NIAID Ontology Workshop

Focus on Immunology

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UMLS and Semantic Web

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What does UMLS stand for?

- Unified
- Medical
- Language
- System

UMLS®
Unified Medical Language System®
UMLS Metathesaurus®

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.»

Unified Medical Language System

- SPECIALIST Lexicon
  - 200,000 lexical items
  - Part of speech and variant information
- Metathesaurus
  - 5M names from over 100 terminologies
  - 1M concepts
  - 16M relations
- Semantic Network
  - 135 high-level categories
  - 7000 relations among them

Lexical resources

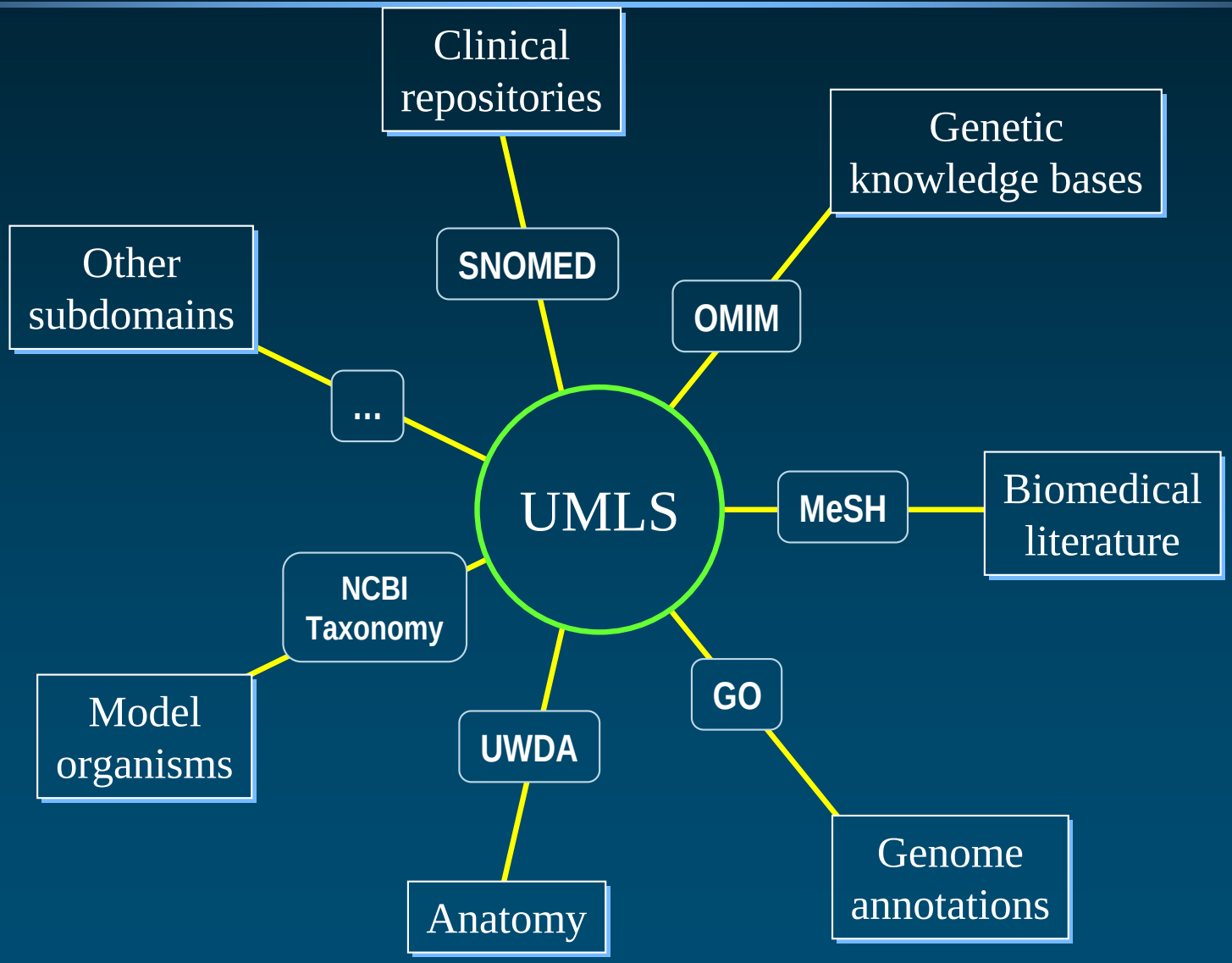
Terminological resources

Ontological resources

Source Vocabularies (2006AA)

- 140 source vocabularies
  - 17 languages
- Broad coverage of biomedicine
  - 5M names
  - 1.3M concepts
  - 16M relations
- Common presentation

Integrating subdomains



Addison's Disease: Concept

Disease or Syndrome

Addison's Disease

ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)
ADRENOCORTICAL INSUFFICIENCY, PRIMARY FAILURE
Addison melanoderma
Melasma addisonii
Primary adrenal deficiency
Asthenia pigmentosa
Bronzed disease
Insufficiency, adrenal primary
Primary adrenocortical insufficiency
Addison's, disease

MALADIE D'ADDISON - French
Addison-Krankheit - German
Morbo di Addison - Italian
DOENCA DE ADDISON - Portuguese
ADDISONOVA BOLEZN' - Russian
ENFERMEDAD DE ADDISON - Spanish

SNOMED
MeSH
AOD
Read Codes
…

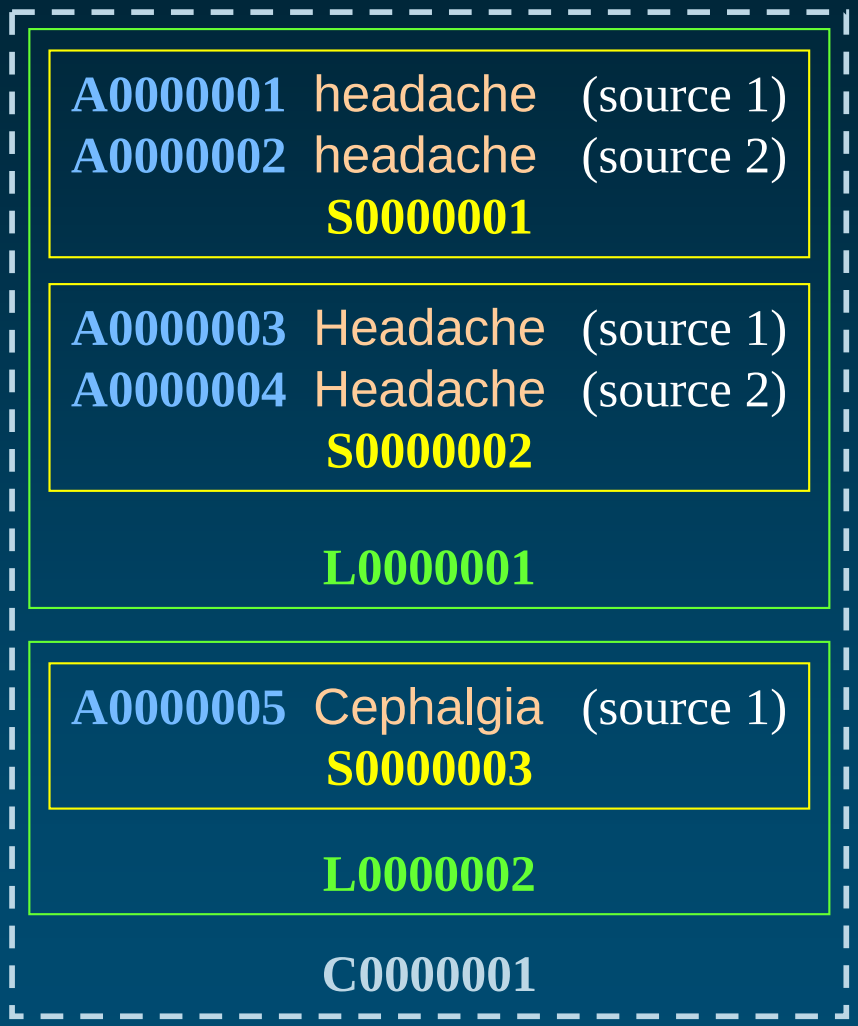
C0001403

A disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin. It is due to tuberculosis- or autoimmune-induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. In the absence of replacement therapy, it is usually fatal.

NLM

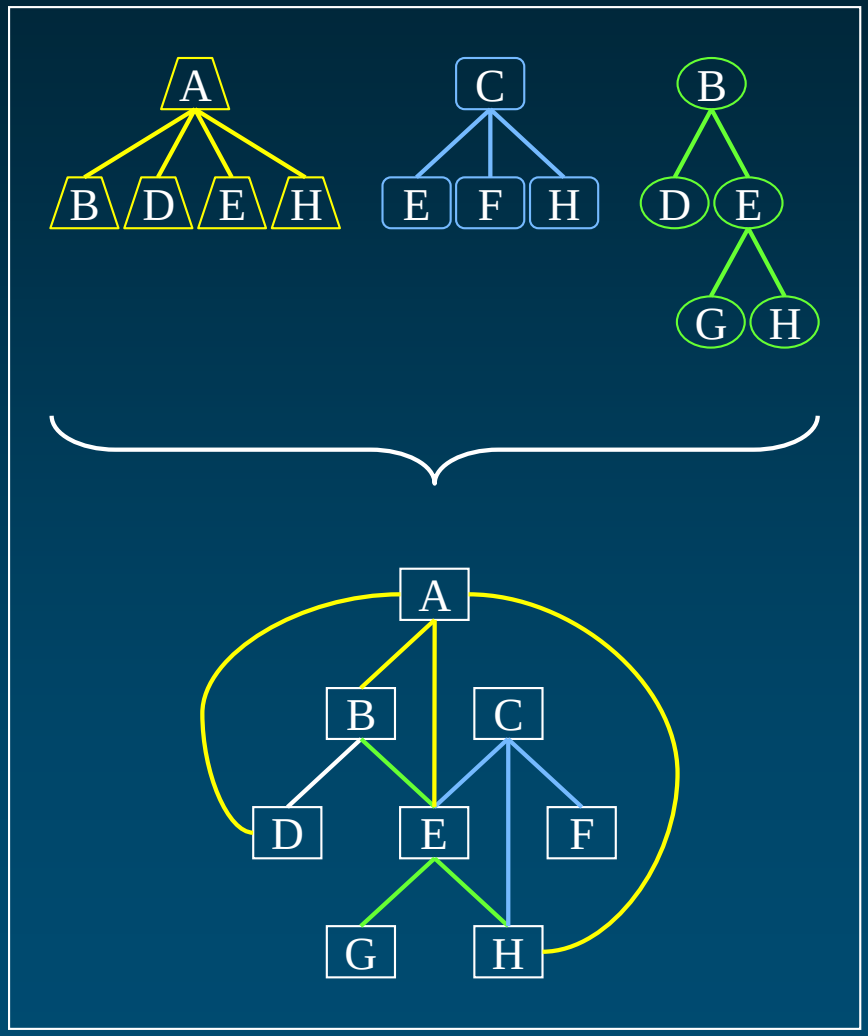
Metathesaurus Concepts (2005AB)

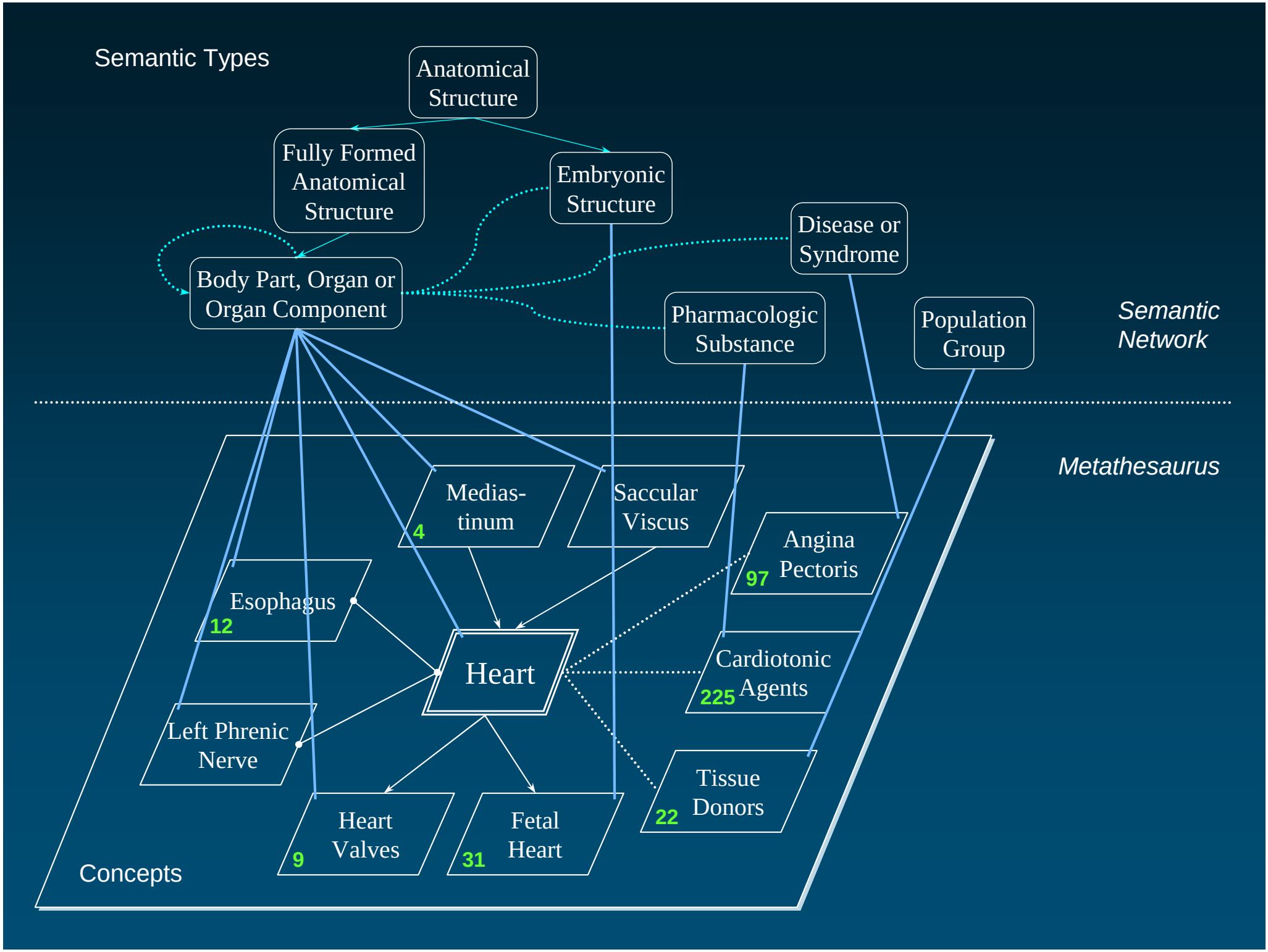
- Concept (~ 1.2 M) CUI
  - Set of synonymous concept names
- Term (~ 4.2 M) LUI
  - Set of normalized names
- String (~ 4.8 M) SUI
  - Distinct concept name
- Atom (~ 5.6 M) AUI
  - Concept name in a given source



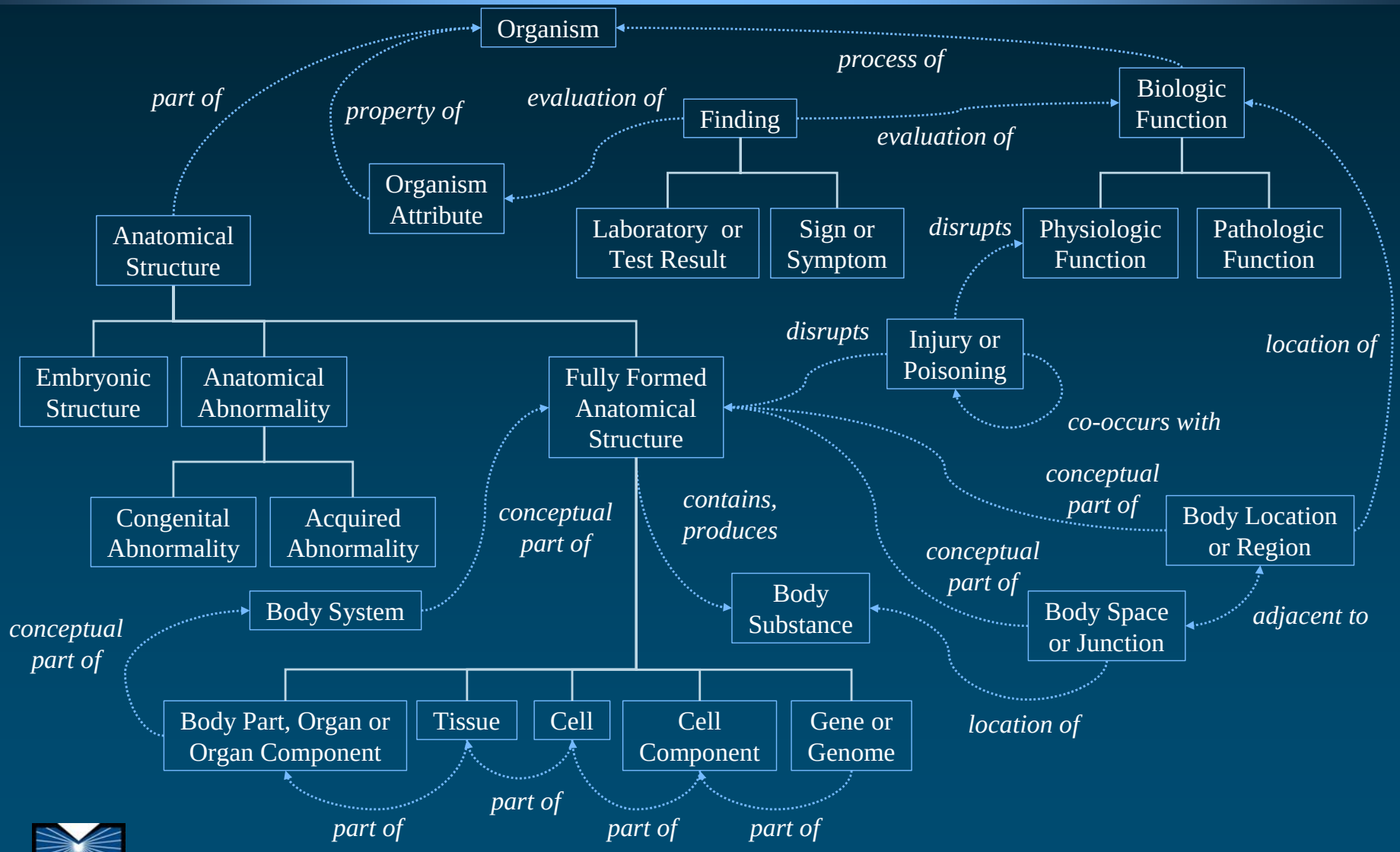
Organize concepts

- Inter-concept relationships: hierarchies from the source vocabularies
- Redundancy: multiple paths
- One graph instead of multiple trees (multiple inheritance)

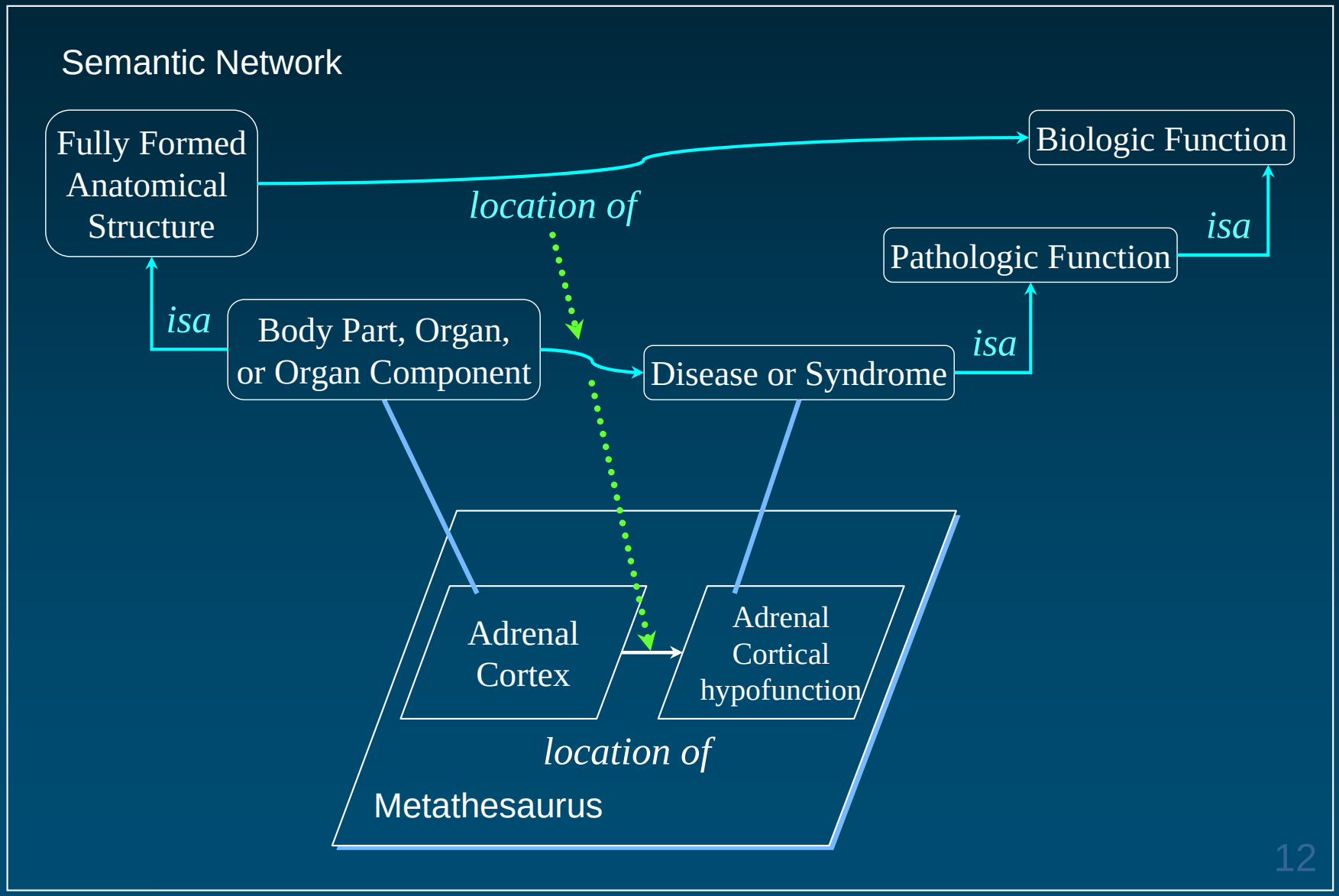



Semantic Types
Anatomical Structure
Fully Formed Anatomical Structure
Embryonic Structure
Disease or Syndrome
Body Part, Organ or Organ Component
Pharmacologic Substance
Population Group
Semantic Network
Metathesaurus
Medias-tinum
4
Saccular Viscus
Angina Pectoris
97
Esophagus
12
Heart
Cardiotonic Agents
225
Left Phrenic Nerve
Tissue Donors
22
Heart Valves
9
Fetal Heart
31
Concepts

Semantic Network relations



Relationships can inherit semantics



Immunology in the UMLS

- Semantic Network
  - Immunologic Factor
- Metathesaurus
  - Immunologic factors (C0021054)
  - Immunity (C0020964)
  - Autoimmune diseases (C0004364)
  - Vaccines (C0042210)

{UMLS_2005AA} UMLS® Semantic Navigator ¤ [2.12] - Mozilla Firefox

File Edit View Go Bookmarks Tools Help

http://mor.nlm.nih.gov/perl/semnav.pl

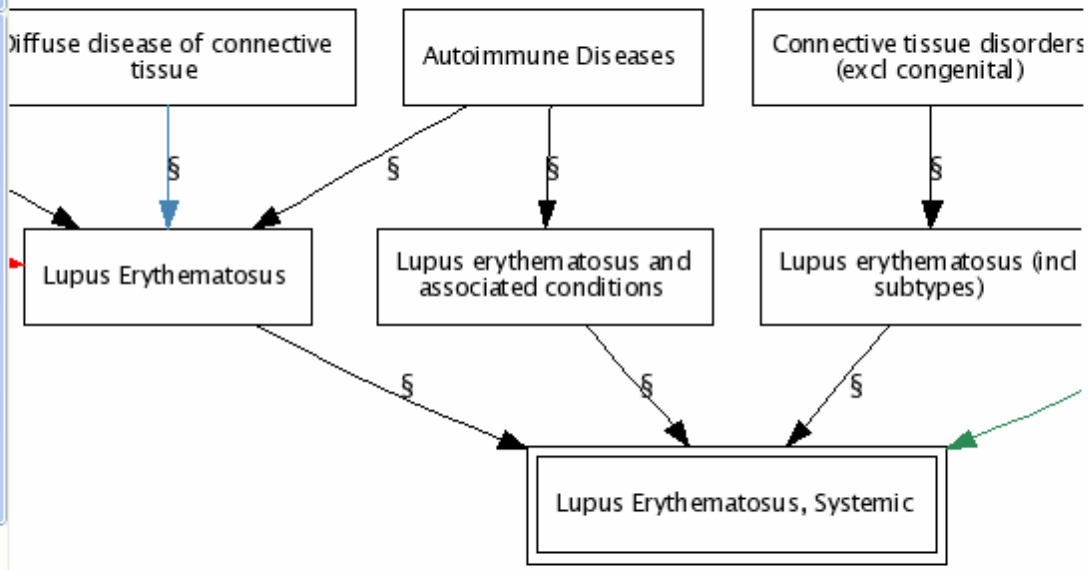
Siblings

Concepts & Ideas

- None of the above ¤
- Unsure of diagnosis ¤

Disorders

- Inflammatory Monoarthritis ¤
- Lupoid hepatic cirrhosis ¤
- Lupus endocarditis ¤
- Lupus Erythematosus, Cutaneous ¤
- Lupus erythematosus, localized, discoid ¤
- Lupus erythematosus, subacute ¤
- Lupus-like syndrome ¤



Descendants (first generation)

Disorders

- Acute systemic lupus erythematosus ¤
- Bullous systemic lupus erythematosus ¤
- dementia; systemic lupus erythematosus (etiology) ¤
- Drug-induced lupus erythematosus ¤
- Fulminating systemic lupus erythematosus ¤
- LE syndrome aggravated ¤
- LE test abnormality ¤

Other Related Concepts

Anatomy

- Connective Tissue ¤
- LE Cells ¤

Chemicals & Drugs

- Aurothioglucose ¤
- Aurothioglucose

- Anti-Arrhythmia Agents [8] ¤
- Anti-Bacterial Agents [30] ¤
- Antibodies [39] ¤
- Antibodies, Anticardiolipin [12
- Antibodies, Anti-Idiotypic [34
- Antibodies, Antinuclear [407]
- Antibodies, Bacterial [9] ¤
- Antibodies, Catalytic [8] ¤

BCI **Lupus Erythematosus, Systemic** LEGEND *

Start again | Apply new parameters

Restrict to vocabulary: Show all

Highlight vocabulary: Nothing

Similar Concepts

- Lupus Nephritis ¤
- SLE

Closest MeSH Terms

Main Headings

Done

Semantic Web

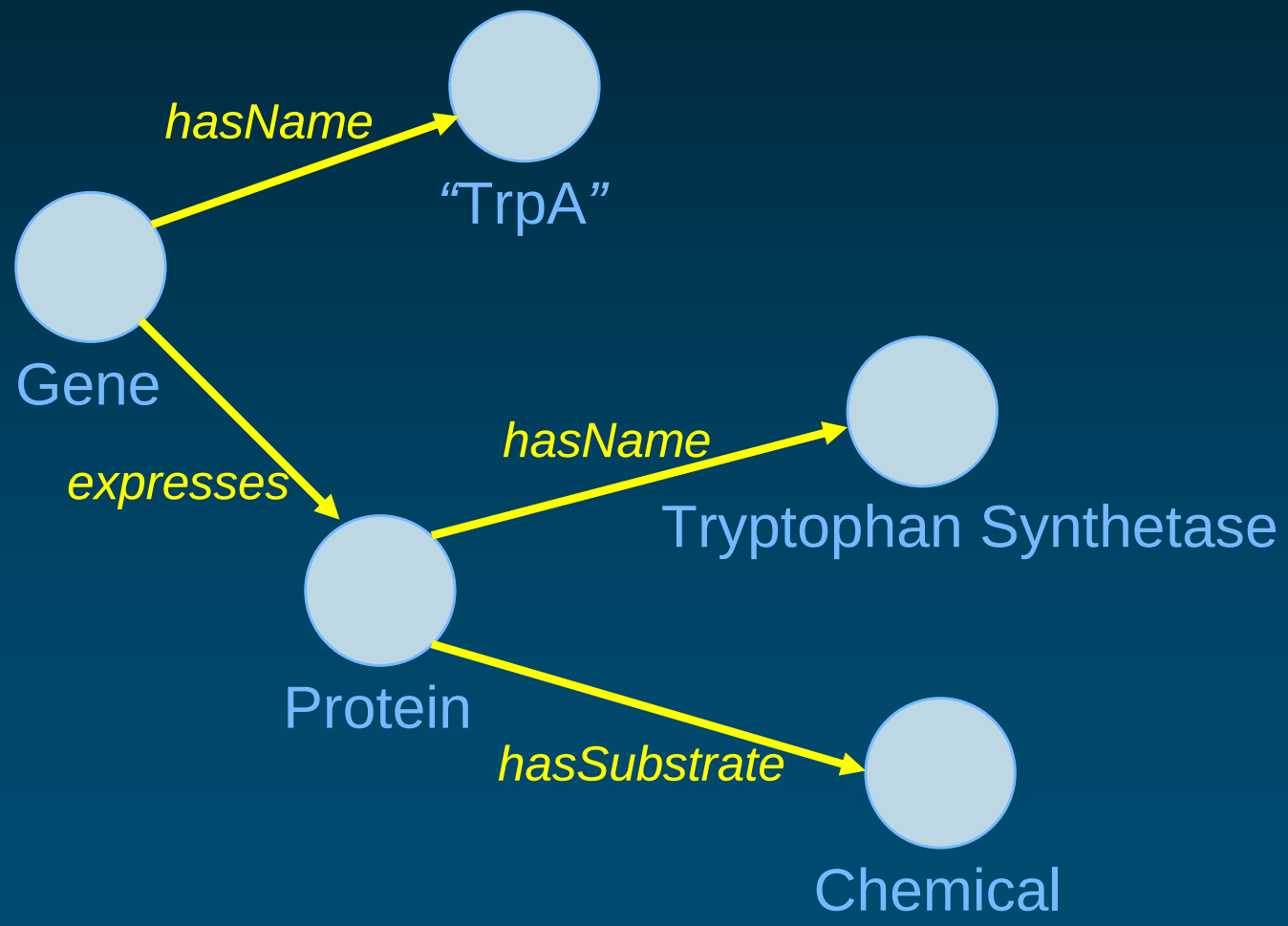
Web vs. Semantic web

- Web
  - For humans
  - Cosmetic markup
  - Knowledge embedded in text
  - Heterogeneity
  - Limited interoperability
- Semantic web
  - For humans and machines
  - Semantic markup
  - Knowledge represented explicitly
    - Semantic annotation (RDF)
    - Ontologies
  - Integrates heterogeneous resources
  - Interoperability

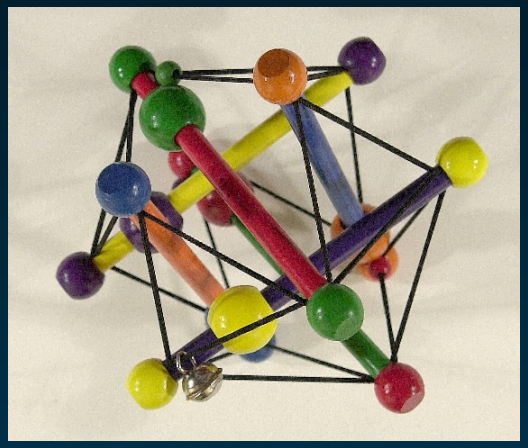
Knowledge in the semantic web

- Uniform Resource Identifier (URI)
  - Identify entities and relations
  - E.g., GO or UMLS identifier
- Resource Description Framework (RDF)
  - Triples ($entity_1$, relationship, $entity_2$)
  - Common syntax
  - Semantic bus for applications

RDF triples


hasName
"TrpA"
Gene
expresses
hasName
Tryptophan Synthetase
Protein
hasSubstrate
Chemical

Issues and Challenges



Medical Ontology Research

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UMLS References

- UMLS
  umlsinfo.nlm.nih.gov
- UMLS browsers
  (free, but UMLS license required)
  - Knowledge Source Server: **umlsks.nlm.nih.gov**
  - Semantic Navigator:
    http://mor.nlm.nih.gov/perl/semnav.pl
  - RRF browser
    (standalone application distributed with the UMLS)

UMLS References

- Recent overviews
  - Bodenreider O. (2004). The Unified Medical Language System (UMLS): Integrating biomedical terminology. *Nucleic Acids Research*; D267-D270.
  - Nelson, S. J., Powell, T. & Humphreys, B. L. (2002). The Unified Medical Language System (UMLS) Project. In: Kent, Allen; Hall, Carolyn M., editors. *Encyclopedia of Library and Information Science*. New York: Marcel Dekker. p.369-378.

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- UMLS as a research project
  - Lindberg, D. A., Humphreys, B. L., & McCray, A. T. (1993). The Unified Medical Language System. *Methods Inf Med*, *32*(4), 281-91.
  - Humphreys, B. L., Lindberg, D. A., Schoolman, H. M., & Barnett, G. O. (1998). The Unified Medical Language System: an informatics research collaboration. *J Am Med Inform Assoc*, *5*(1), 1-11.

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- Technical papers
  - McCray, A. T., & Nelson, S. J. (1995). The representation of meaning in the UMLS. *Methods Inf Med*, *34*(1-2), 193-201.
  - Bodenreider O. & McCray A. T. (2003). Exploring semantic groups through visual approaches. *Journal of Biomedical Informatics*, 36(6), 414-432.

Semantic Web References

- World Wide Web Consortium (W3C)
  - http://www.w3.org/
- W3C Health Care and Life Sciences Interest Group
  - http://www.w3.org/2001/sw/hcls/