Large Formal Ontologies for Biomedicine

Panel

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What does UMLS stand for?

- **Unified**
- **Medical**
- **Language**
- **System**
Motivation

- Started in 1986
- National Library of Medicine
- “Long-term R&D project”

«[...] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

- The first is the variety of ways the same concepts are expressed in different machine-readable sources and by different people.
- The second is the distribution of useful information among many disparate databases and systems.»
UMLS 3 components

- **Metathesaurus**
  - Concepts
  - Inter-concept relationships

- **Semantic Network**
  - Semantic types
  - Semantic network relationships

- **Lexical resources**
  - SPECIALIST Lexicon
  - Lexical tools
UMLS Metathesaurus

Metathesaurus

Concepts

Heart

Esophagus

Mediastinum

Saccular Viscus

Angina Pectoris

Cardiotonic Agents

Tissue Donors

Left Phrenic Nerve

Heart Valves

Fetal Heart

4

9

31

97

225

22

12

9

31
Metathesaurus Large (2004AB)

- 134 source vocabularies
  - 73 families of vocabularies

- 1M concepts
  - 4.3M distinct concept names

- 15M relations
Metathesaurus Essentially informal

- By design
  - Terminology integration project
- By mandate
  - Vehicle for what is asserted in the source vocabularies
- Because if its purpose
  - Influenced by information retrieval
- No formalism
- Manual curation, but insufficient guidelines
UMLS Semantic Network
Semantic Network  More formal

- Manually created
  - Regardless of the position of the corresponding terms in existing vocabularies

- No particular formalism
  - Triples $<ST_1, rel, ST_2>$

- Quantification: some value from
  - $<Drug, treats, Disease or Syndrome>$
  - some Drug treats some Disease or Syndrome
Semantic Network  But small

- 135 semantic types
- 54 relationships
- 558 relations
Can the Metathesaurus be more formal?

◆ Probably not given its design

◆ Trade-offs
  ● Ingredient/drug synonymy
  ● Clinical synonymy/linguistic synonymy

◆ But some recent changes in its representation may help
  ● Source transparency: relations are no longer recorded at the concept level, but at the atom level
Couldn’t the Metathesaurus use another formalism (e.g., OWL)?

- Simply changing the formalism would not help
- What is needed are more precise, better defined relationships (isa vs. parent/child)
  - A trivial change of formalism would (wrongly) assume isa for each parent/child relationship
- Transforming portions of the UMLS in DL is resource intensive
- The OWLization of other terminology systems has not produced convincing results
Towards more consistency

- Many studies have investigated consistency issues in the Metathesaurus
- Some have developed algorithms, e.g.,
  - Identify and remove circular hierarchical relations
  - Use Semantic Network relations to assess the validity of Metathesaurus relations
  - ...

...
Conclusions

◆ Metathesaurus
  ● Large and informal
  ● But could be used as the basis for deriving an ontology of biomedicine
    – Manual curation
    – Lexical methods for acquiring ontological relations from terminology
    – Would still require an upper-level ontology
    – And a formalism
Medical Ontology Research

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