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The UMLS and the Semantic Web

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Outline

- The UMLS (in a nutshell)
  - Lexical resources
  - Metathesaurus
  - Semantic Network
- Why is the UMLS relevant to the Semantic Web?
- Issues and challenges

Unified Medical Language System (UMLS)

UMLS: 3 components

- 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

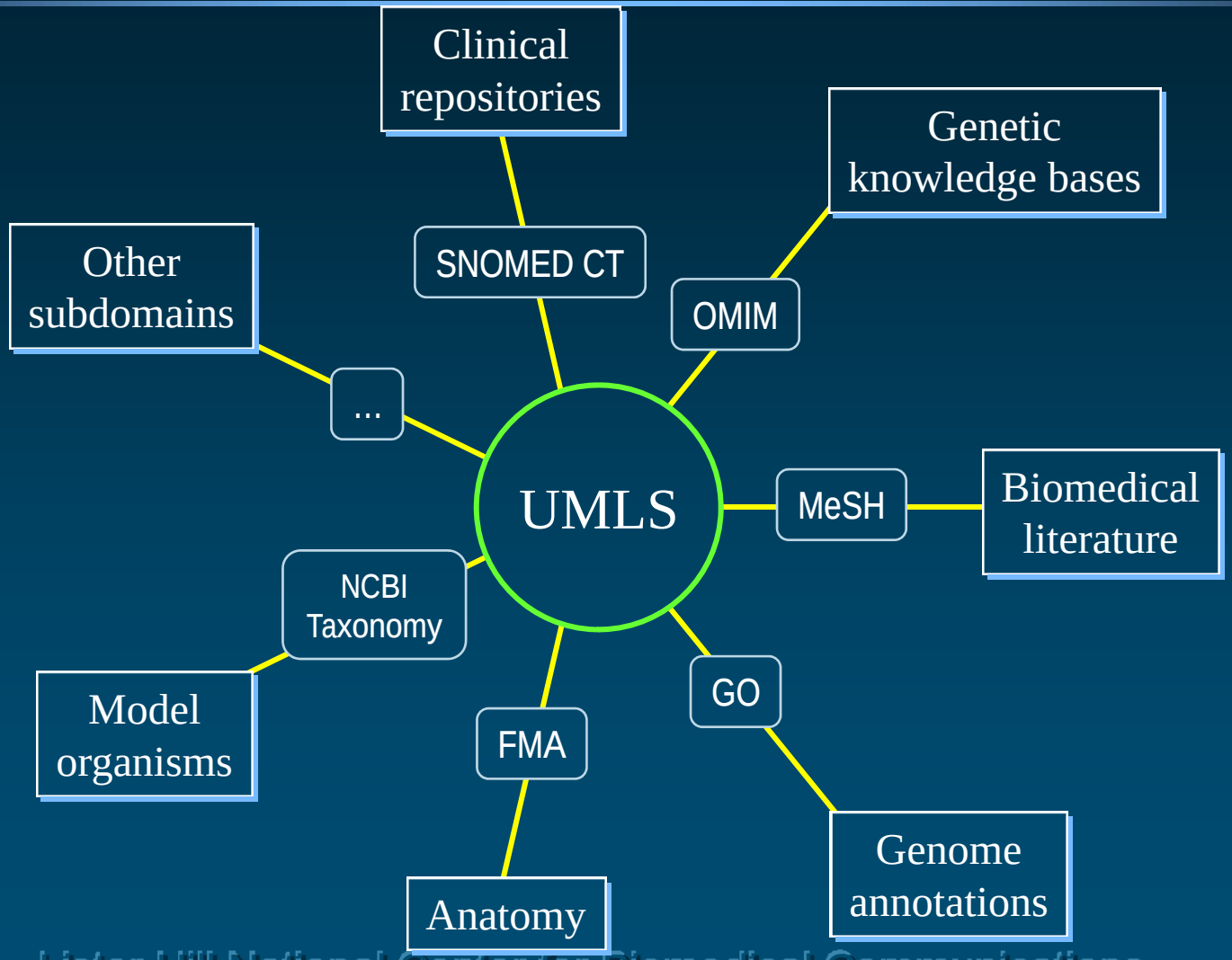
UMLS Characteristics (1)

- Current version: 2008AA (2-3 annual releases)
- Type: Terminology integration system
- Domain: Biomedicine
- Developer: NLM
- Funding: NLM (intramural)
- Availability
  - Publicly available: Yes* (cost-free license required)
  - Repositories: UMLS
- URL: http://umlsks.nlm.nih.gov/

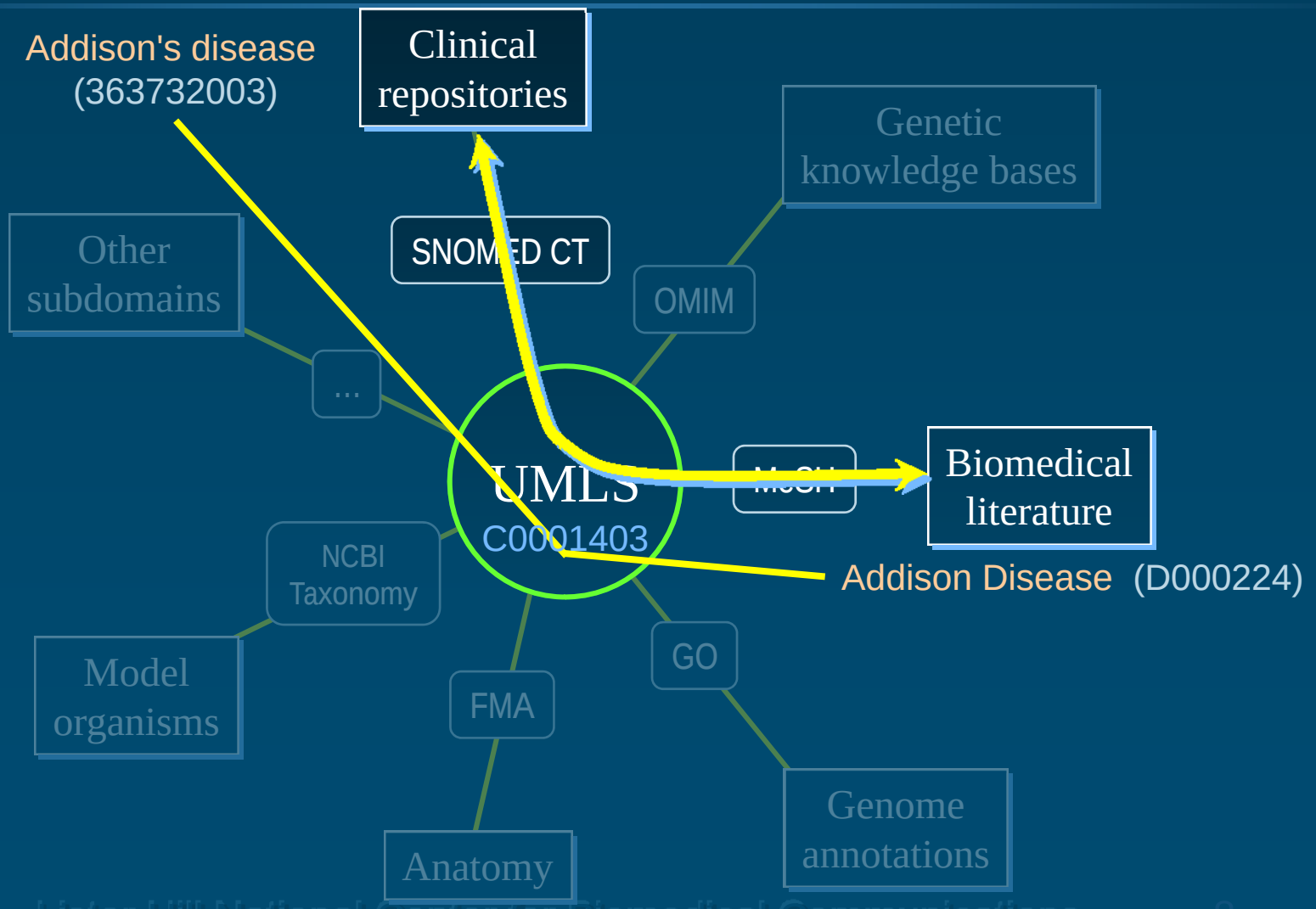
UMLS Characteristics (2)

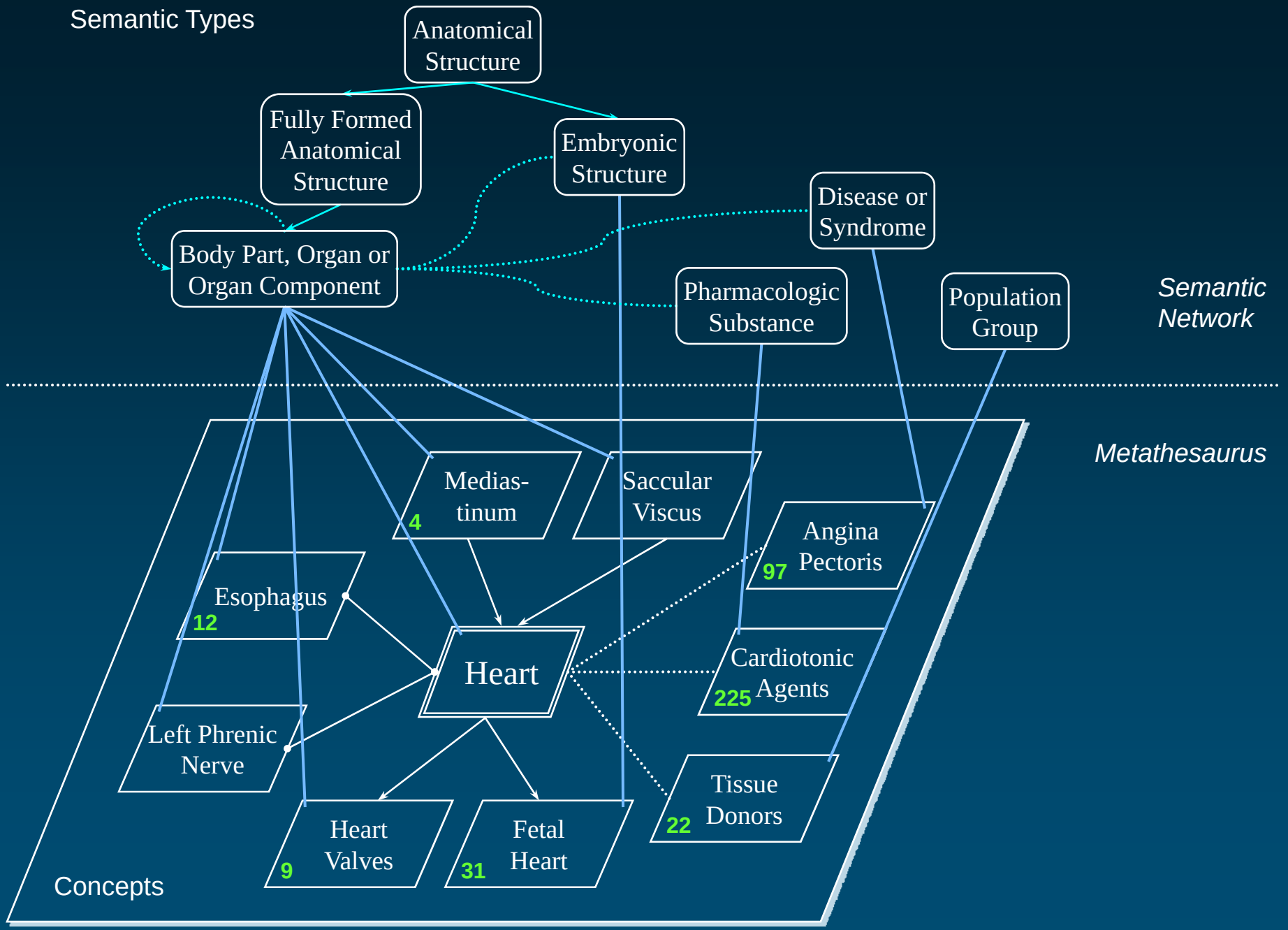
- Number of
  - Concepts: 1.5M (2008AA)
  - Terms: ~6M
- Major organizing principles (Metathesaurus):
  - Concept orientation
  - Source transparency
  - Multi-lingual through translation
- Formalism: Proprietary format (RRF)

UMLS Integrating subdomains



Trans-namespace integration




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

Why is the UMLS relevant to the Semantic Web?

Relevance to the SW Metathesaurus

- Terminology integration system
  - Trans-namespace integration
  - Integration beyond shared identifiers
- Repository of biomedical terminologies/ontologies
- Many UMLS vocabularies used for the annotation of datasets (including clinical records)

Relevance to the SW Metathesaurus

- Broad coverage of biomedicine
- Large user base
- Tooling available
  - E.g, visualization, named entity recognition, etc.

Relevance to the SW Semantic Network

- Top-level ontology of the biomedical domain
- Broad biomedical categories
- Helps partition biomedical concepts
- Semantic relations

Issues and Challenges

Issues and challenges

- Availability
  - Mandatory license agreement
- Discoverability
  - No metadata
- Formalism
  - No easy conversion to SKOS/RDF(S)/OWL
- Identifiers

- Steep learning curve

Availability

- Some source vocabularies have intellectual property restrictions
  - E.g., most drug vocabularies
  - Complex agreement for SNOMED CT: available at no cost for member countries of the IHTSDO
- Mandatory license agreement
  - No cost for research
  - May require negotiation with the vocabulary developer for production applications
- MetamorphoSys helps extract selected sources from the UMLS

Discoverability

- Discoverability of individual concepts
  - UMLSKS web services
  - Search all UMLS source vocabularies at the same time
  - Named entity recognition/normalization (e.g., MetaMap)
- Discoverability of terminologies/ontologies
  - No comprehensive registries
  - No rich registries
    - With rich metadata supporting the discoverability of terminologies/ontologies

Formalism

- UMLS: Proprietary format
  - Rich Release Format (RRF)
  - All terminologies/ontologies represented in the same format
- No easy conversion to SKOS/RDF(S)/OWL
  - Underspecified semantics
    - Child/parent ≠ subClassOf
  - Complex semantics
    - Descriptors / concepts / terms
  - Rich attribute set

Identifiers for biomedical entities

- What is identified?
  - Entity vs. resource about the entity
- Which identifier to pick?
  - E.g., Addison's disease
    - 363732003 (SNOMED CT)
    - D000224 (MeSH)
    - C0001403 (UMLS Metathesaurus)
- Which format?
  - URI vs. LSID
- Which authoritative source for minting URIs?
  - Ontology developers vs. (e.g.) Bio2RDF

Steep learning curve

- Large resource
  - 1.5M concepts
  - 6M terms
  - Over 20M relations
- Complex structure
  - Metathesaurus
  - Semantic Network
- Rich set of attributes
- Rich set of relations
  - Terminological
  - Semantic
  - Statistical
  - Mapping
- Multiple languages
- Complex domain

Conclusions

Conclusions

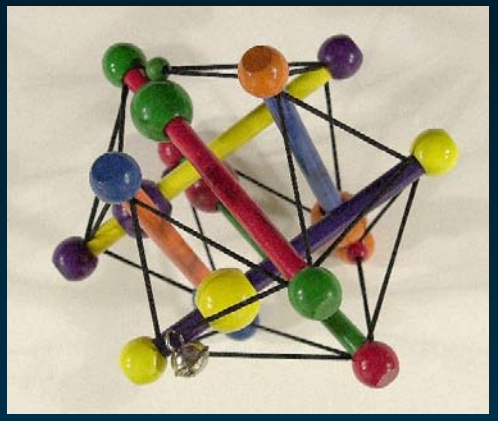
- UMLS as a terminology integration system
  - Helps bridge across namespaces
  - Helps integrate information sources
    - Beyond shared identifiers
- UMLS as a repository of terminologies/ontologies
  - Single source, single format for 143 vocabularies
- Issues with availability, discoverability and formalism
- Identifiers for biomedical entities

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)

References

- Recent overviews
  - Bodenreider O. (2004). The Unified Medical Language System (UMLS): Integrating biomedical terminology. *Nucleic Acids Research*; D267-D270.
  - Bodenreider O. From terminology integration to information integration: Unified Medical Language System (UMLS). BioRDF Teleconference, W3C Semantic Web Health Care and Life Sciences Interest Group, June 5, 2006. http://mor.nlm.nih.gov/pubs/pres/060605-BioRDF.pdf



Medical Ontology Research

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