Categorizing the Relationships between Structurally Congruent Concepts from Pairs of Terminologies for Semantic Harmonization

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Disclosure

- Zhe He discloses that he has no relationships with commercial interests.
- James Geller discloses that he has no relationships with commercial interests.
- Gai Elhanan discloses that he has no relationships with commercial interests.

Learning Objective

- Use structural method to find potential concepts for enriching the conceptual content of a biomedical terminology

Overview

- Motivation
  - Exploring structural method for semantic harmonization
- Background
  - Importance of the conceptual content of SNOMED CT
- Methods
  - Structural matching of pairs of terminologies in the UMLS
- Results
  - Reusable knowledge can be derived by structural matching, including discovery of possible synonym
- Limitations and Future Work
- Conclusions

Motivation

- Need of well-developed and well-maintained terminologies
- NLP tools that process clinical text need a terminology with fruitful concepts and synonyms.
- Complex clinical research texts require combined use of multiple terminologies (Weng *et al.* 2010)
- Terminologies need harmonization to achieve semantic interoperability (Bittner *et al.* 2005)

Semantic Harmonization Between Different Terminologies

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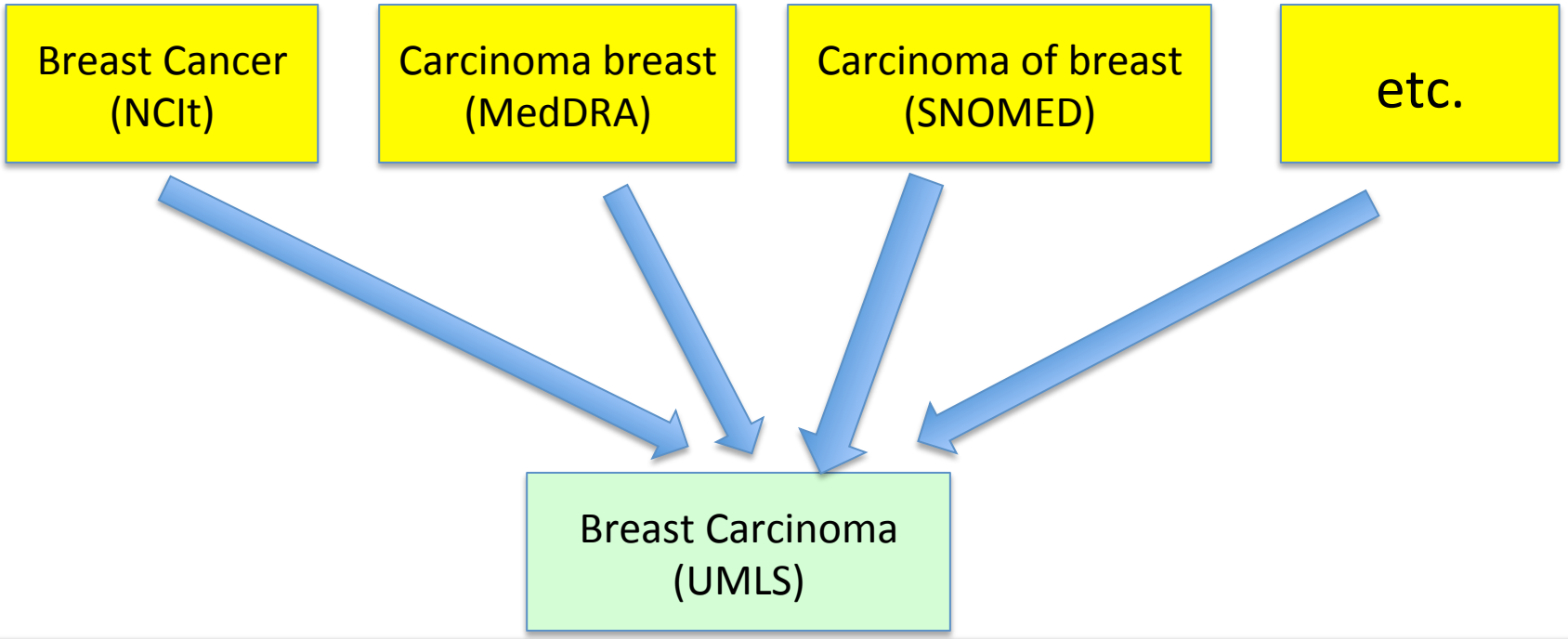
  (Weng *et al.* 2010)
- Harmonized existing time ontologies for annotating temporal relation in clinical narratives (Tao *et al.* 2011)
- Semantic harmonization efforts have recently been extended for various terminologies
  - SNOMED CT and LOINC (AMIA 2013 Informatics Year in Review)
  - SNOMED CT and ICD 11 (Rodrigues *et al.* 2013)

Importance of Conceptual Content of SNOMED CT

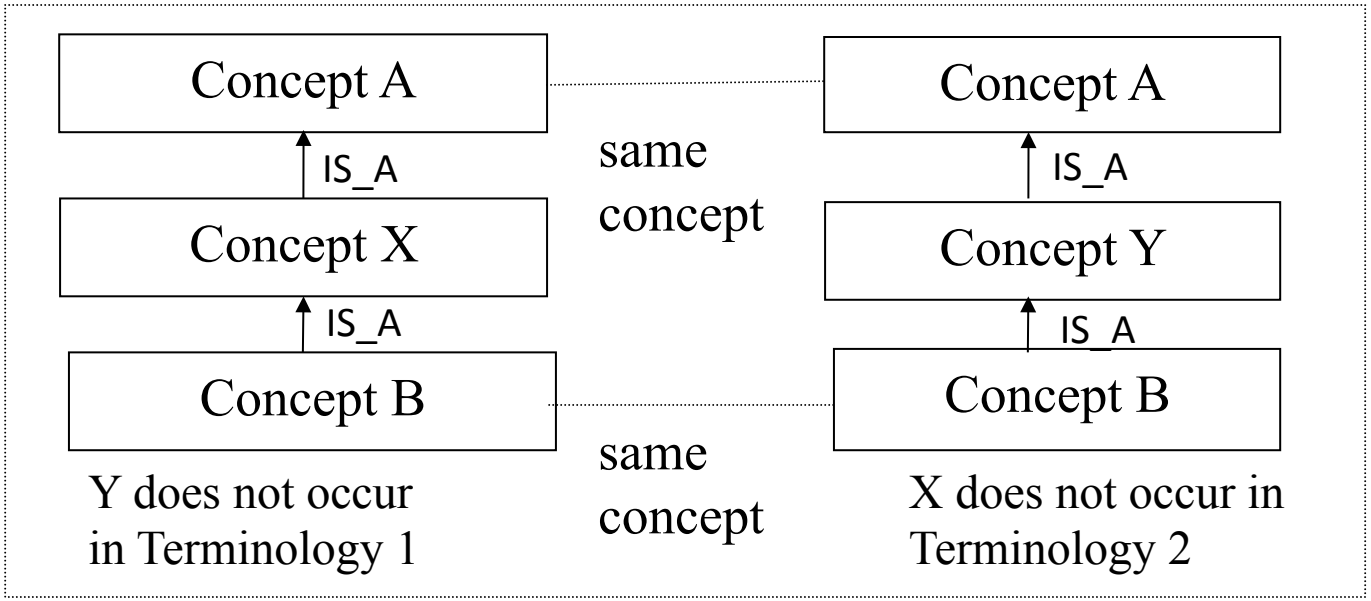
- SNOMED CT is going to be a major terminology for EHR encoding of diagnoses and problem lists by 2015
- SNOMED CT has many problems!
- Top two mentioned deficiencies of SNOMED CT (Elhanan 2011):
  - Missing concepts – 23%
  - Missing synonyms – 17%
- Users will expect SNOMED CT to have correct synonyms and sufficient concepts to be used in EHR

Leveraging Common Structure of Pairs of Terminologies in the UMLS

- UMLS (Unified Medical Language System):
  - More than 170 source terminologies, 8.9 million terms, 2012AB release



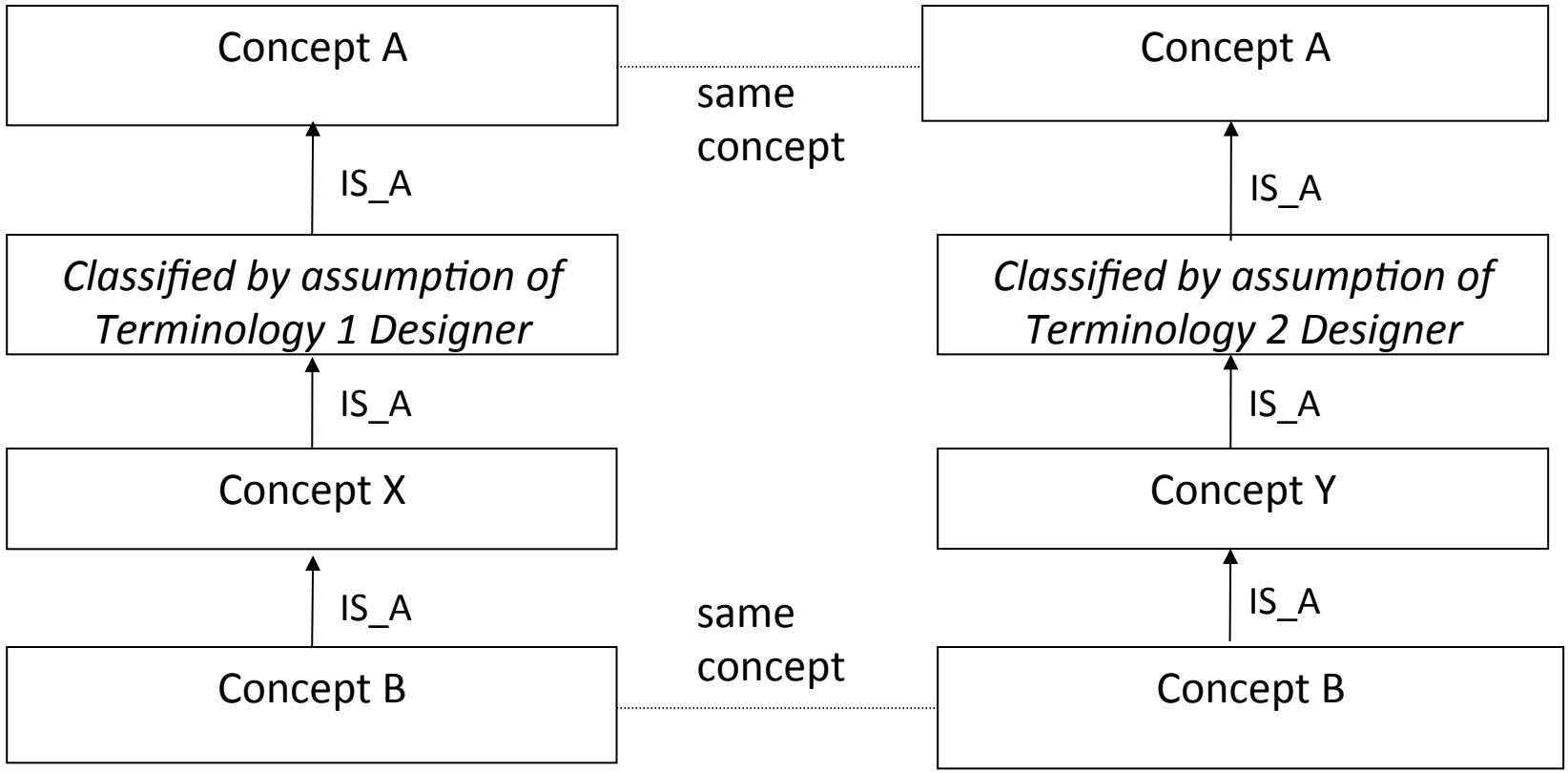
Structurally Congruent Concepts



Cycles were eliminated during the processing

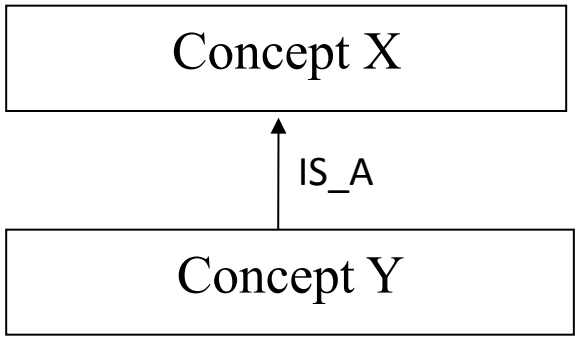
1) X and Y are alternative classifications
2) X can be a parent of Y
3) Y can be a parent of X
4) X and Y are synonymous
5) Structural errors in Terminology 1
6) Structural errors in Terminology 2

Alternative Classification

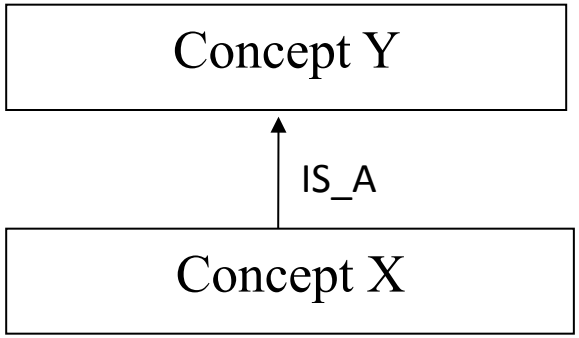


Parent-Child Relationship

X can be a parent of Y



Y can be a parent of X



Synonymous

X and Y are synonymous but have not been identified by the UMLS

Concept X (Synonym: Y)

META Terminologies in this Study

- Metathesaurus terminologies with "PAR" relationship and "inverse_isa" relationship attribute were chosen
- SCTUSX and UWDA were excluded
- Reference Terminologies (Terminology 1):
  - MEDCIN (MEDCIN)
  - National Cancer Institute Thesaurus (NCIt)
  - Gene Ontology (GO)
  - Medical Entities Dictionary (CPM)
  - UMDNS: product category thesaurus (UMD)
  - Foundational Model of Anatomy Ontology (FMA)
- Terminology 2: SNOMED CT

Evaluation: Pairs of Congruent Concepts Found

Reference Terminology	Size of Terminology	# of Pairs of Congruent Concepts	Sample Size
MEDCIN	279529	655	70
NCI	95523	582	70
FMA	82062	116	70
UMD	15956	18	18
GO	61925	6	6
CPM	3078	7	7
Total	--	1384	**241**

Review Results for Pairs of Congruent Concepts

Co-author GE reviewed the sample

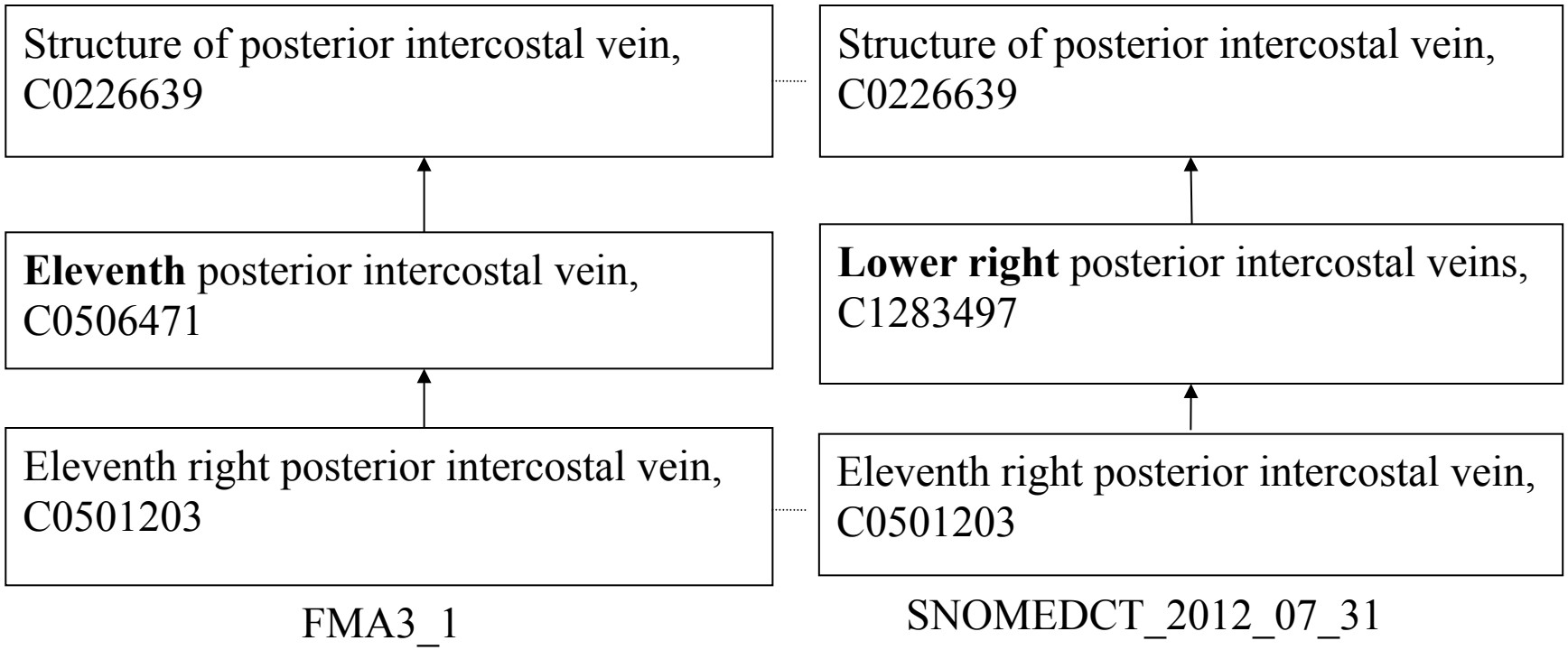
Reference Terminology	Sample Size	Alternative Classification	Y → X	X → Y
MEDCIN	70	44	10	7
NCI	70	38	12	6
GO	6	2	--	4
CPM	7	5	--	--
UMD	18	9	1	--
FMA	70	45	13	4
Total	241	143	36	21
Percentage	100%	**59.3%**	**14.9%**	**8.7%**

23.6%

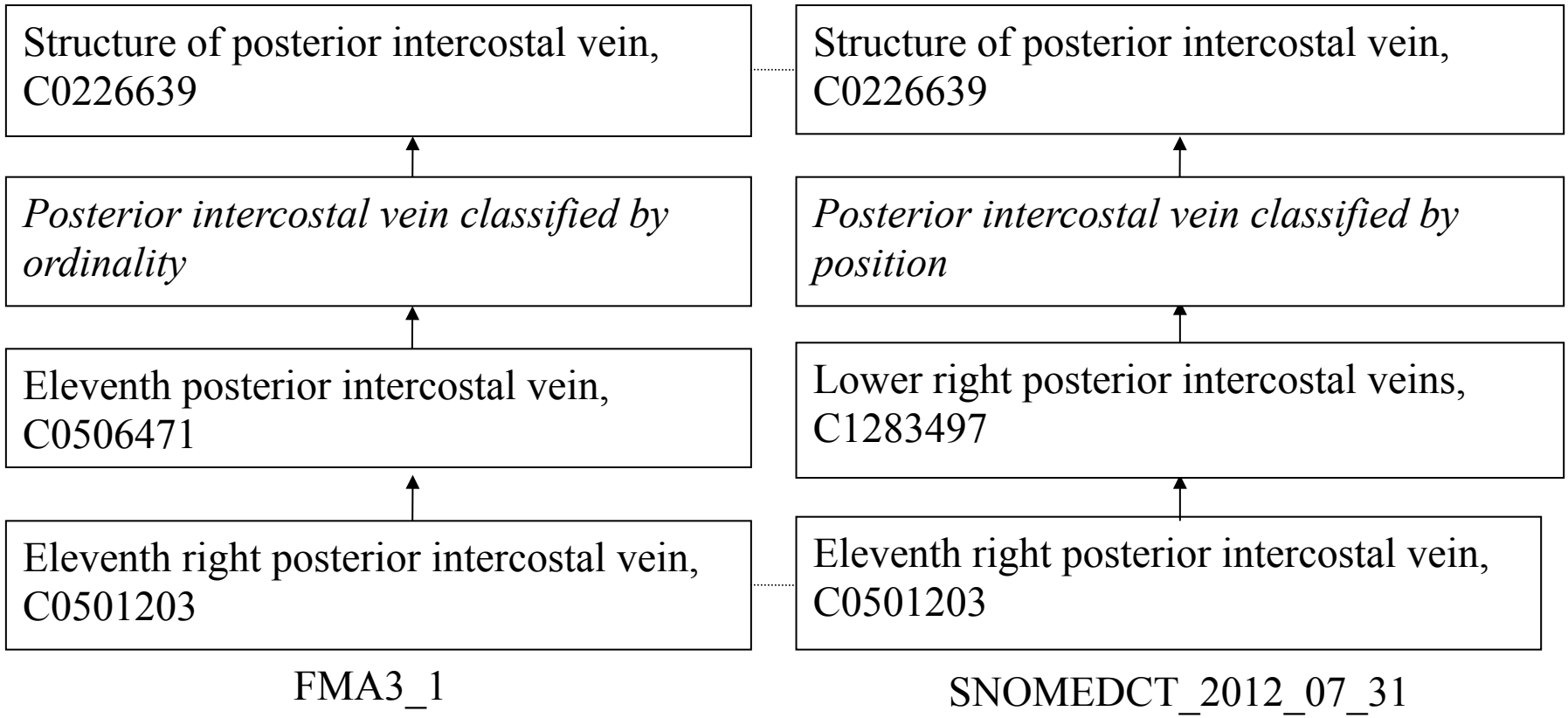
Review Results for Pairs of Congruent Concepts

Reference Terminology	Sample Size	Error in Termi-nology 1	Error in Termi-nology 2	Synonym
MEDCIN	70	--	1	8
NCI	70	--	3	11
GO	6	--	--	--
CPM	7	--	--	2
UMD	18	--	--	8
FMA	70	2	--	6
Total	181	2	4	35
Percentage	100%	0.8%	1.7%	**14.5%**

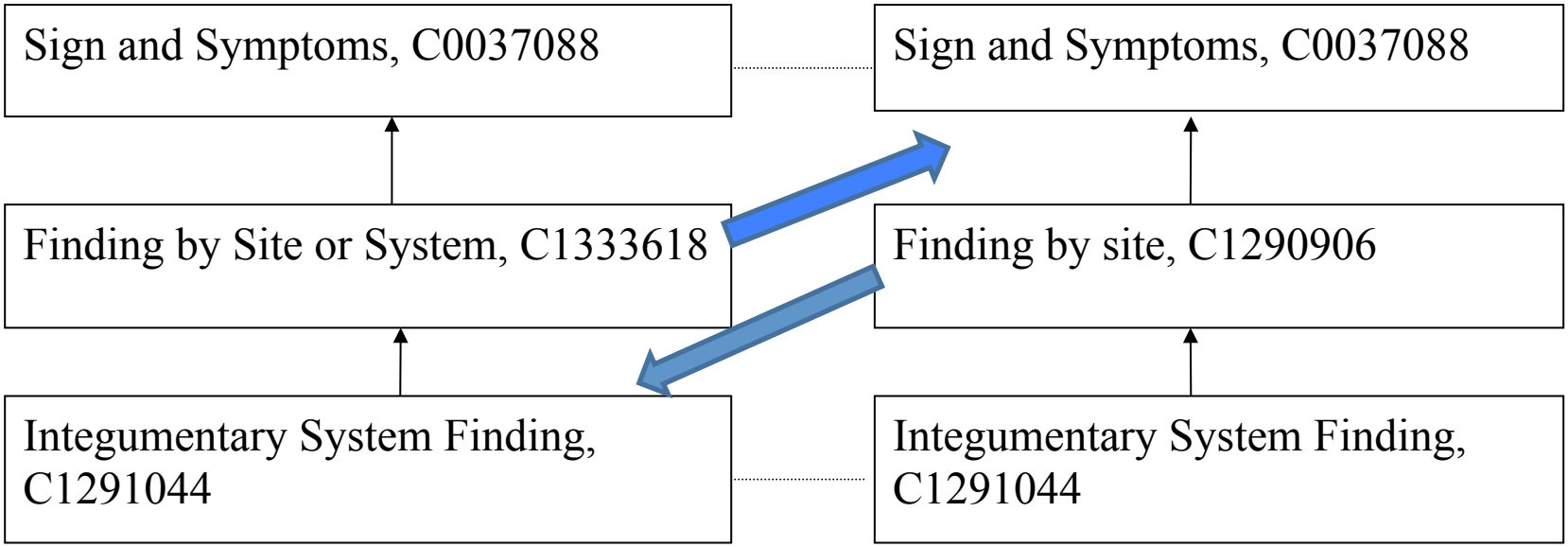
Example: Alternative Classification



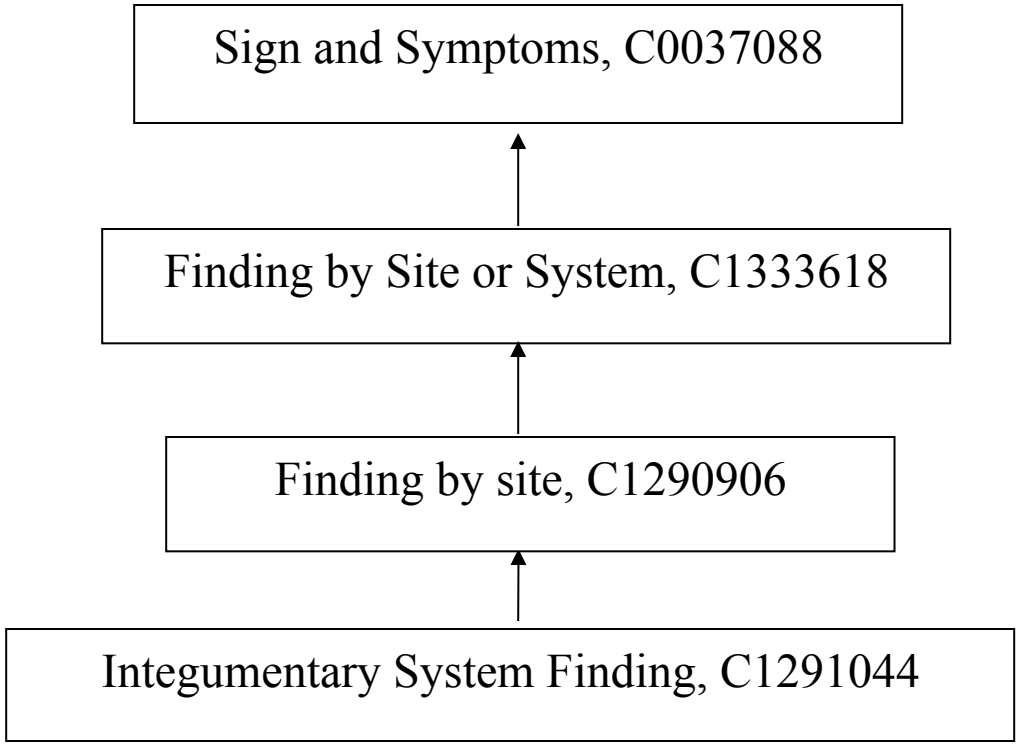
Making Explicit an Implicit Assumption of the Two Original Terminology Designers



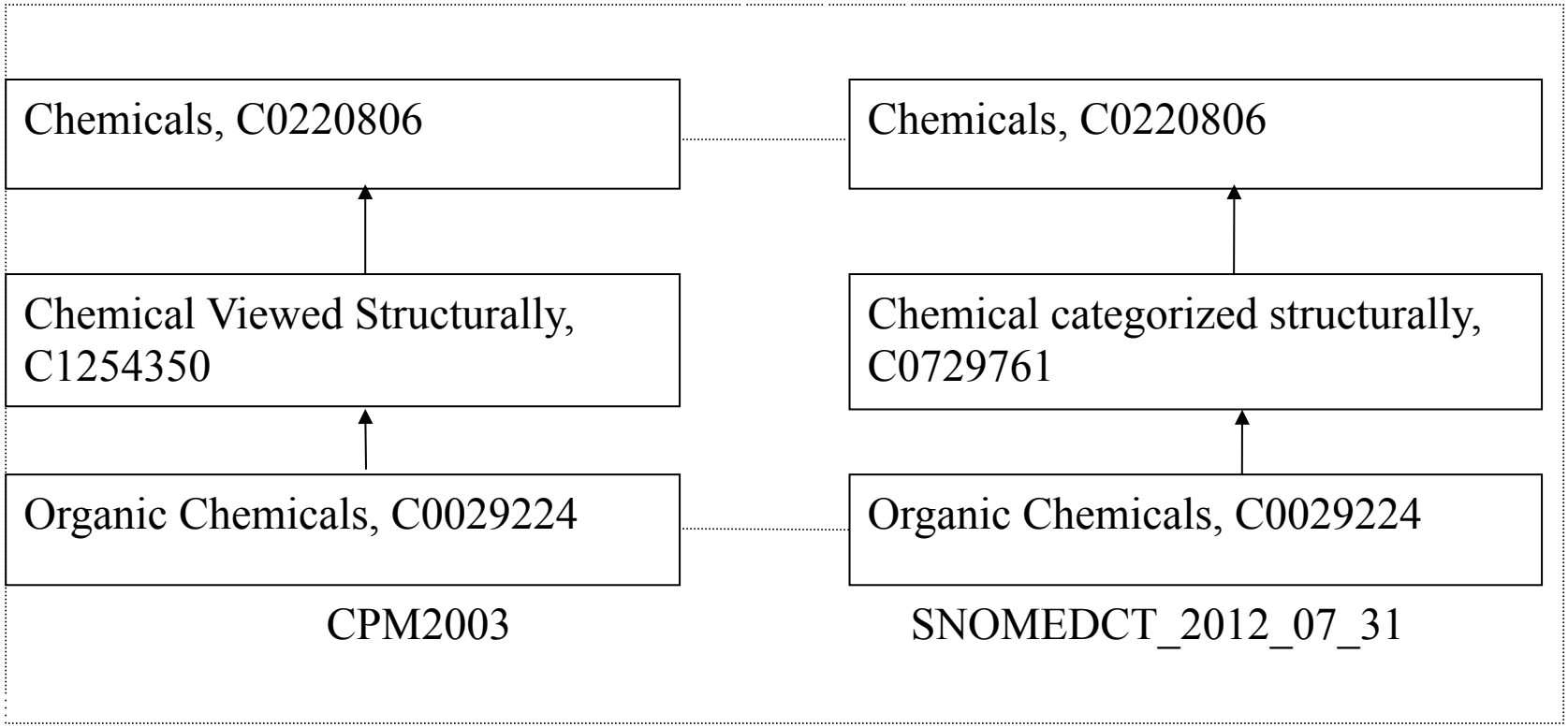
Example: Parent of the Other



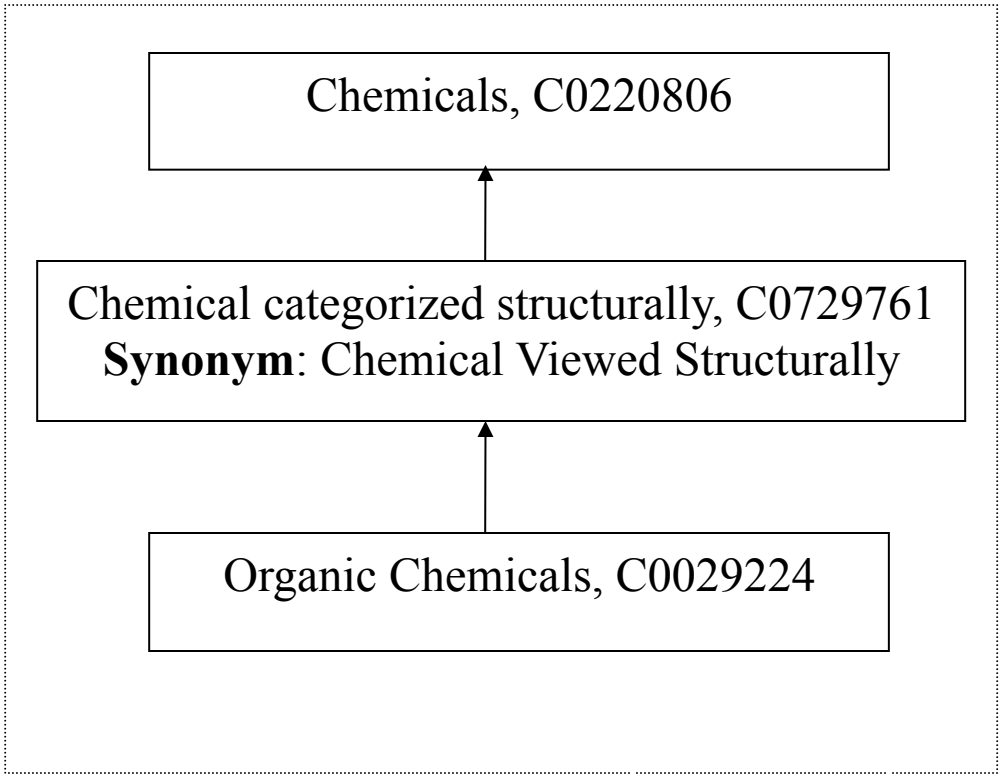
Suggestion: Concept Import



Example: Synonym of the Other



Suggestion: Merge Two Synonymous Concepts



Limitations

- Harmonization cannot be done without the consent of terminology curators
- All the terminologies are in UMLS Rich Release format

Future Work

- More complex configurations: more intermediate concepts
- Algorithm to identify the relationships between intermediate concepts in complex configurations
- Pairs of any two terminologies

Conclusions

- Six reference terminologies of the UMLS vs. SNOMED
- In a sample of 241 congruency pairs
  - 143 out of 241 (59.3%) concept pairs: alternative classification.
  - 47 out of 241 (23.6%) concept pairs: parent-child relationships.
  - 35 (14.5%) new synonyms
  - Three pairs of concepts indicated errors
- Take home message:
  - A semi-automated way based on common structure of the UMLS may complement existing human-expert centered methods to find potential concepts for import and export to a terminology.

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Thanks!

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