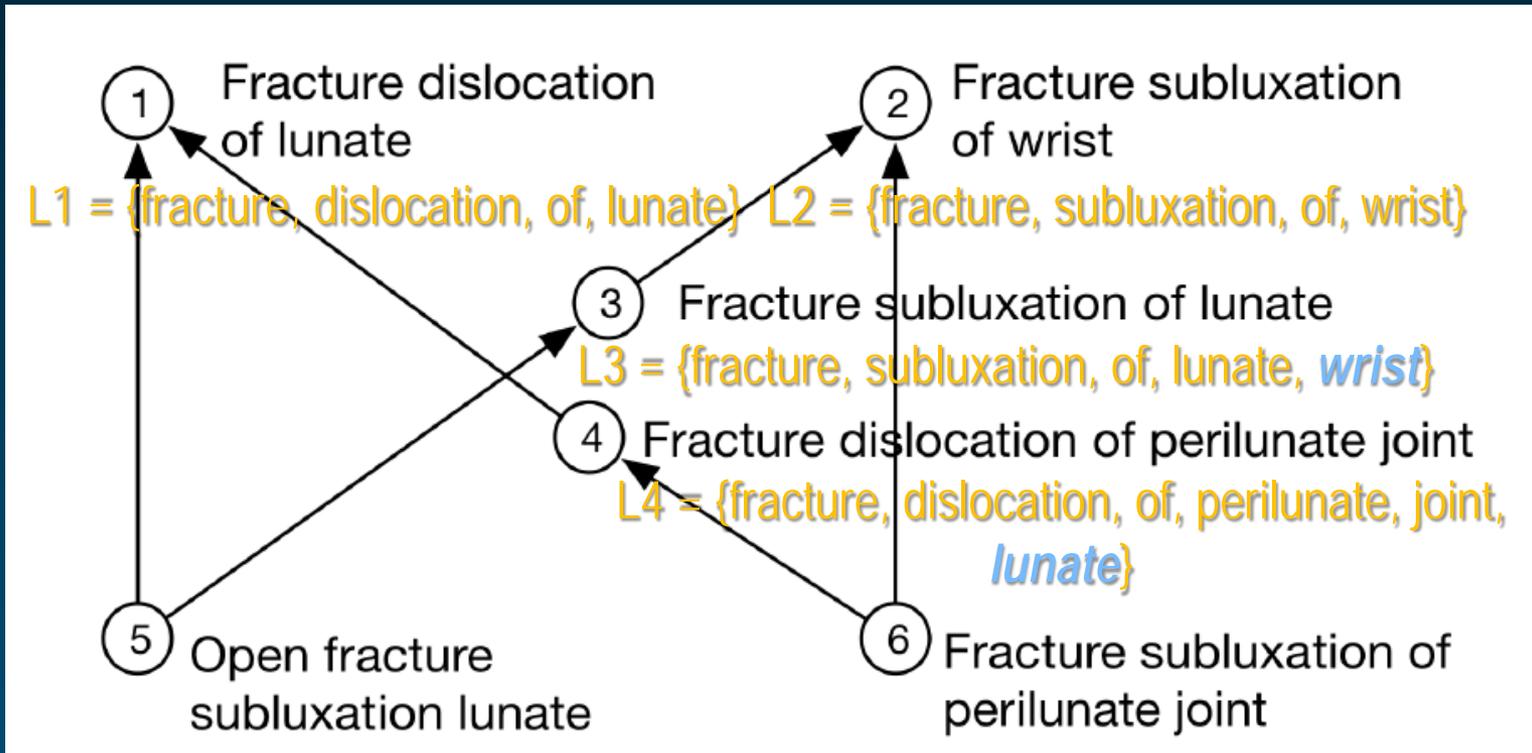
Additional hypernyms



L4 = {fracture **dislocation**, of, perilunate, joint}

L6 = {fracture **subluxation**, of, perilunate, joint}

Lexical features enriched with words from ancestors



L5 = (open, fracture, subluxation, lunate, *dislocation, of, wrist*)

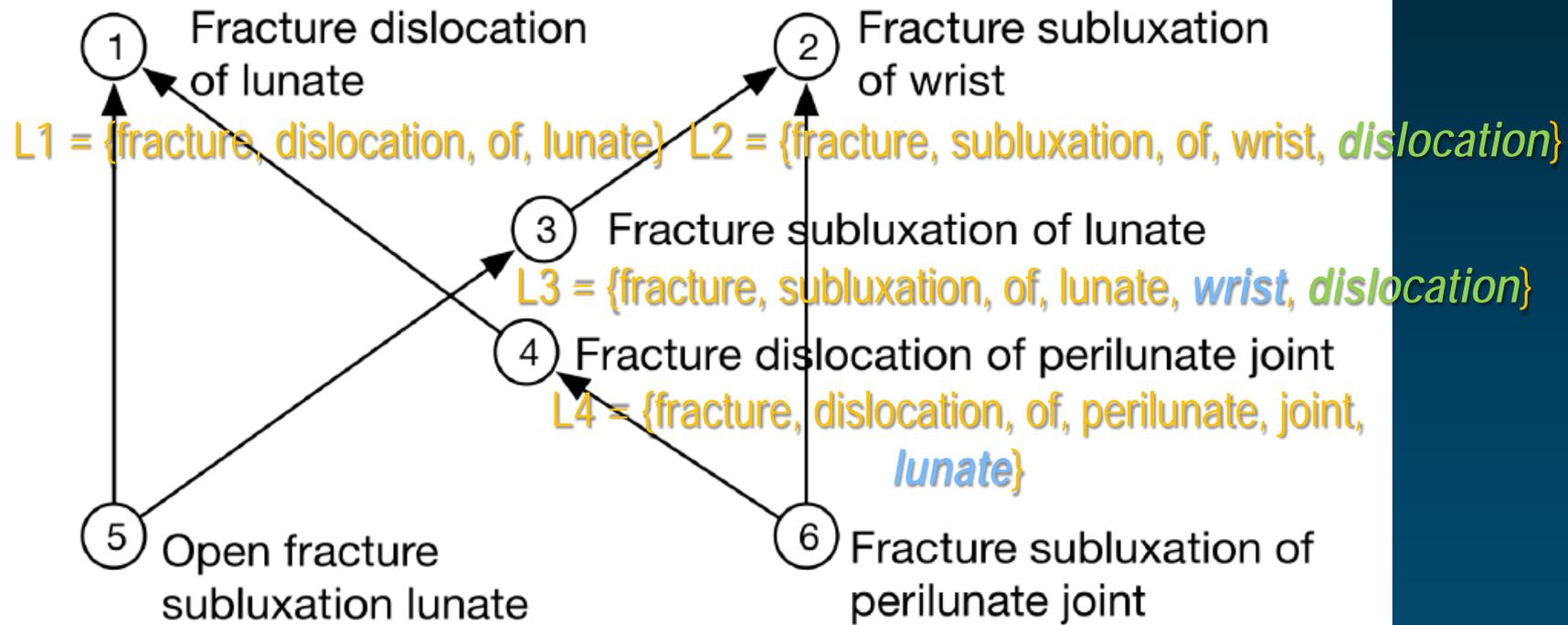
L6 = {fracture, subluxation, of, perilunate, joint, *dislocation, lunate, wrist*}



Lexical features enriched with ancestors and hypernyms

dislocation

subluxation

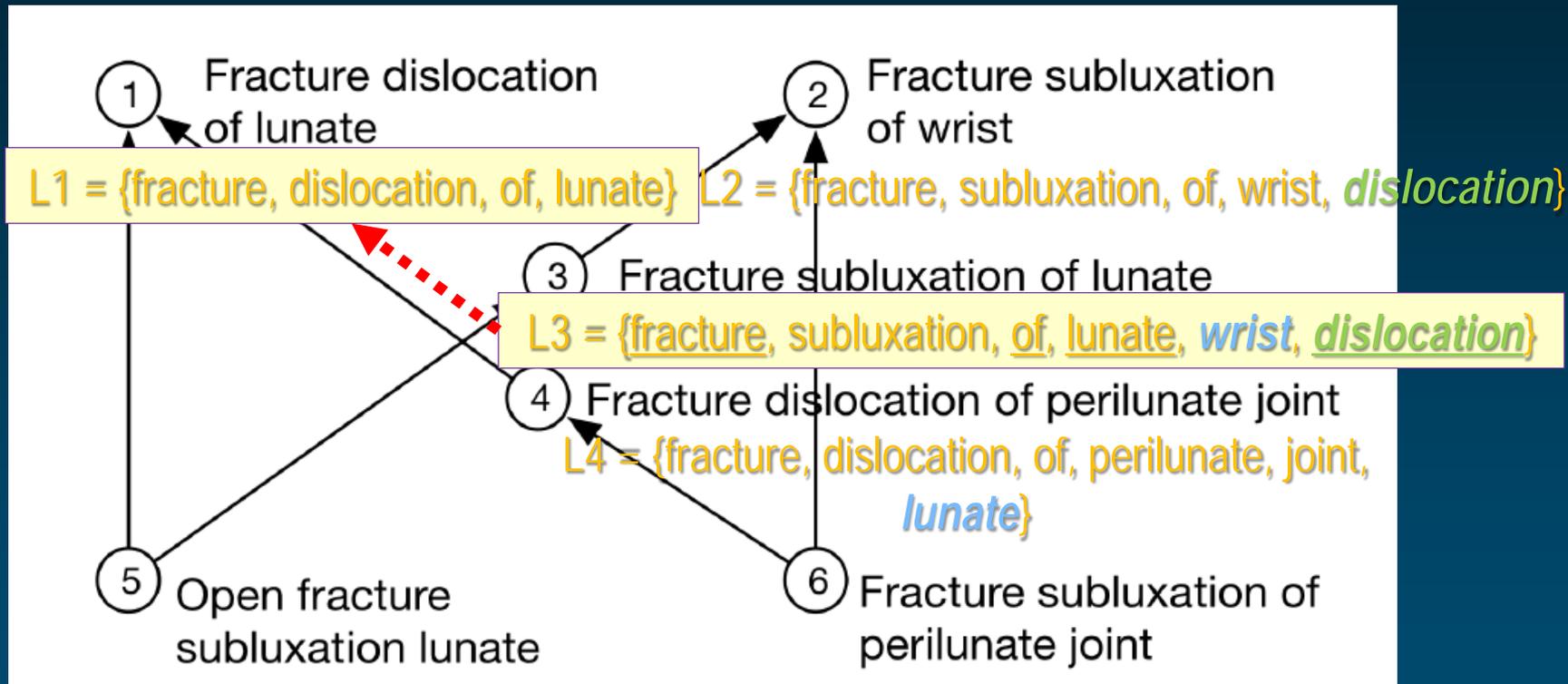


$L5 = (\text{open, fracture, subluxation, lunate, dislocation, of, wrist})$

$L6 = \{\text{fracture, subluxation, of, perilunate, joint, dislocation, lunate, wrist}\}$



Missing hierarchical relation

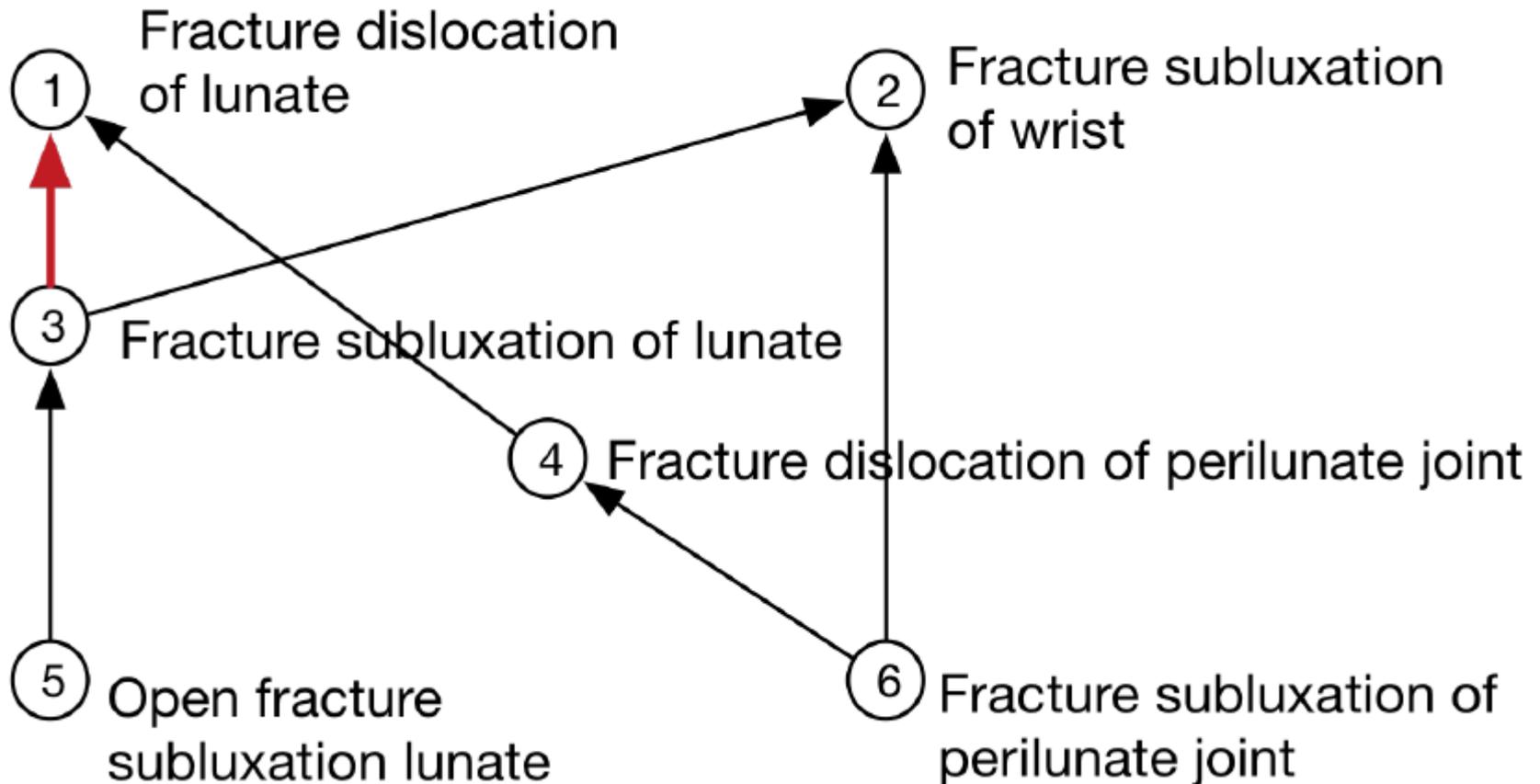


L5 = (open, fracture, subluxation, lunate, *dislocation, of, wrist*)

L6 = {fracture, subluxation, of, perilunate, joint, *dislocation, lunate, wrist*}

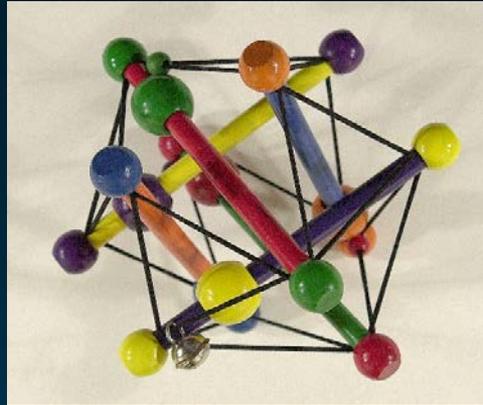


Non-lattice → lattice subgraph



Summary

- ◆ The combination of structural (non-lattice) and lexical (words from terms) features is effective in identifying missing hierarchical relations in SNOMED CT with high precision
- ◆ This approach would be applicable to a large number of ontologies



Medical Ontology Research

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