Semantic Data Integration Workshop

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UMLS and semantic integration

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Outline

- Unified Medical Language System overview
  - *UMLS Metathesaurus*
  - *UMLS Semantic Network*
- Data integration questions

Uses of biomedical ontologies

- Knowledge management
  - Annotating data and resources
  - Accessing biomedical information
  - Mapping across biomedical ontologies
- Data integration, exchange and semantic interoperability
- Decision support
  - Data selection and aggregation
  - Decision support
  - NLP applications
  - Knowledge discovery

[Bodenreider, YBMI 2008]

Unified Medical Language System

Overview

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.»

The UMLS in practice

- Database
  - Series of relational files
- Interfaces
  - Web interface: Knowledge Source Server (UMLSKS)
  - Application programming interfaces (Java and XML-based)
- Applications
  - lvg (lexical programs)
  - MetamorphoSys (installation and customization)
  - RRF browser (browsing subsets)

The UMLS is *not* an end-user application

UMLS 3 components

- Lexical resources
  - SPECIALIST Lexicon
  - Lexical tools
- Metathesaurus
  - Concepts
  - Inter-concept relationships
- Semantic Network
  - Semantic types
  - Semantic network relationships

Lexical resources

Terminological resources

Ontological resources

UMLS Knowledge Sources

UMLS Metathesaurus

Metathesaurus Basic organization

- Concepts
  - Synonymous terms are clustered into a concept
  - Properties are attached to concepts, e.g.,
    - Unique identifier
    - Definition
- Relations
  - Concepts are related to other concepts
  - Properties are attached to relations, e.g.,
    - Type of relationship
    - Source

Source Vocabularies (2009AA)

- 152 source vocabularies
  - 19 languages
- Broad coverage of biomedicine
  - 9.7M names
  - 2.1M concepts
  - >10M relations
- Common presentation

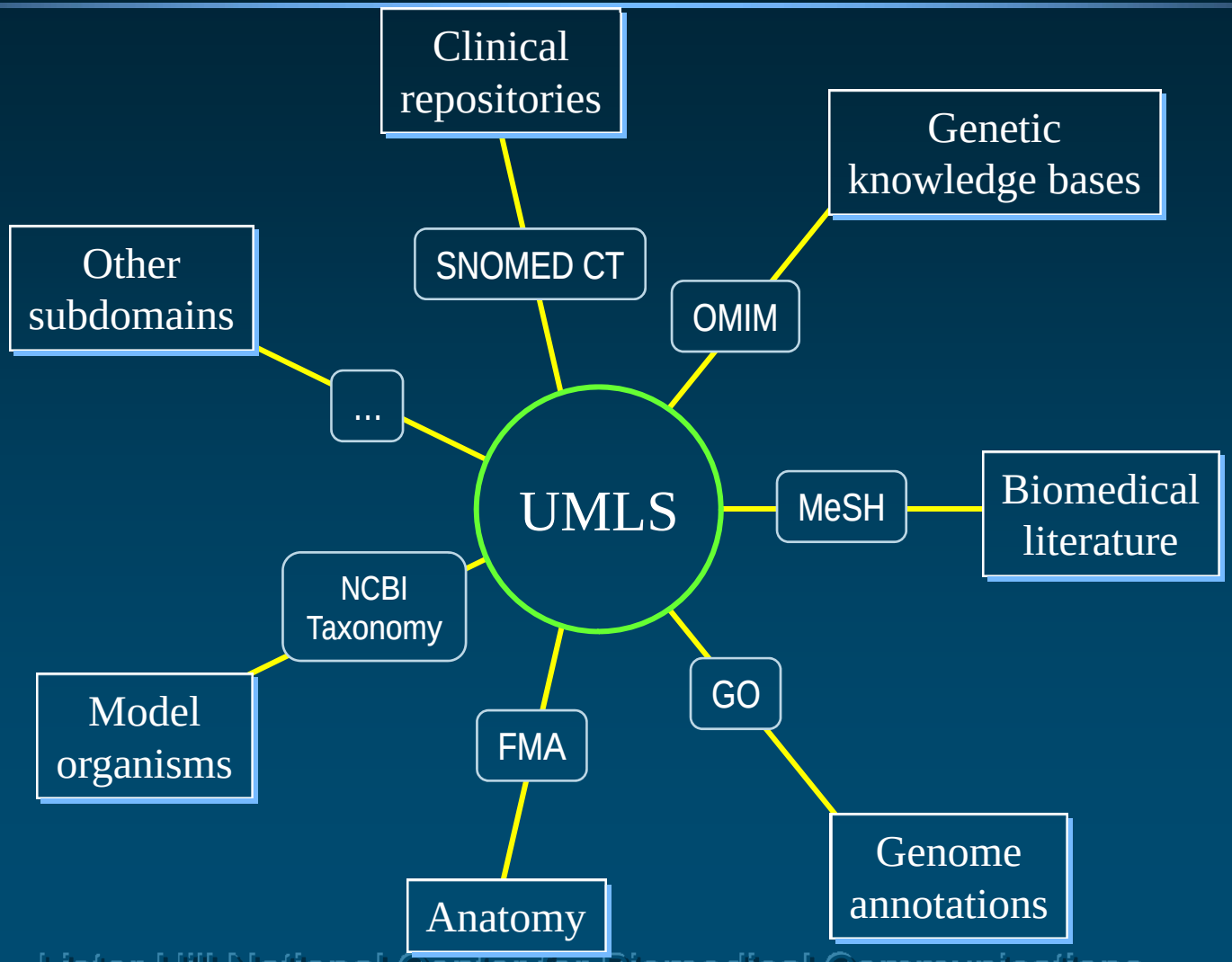
Biomedical terminologies

- General vocabularies
  - anatomy (UWDA, Neuronames)
  - drugs (RxNorm, First DataBank, Micromedex)
  - medical devices (UMD, SPN)
- Several perspectives
  - clinical terms (SNOMED CT)
  - information sciences (MeSH, CRISP)
  - administrative terminologies (ICD-9-CM, CPT-4)
  - data exchange terminologies (HL7, LOINC)

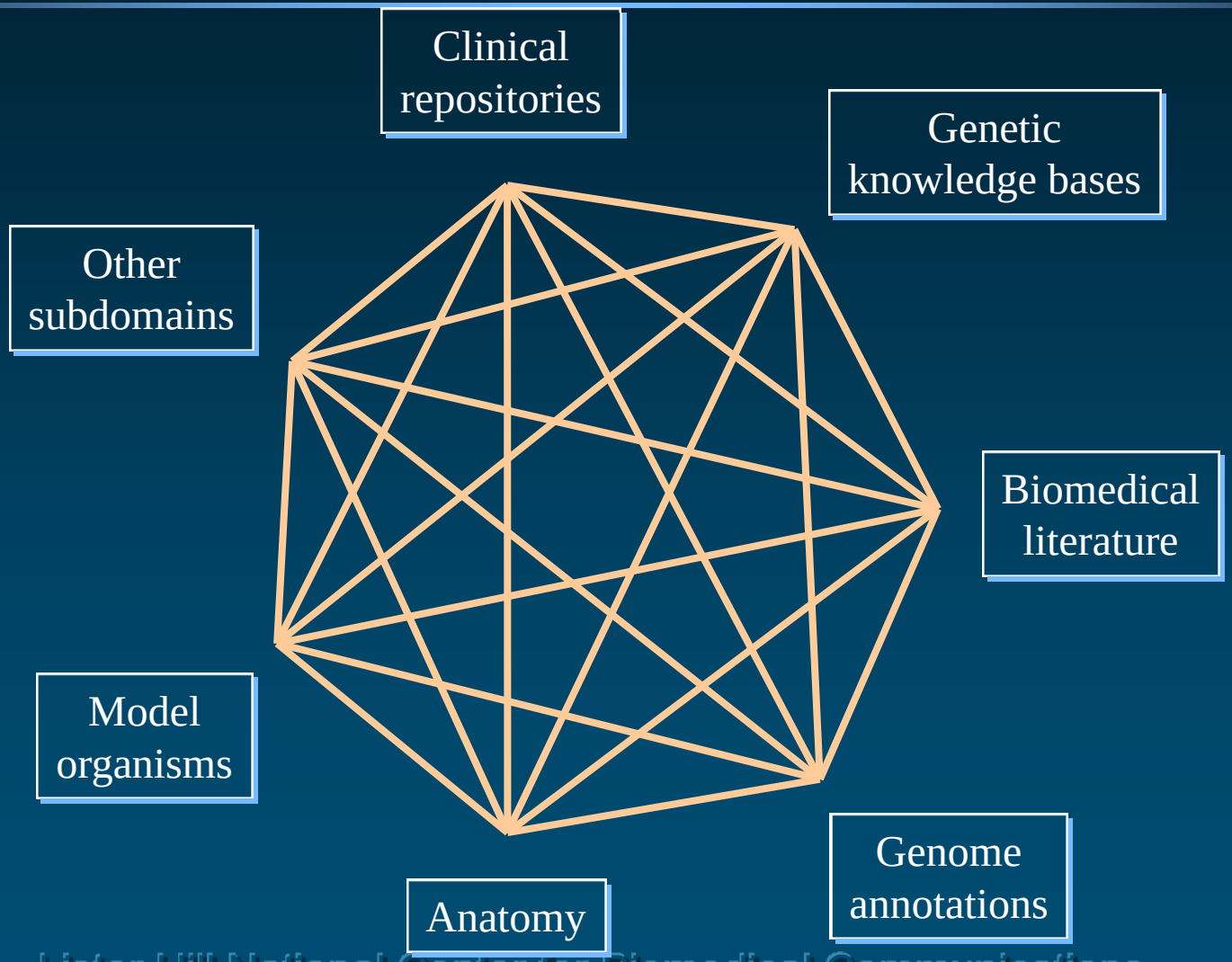
Biomedical terminologies (cont'd)

- Specialized vocabularies
  - nursing (NIC, NOC, NANDA, Omaha, PCDS)
  - dentistry (CDT)
  - oncology (PDQ)
  - psychiatry (DSM, APA)
  - adverse reactions (COSTART, WHO ART)
  - primary care (ICPC)
- Terminology of knowledge bases (AI/Rheum, DXplain, QMR)

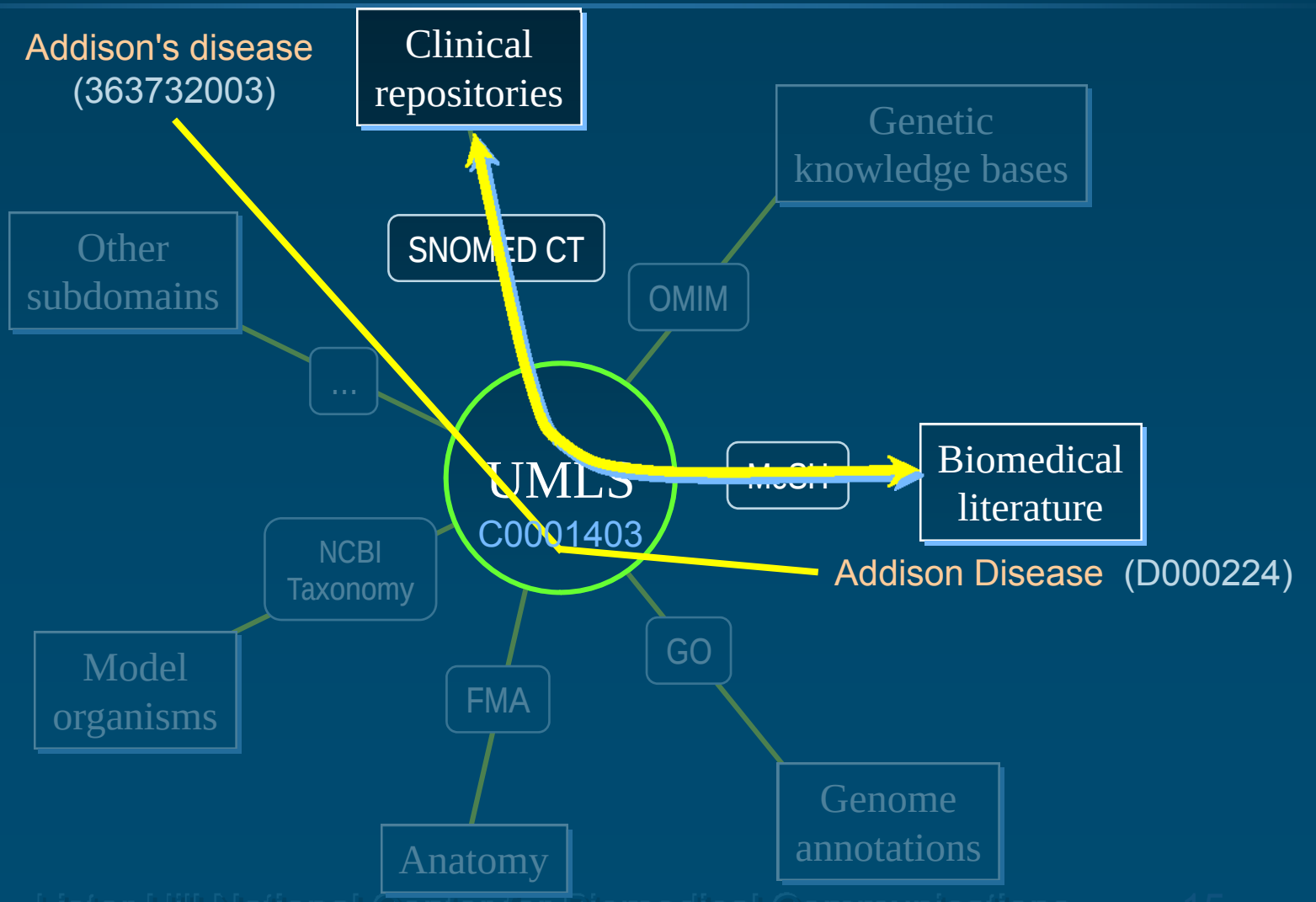
Integrating subdomains



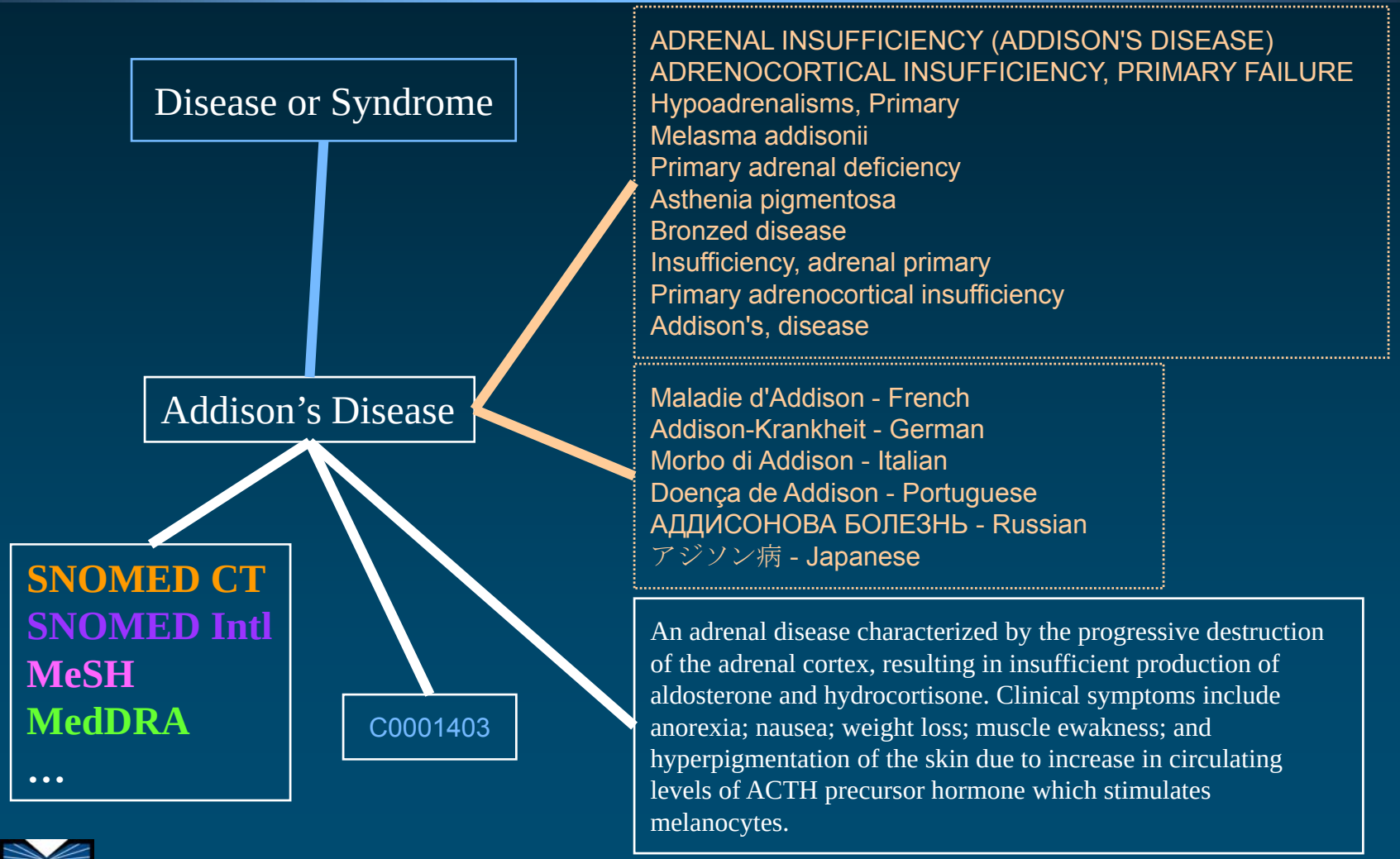
Integrating subdomains



Trans-namespace integration



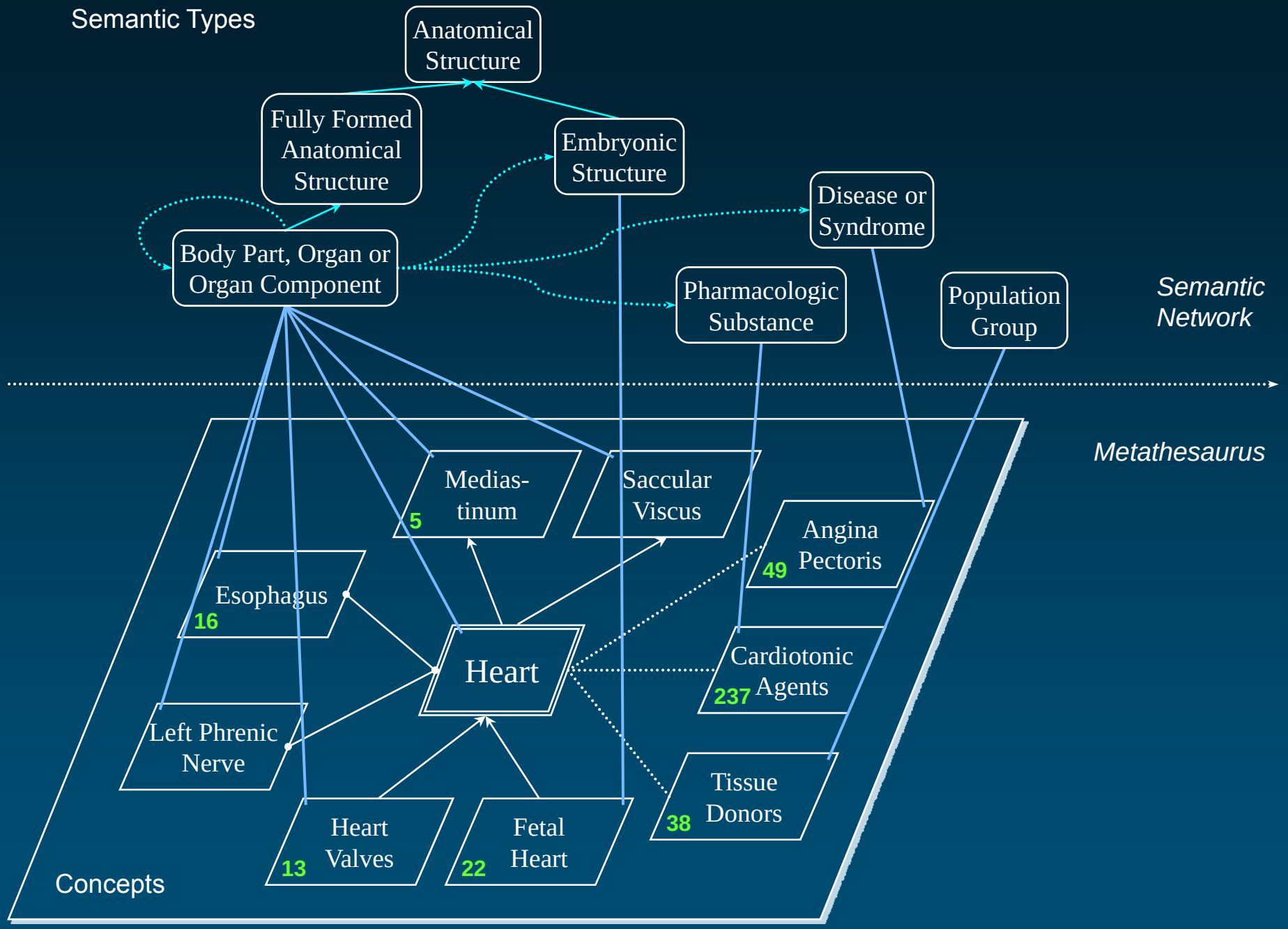
Addison's Disease: Concept



Metathesaurus Relationships

- Symbolic relations: ~8 M pairs of concepts
- Statistical relations : ~6 M pairs of concepts (co-occurring concepts)
- Mapping relations: ~150,000

- Categorization: Relationships between concepts and semantic types from the Semantic Network


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
5
Saccular Viscus
Angina Pectoris
49
Esophagus
16
Heart
Cardiotonic Agents
237
Left Phrenic Nerve
Tissue Donors
38
Heart Valves
13
Fetal Heart
22
Concepts

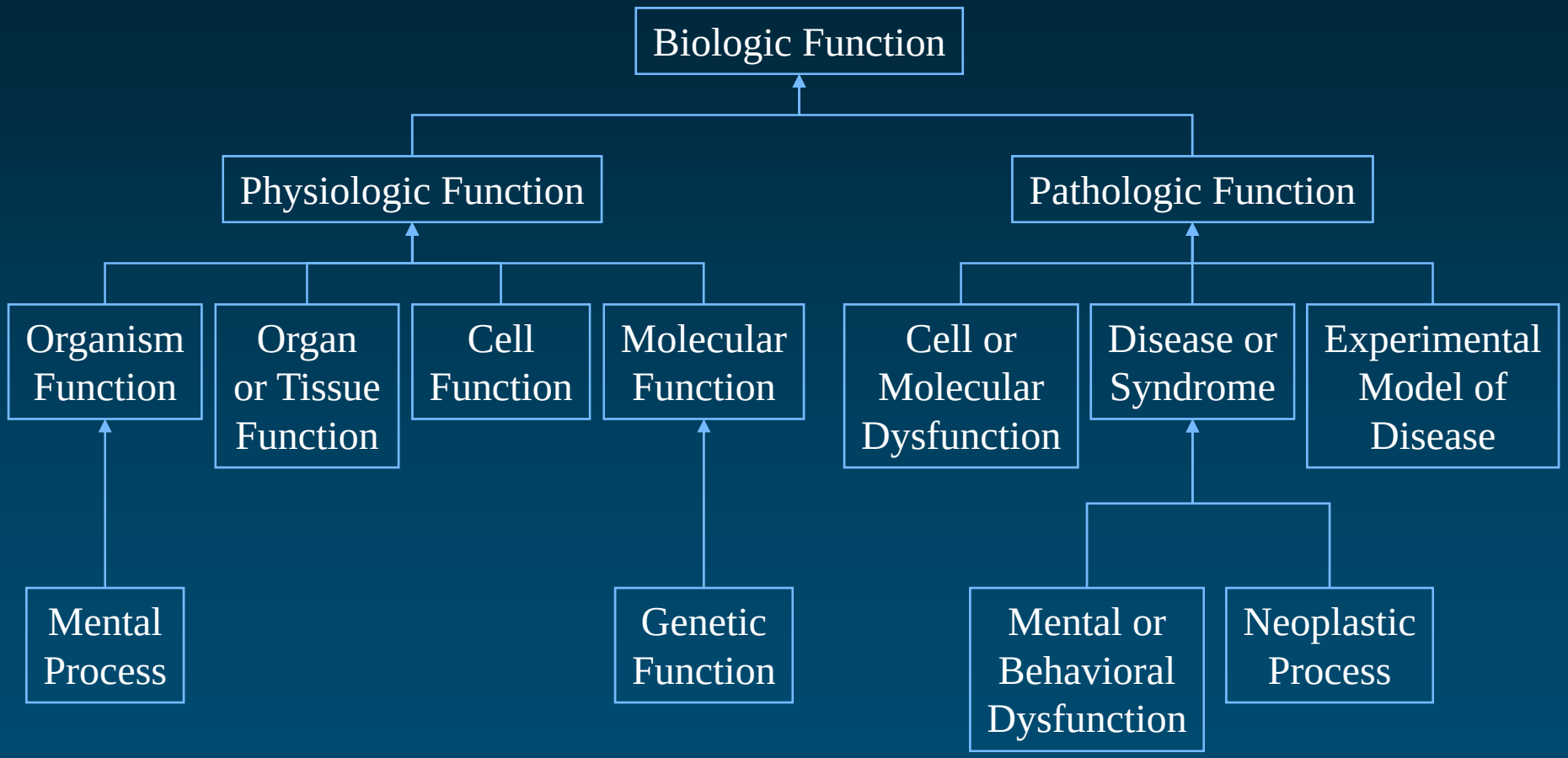
UMLS Knowledge Sources

UMLS Semantic Network

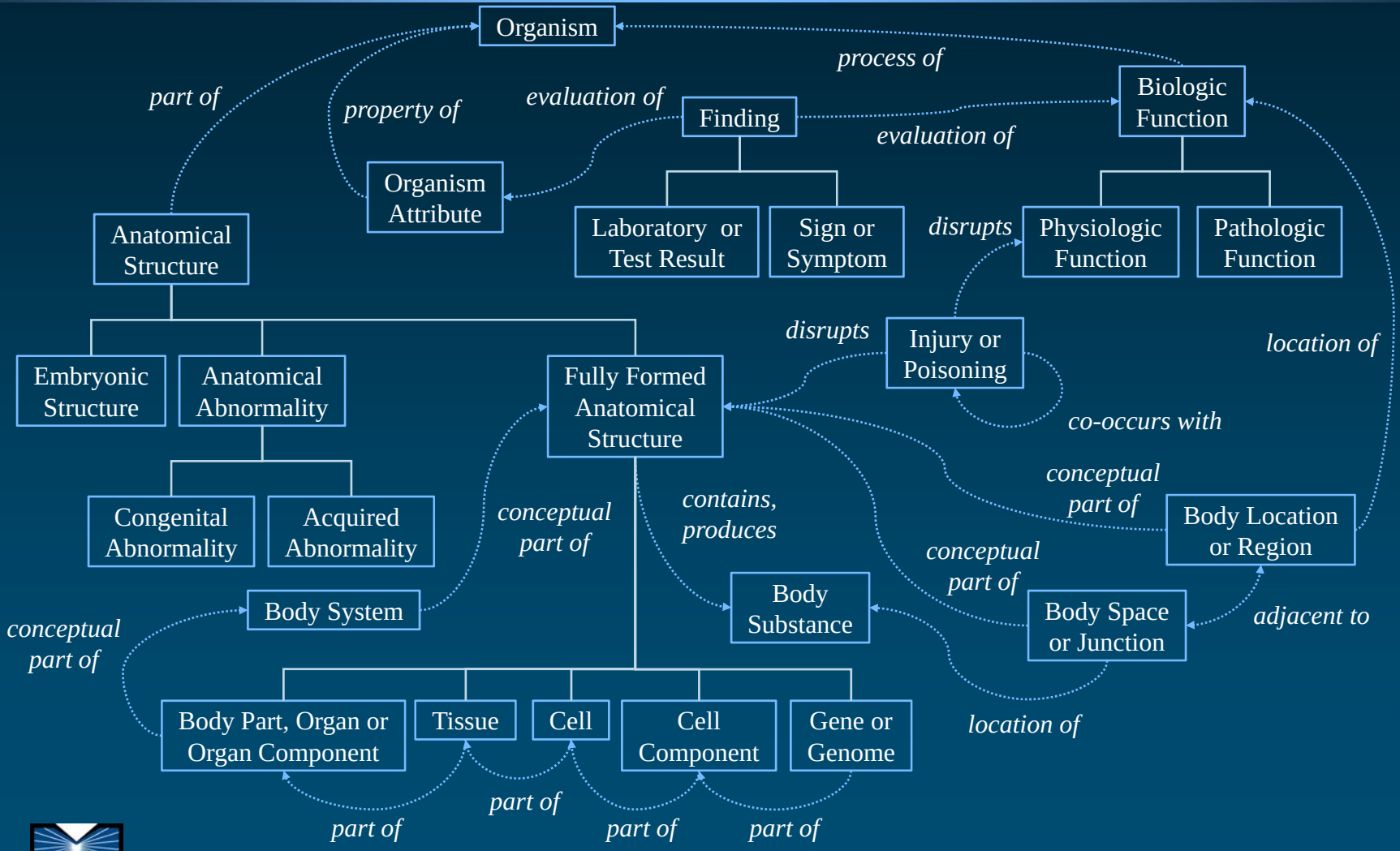
Semantic Network

- Semantic network relationships (54)
  - hierarchical (isa = is a kind of)
    - among types
      - Animal *isa* Organism
      - Enzyme *isa* Biologically Active Substance
    - among relations
      - treats *isa* affects
  - non-hierarchical
    - Sign or Symptom *diagnoses* Pathologic Function
    - Pharmacologic Substance *treats* Pathologic Function

“Biologic Function” hierarchy (isa)



Associative (non-isa) relationships

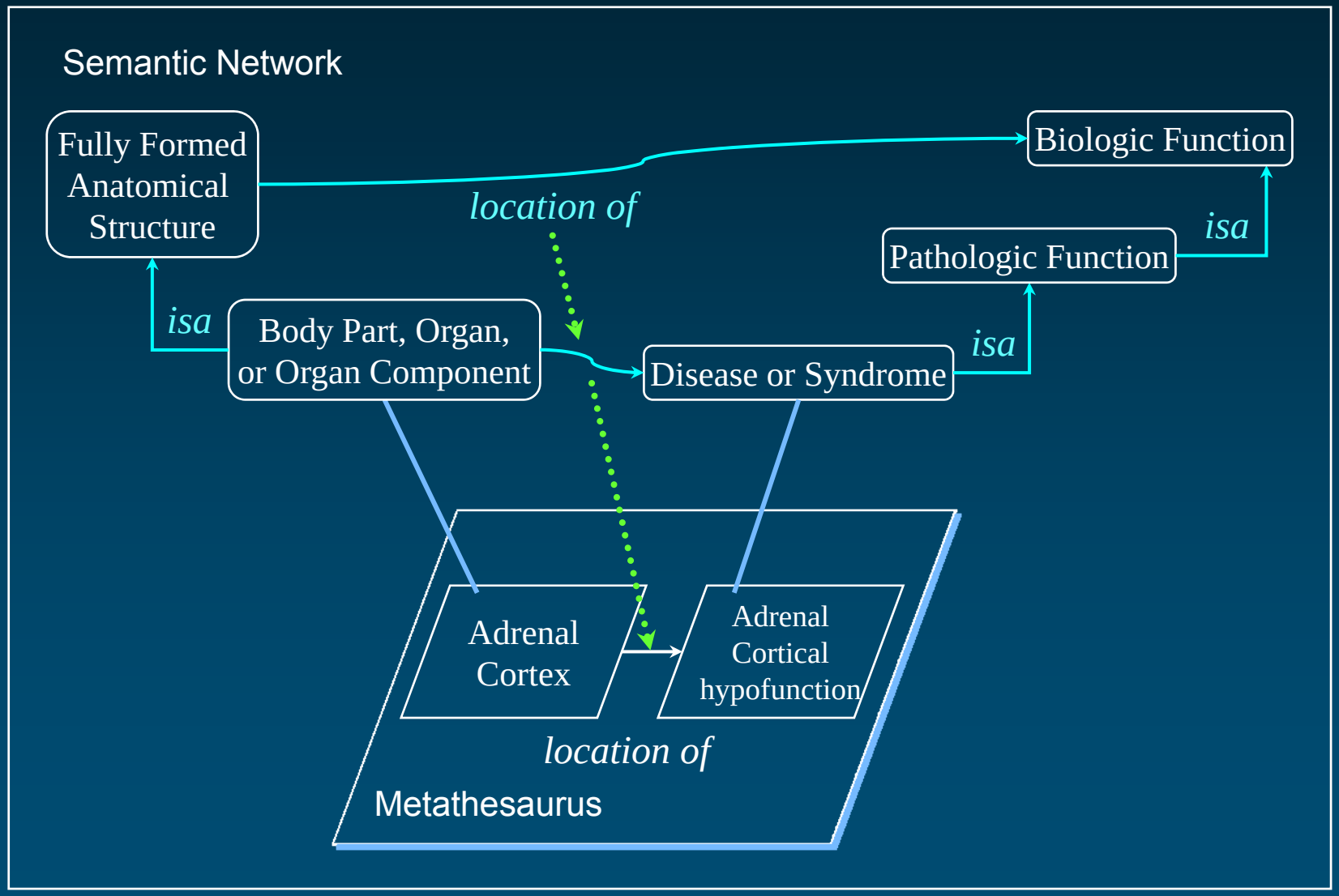


Why a semantic network?

- Semantic Types serve as high level categories assigned to Metathesaurus concepts, *independently of their position in a hierarchy*

- A relationship between 2 Semantic Types (ST) is a possible link between 2 concepts that have been assigned to those STs
  - The relationship may or may not hold at the concept level
  - Other relationships may apply at the concept level

Relationships can inherit semantics



UMLS and semantic integration

Data integration questions

Semantic interoperability through the UMLS

- Metathesaurus:
  Terminology/ontology integration
  - Terms from various terminologies linked through UMLS
- Semantic Network:
  Top domain ontology
  - Framework for semantic categorization of concepts
  - Template for potential relations among concepts

Potential contribution of UMLS to integration

- Data consistency
  - SN as a source of domain and range constraints for relations
- Data query
  - Resolve terms into concepts
  - Source of synonymy
  - [Lexical variants, normalization]
- Service query
- Service interoperability

Potential contribution of UMLS to integration

- Provenance
  - Rich source of metadata about terms
- Data integration
  - Map terms/concepts across vocabularies
  - Data integration through terminology integration
- Semantic mediation
  - UMLS as a the global schema

[Mougin, DILS 2008]

- Reasoning
  - Limited

Data, metadata and semantics

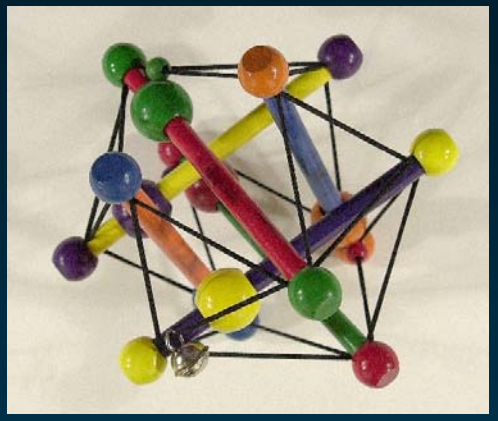
- Not specifically in UMLS
- caBIG
  - Cancer Biomedical Informatics Grid
    http://cabig.cancer.gov/
  - National Cancer Institute
  - Cancer Data Standards Registry and Repository (caDSR)
    http://ncicb.nci.nih.gov/NCICB/infrastructure/cacore_overview/cadsr
    - Common data elements
    - Metadata repository

Use of the Metathesaurus in applications

- Indexing, semantic annotation, coding
- Mapping across vocabularies
- Aggregation
- Support for Natural Language Processing applications (entity recognition)
- Source of value sets for information models

Use of the Semantic Network in applications

- Partition concepts into subdomains
  - Aggregation
- Support for Natural Language Processing applications (language understanding)
- Consistency checking of relations



Medical Ontology Research

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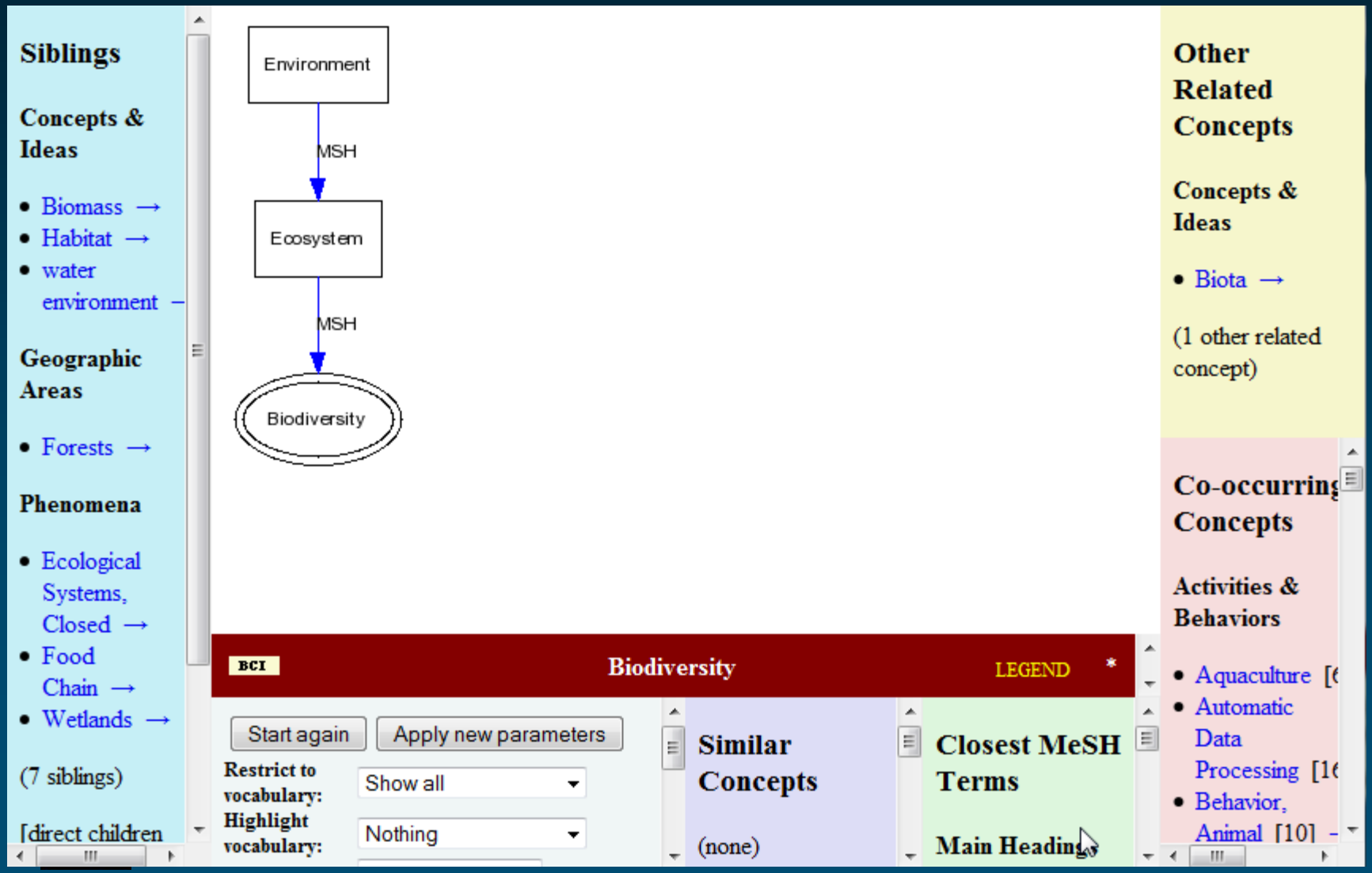
References

- 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

Biodiversity in the UMLS