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UMLS Webcasts series  
February 29, 2008



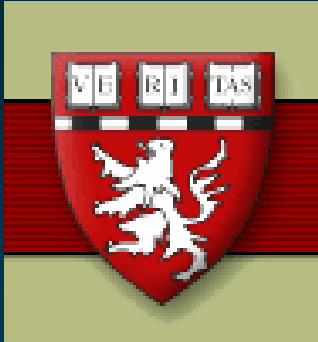
## UMLS Semantic Network



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# Acknowledgments



- ◆ Alexa T. McCray  
Harvard Medical School  
Boston, Massachusetts

# Outline

- ◆ The UMLS: A two-level structure
- ◆ UMLS Semantic Network
  - What is it?
    - Semantic types
    - Semantic relationships
    - Semantic relations
  - How to use it?
  - Where it get it from?
- ◆ Future directions



# Unified Medical Language System



## ◆ SPECIALIST Lexicon

- 360,000 lexical items
- Part of speech and variant information

## ◆ Metathesaurus

- 6M names from over 100 terminologies
- 1.5M concepts
- 8M relations

## ◆ Semantic Network

- 135 high-level categories
- 7000 relations among them

Lexical  
resources

Terminological  
resources

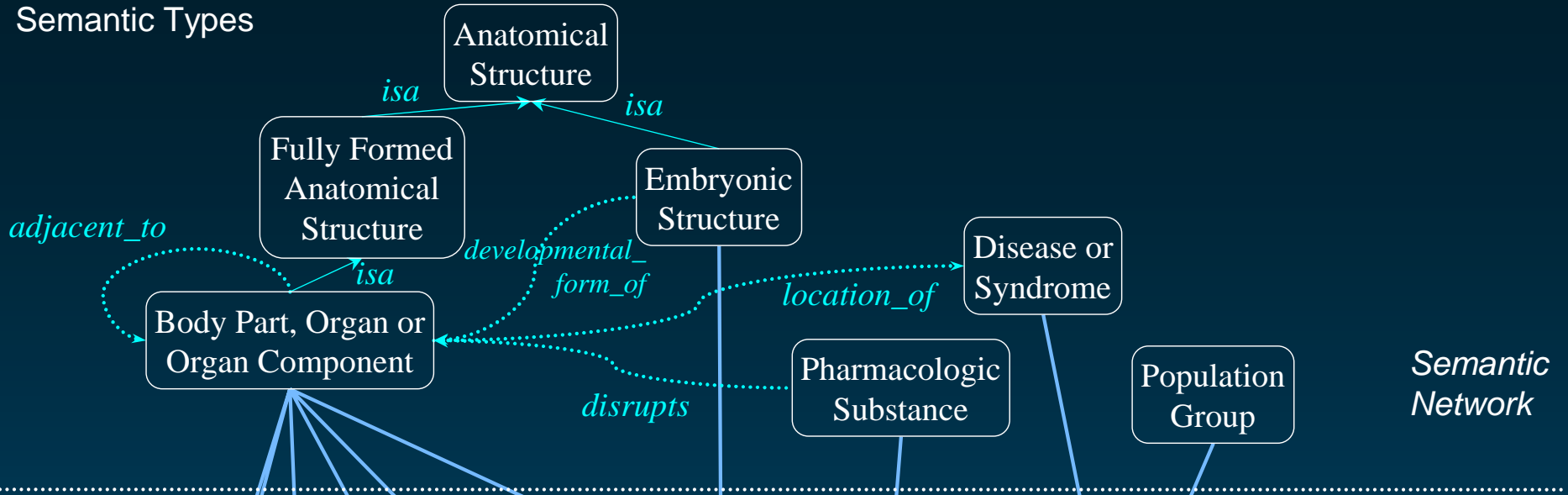
Ontological  
resources



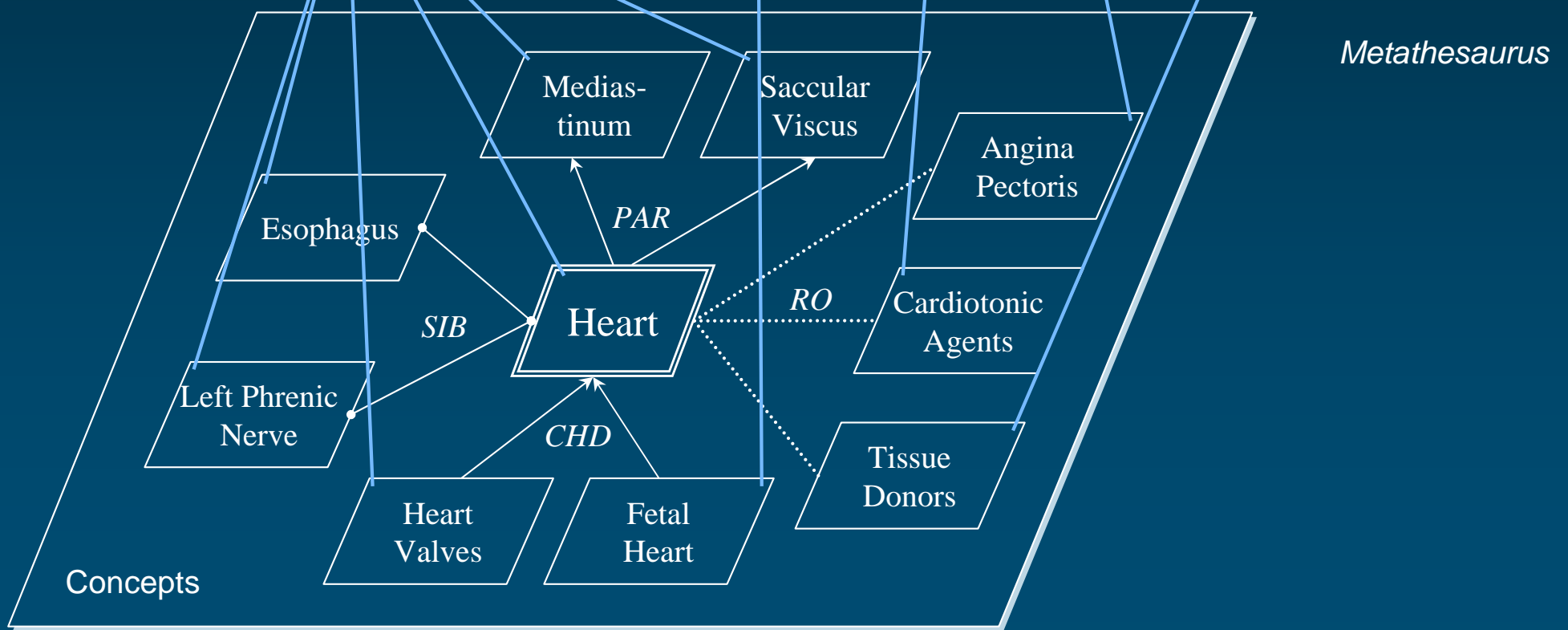
# The UMML

*A two-level structure*

Semantic Types

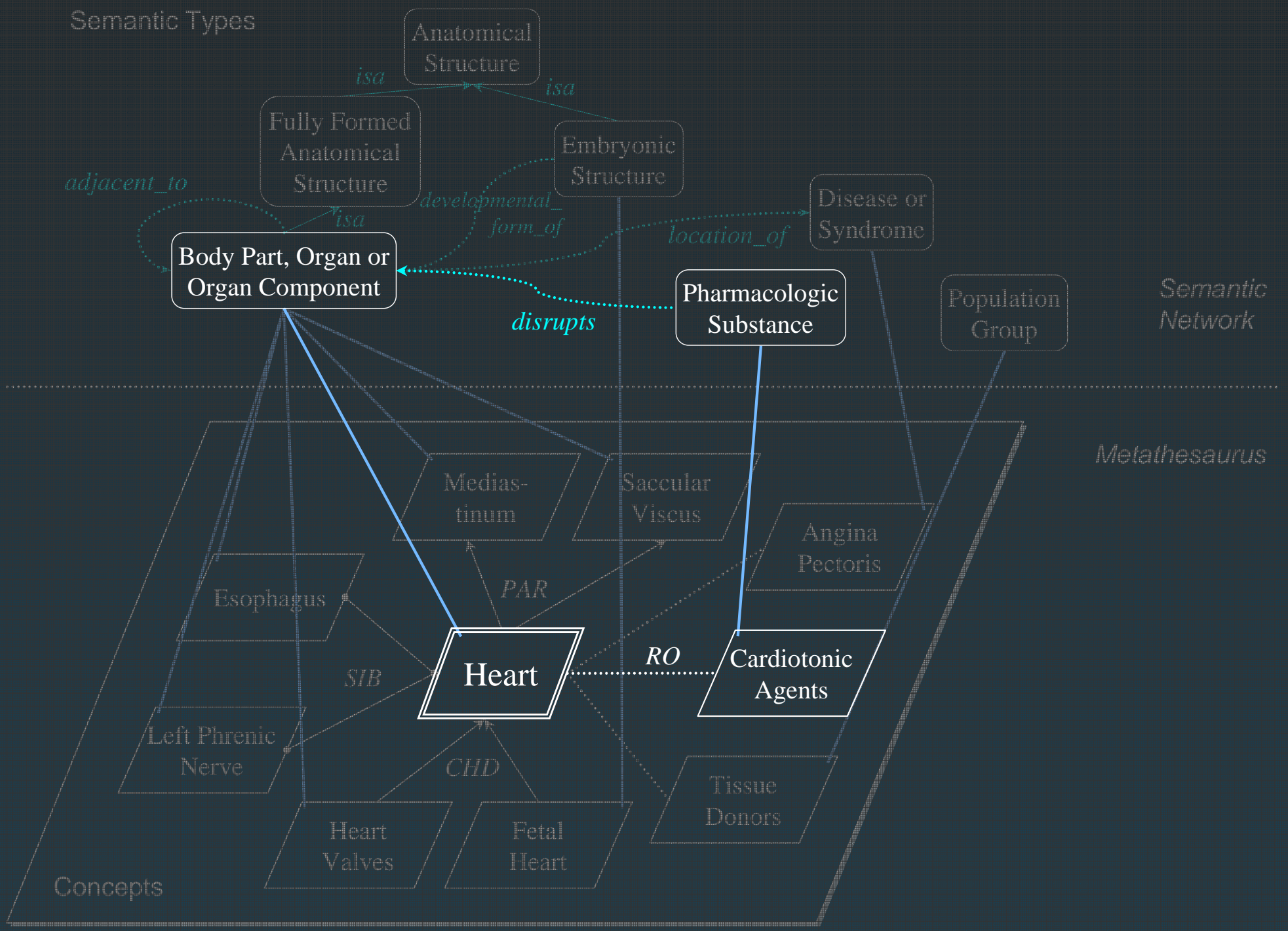


Semantic Network



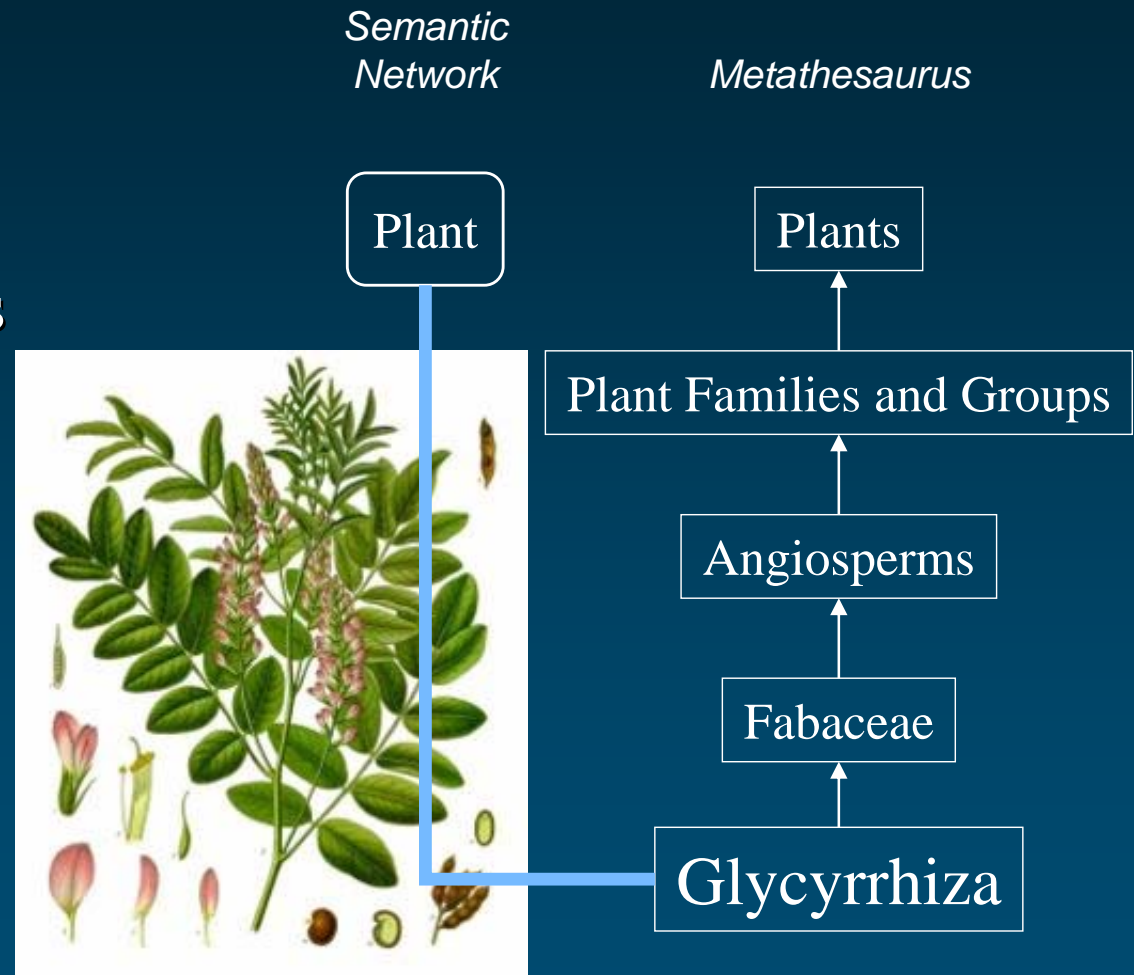
Metathesaurus

Concepts



# Concept categorization

- ◆ High-level categories (semantic types)
- ◆ Assigned by the Metathesaurus editors
- ◆ Independently of the hierarchies in which these concepts are located



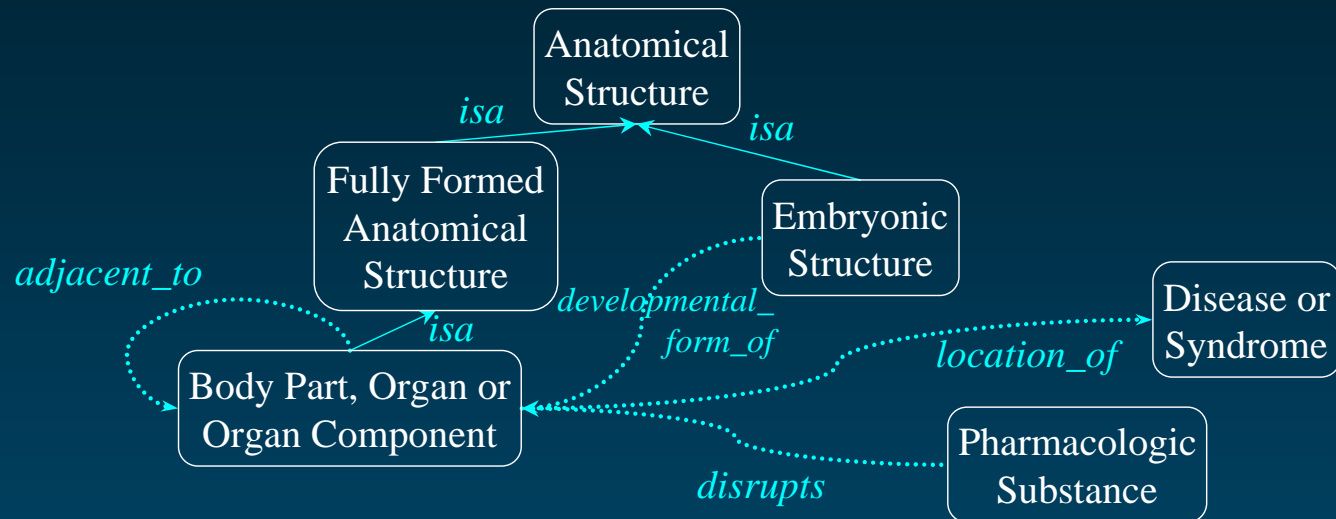
<http://en.wikipedia.org/wiki/Glycyrrhiza>



# UMLS Semantic Network

*What is it?*

# Semantic network



- ◆ *Semantic types*: Nodes
- ◆ *Semantic relationships*: Links
- ◆ *Semantic relations*: Triples (node<sub>1</sub>, link, node<sub>2</sub>)

# Semantic types

## ◆ Semantic types (135)

- tree structure
- 2 major hierarchies
  - Entity
    - Physical Object
    - Conceptual Entity
  - Event
    - Activity
    - Phenomenon or Process



- |  |   |   |
|--|---|---|
| Entity .....                                 |   | Entity [continued]                        |
| ■ Physical Object .....                      | ■ Physical Object [continued]                       | ■ Conceptual Entity                       |
| ■ ■ Organism                                 | ■ ■ Manufactured Object                             | ■ ■ Idea or Concept                       |
| ■ ■ ■ Plant                                  | ■ ■ ■ Medical Device                                | ■ ■ ■ Temporal Concept                    |
| ■ ■ ■ ■ Alga                                 | ■ ■ ■ Research Device                               | ■ ■ ■ Qualitative Concept                 |
| ■ ■ ■ ■ Fungus                               | ■ ■ ■ Clinical Drug                                 | ■ ■ ■ Quantitative Concept                |
| ■ ■ ■ ■ Virus                                | ■ ■ Substance                                       | ■ ■ ■ Functional Concept                  |
| ■ ■ ■ ■ Rickettsia or Chlamydia              | ■ ■ ■ Chemical                                      | ■ ■ ■ ■ Body System                       |
| ■ ■ ■ ■ Bacterium                            | ■ ■ ■ ■ Chemical Viewed Functionally                | ■ ■ ■ ■ Spatial Concept                   |
| ■ ■ ■ ■ Archaeon                             | ■ ■ ■ ■ ■ Pharmacologic Substance                   | ■ ■ ■ ■ Body Space or Junction            |
| ■ ■ ■ ■ Animal                               | ■ ■ ■ ■ ■ Antibiotic                                | ■ ■ ■ ■ Body Location or Region           |
| ■ ■ ■ ■ ■ Invertebrate                       | ■ ■ ■ ■ ■ Biomedical or Dental Material             | ■ ■ ■ ■ Molecular Sequence                |
| ■ ■ ■ ■ ■ Vertebrate                         | ■ ■ ■ ■ ■ Biologically Active Substance             | ■ ■ ■ ■ ■ Nucleotide Sequence             |
| ■ ■ ■ ■ ■ ■ Amphibian                        | ■ ■ ■ ■ ■ Neuroreactive Substance or Biogenic Amine | ■ ■ ■ ■ ■ Amino Acid Sequence             |
| ■ ■ ■ ■ ■ ■ Bird                             | ■ ■ ■ ■ ■ Hormone                                   | ■ ■ ■ ■ ■ Carbohydrate Sequence           |
| ■ ■ ■ ■ ■ ■ Fish                             | ■ ■ ■ ■ ■ Enzyme                                    | ■ ■ ■ ■ ■ Geographic Area                 |
| ■ ■ ■ ■ ■ ■ Reptile                          | ■ ■ ■ ■ ■ Vitamin                                   | ■ ■ Finding                               |
| ■ ■ ■ ■ ■ ■ Mammal                           | ■ ■ ■ ■ ■ Immunologic Factor                        | ■ ■ ■ Laboratory or Test Result           |
| ■ ■ ■ ■ ■ ■ Human                            | ■ ■ ■ ■ ■ Receptor                                  | ■ ■ ■ Sign or Symptom                     |
| ■ ■ Anatomical Structure                     | ■ ■ ■ ■ ■ Indicator, Reagent, or Diagnostic Aid     | ■ ■ Organism Attribute                    |
| ■ ■ ■ Embryonic Structure                    | ■ ■ ■ ■ ■ Hazardous or Poisonous Substance          | ■ ■ ■ Clinical Attribute                  |
| ■ ■ ■ Anatomical Abnormality                 | ■ ■ ■ ■ ■ Chemical Viewed Structurally              | ■ ■ Intellectual Product                  |
| ■ ■ ■ ■ Congenital Abnormality               | ■ ■ ■ ■ ■ Organic Chemical                          | ■ ■ ■ Classification                      |
| ■ ■ ■ ■ Acquired Abnormality                 | ■ ■ ■ ■ ■ Nucleic Acid, Nucleoside, or Nucleotide   | ■ ■ ■ Regulation or Law                   |
| ■ ■ ■ Fully Formed Anatomical Structure      | ■ ■ ■ ■ ■ Organophosphorus Compound                 | ■ ■ Language                              |
| ■ ■ ■ ■ Body Part, Organ, or Organ Component | ■ ■ ■ ■ ■ Amino Acid, Peptide, or Protein           | ■ ■ Occupation or Discipline              |
| ■ ■ ■ ■ Tissue                               | ■ ■ ■ ■ ■ Carbohydrate                              | ■ ■ ■ Biomedical Occupation or Discipline |
| ■ ■ ■ ■ Cell                                 | ■ ■ ■ ■ ■ Lipid                                     | ■ ■ Organization                          |
| ■ ■ ■ ■ Cell Component                       | ■ ■ ■ ■ ■ Steroid                                   | ■ ■ ■ Health Care Related Organization    |
| ■ ■ ■ ■ Gene or Genome                       | ■ ■ ■ ■ ■ Eicosanoid                                | ■ ■ ■ Professional Society                |
|  | ■ ■ ■ ■ ■ Inorganic Chemical                        | ■ ■ ■ Self-help or Relief Organization    |
|  | ■ ■ ■ ■ ■ Element, Ion, or Isotope                  | ■ ■ Group Attribute                       |
|  | ■ ■ ■ Body Substance                                | ■ ■ Group                                 |
|  | ■ ■ ■ Food  | ■ ■ ■ Professional or Occupational Group  |
|  |   | ■ ■ ■ Population Group                    |
|  |   | ■ ■ ■ Family Group                        |
|  |   | ■ ■ ■ Age Group                           |
|  |   | ■ ■ ■ Patient or Disabled Group           |

# Hierarchical structure Entity

Event .....

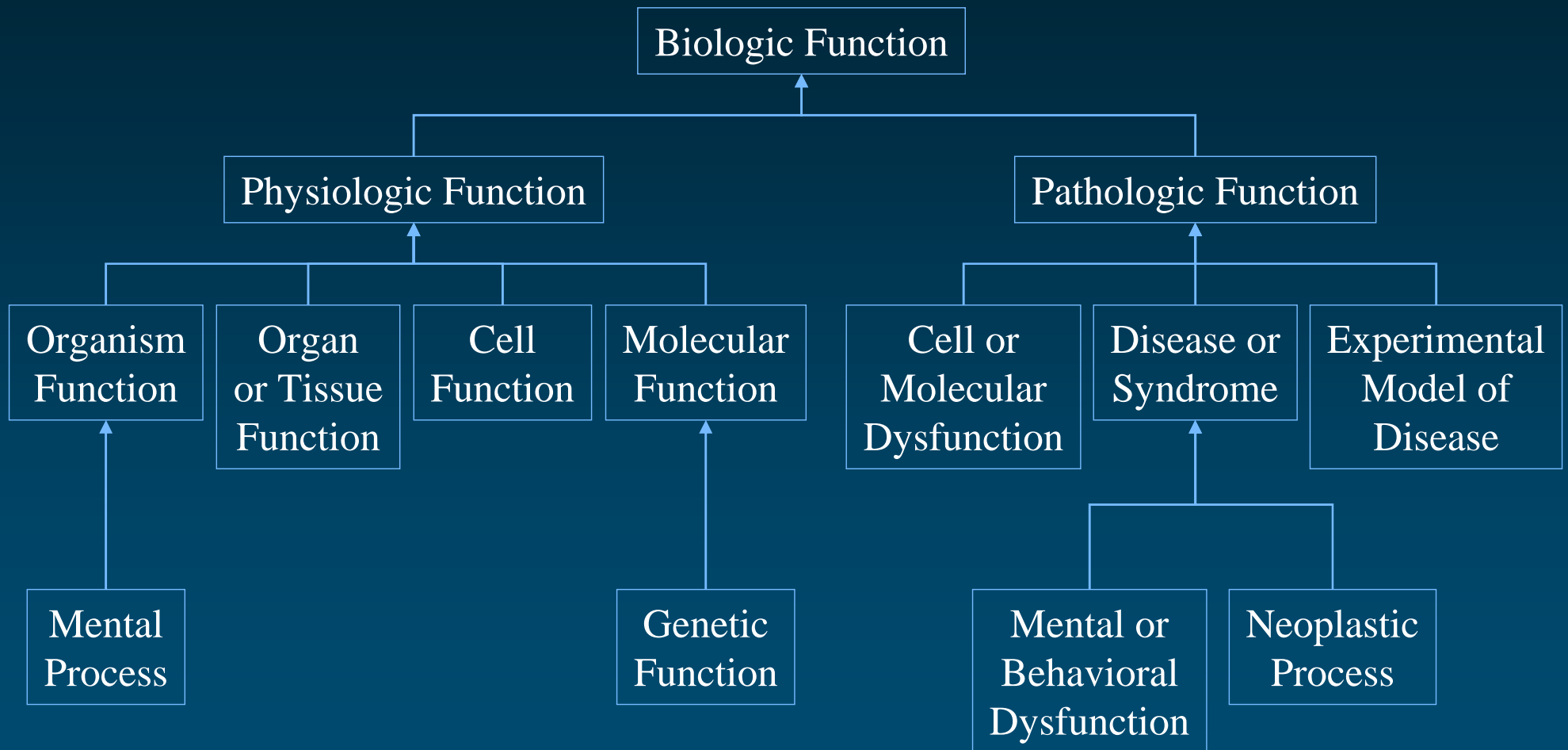
- Activity
  - ■ Behavior
    - ■ ■ Social Behavior
    - ■ ■ Individual Behavior
  - ■ Daily or Recreational Activity
  - ■ Occupational Activity
    - ■ ■ Health Care Activity
      - ■ ■ ■ Laboratory Procedure
      - ■ ■ ■ Diagnostic Procedure
      - ■ ■ ■ Therapeutic or Preventive Procedure
    - ■ ■ Research Activity
      - ■ ■ ■ Molecular Biology Research Technique
    - ■ ■ Governmental or Regulatory Activity
    - ■ ■ Educational Activity
  - ■ Machine Activity

Event [continued]

- Phenomenon or Process
  - ■ Human-caused Phenomenon or Process
    - ■ ■ Environmental Effect of Humans
  - ■ Natural Phenomenon or Process
    - ■ ■ Biologic Function
      - ■ ■ ■ Physiologic Function
        - ■ ■ ■ ■ Organism Function
          - ■ ■ ■ ■ ■ Mental Process
          - ■ ■ ■ ■ Organ or Tissue Function
          - ■ ■ ■ ■ Cell Function
          - ■ ■ ■ ■ Molecular Function
            - ■ ■ ■ ■ ■ Genetic Function
            - ■ ■ ■ ■ Pathologic Function
              - ■ ■ ■ ■ ■ Disease or Syndrome
                - ■ ■ ■ ■ ■ ■ Mental or Behavioral Dysfunction
                - ■ ■ ■ ■ ■ Neoplastic Process
                - ■ ■ ■ ■ Cell or Molecular Dysfunction
                - ■ ■ ■ ■ Experimental Model of Disease
  - ■ Injury or Poisoning

Hierarchical structure Event

# “Biologic Function” hierarchy (isa)



# Semantic type information

- ID** ● T047
- Name** ● Disease or Syndrome
- Tree number** ● B2.2.1.2.1
- Definition** ● *A condition which alters or interferes with a normal process, state, or activity of an organism. It is usually characterized by the abnormal functioning of one or more of the host's systems, parts, or organs. Included here is a complex of symptoms descriptive of a disorder.*
- Examples** ● Diabetes Mellitus; Dumping Syndrome; Malabsorption Syndromes; Nephrotic Syndrome
- Usage note** ● Any specific disease or syndrome that is modified by such modifiers as "acute", "prolonged", etc. will also be assigned to this type. If an anatomic abnormality has a pathologic manifestation, then it will be given this type as well as a type from the 'Anatomical Abnormality' hierarchy, e.g., "Diabetic Cataract" will be double-typed for this reason.
- Abbreviation** ● dsyn



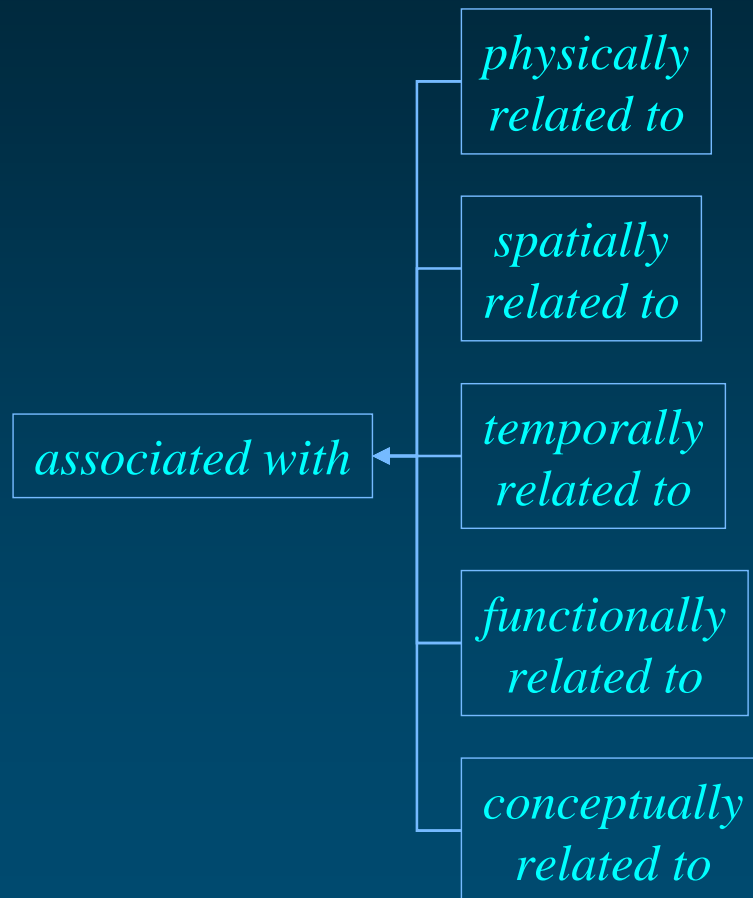
# Semantic relationships

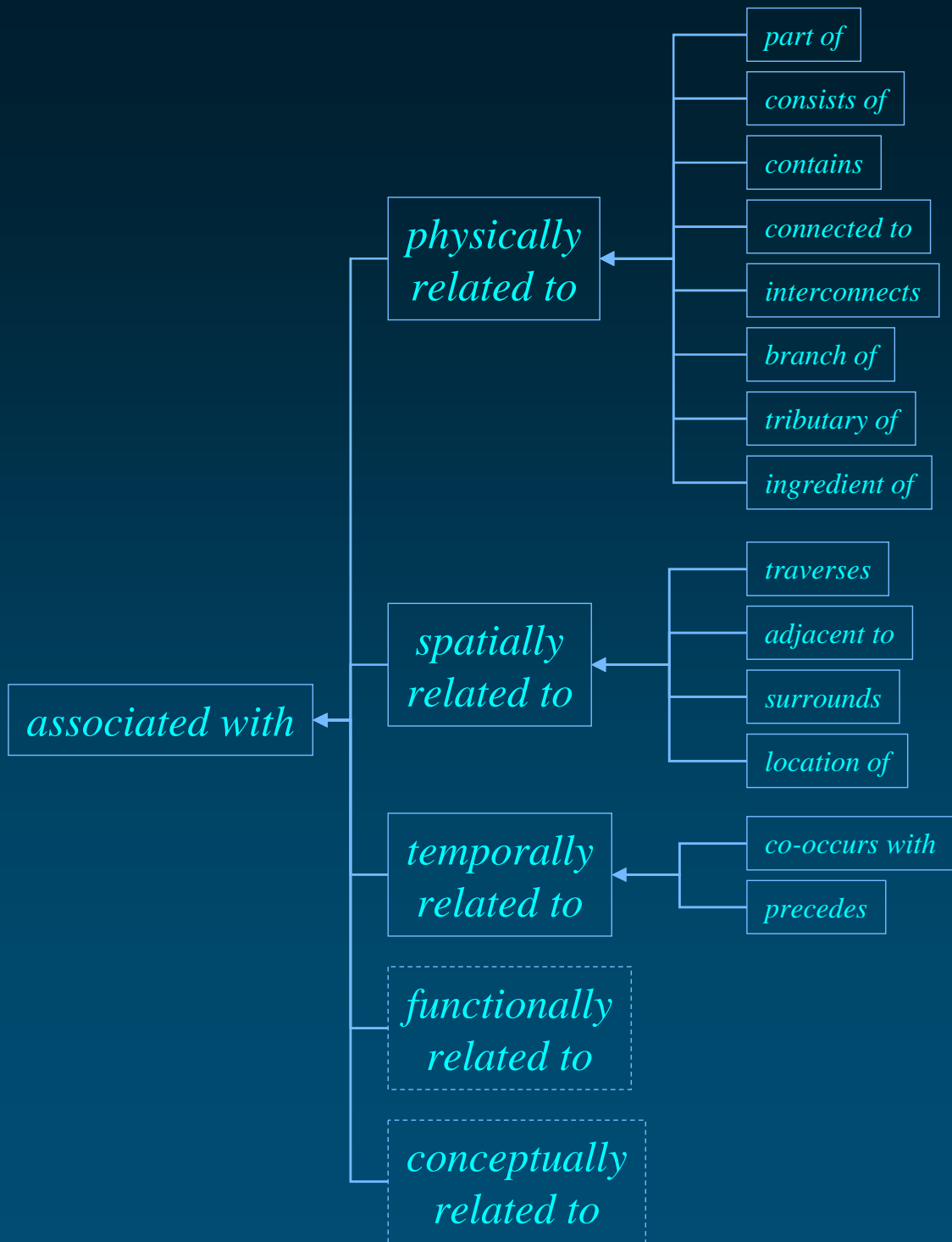
- ◆ Semantic network relationships (54)
  - Hierarchical (*isa* = is a kind of)
    - among types
      - *Animal isa Organism*
      - *Enzyme isa Biologically Active Substance*
    - among relations
      - *treats isa affects*
  - Associative (non-hierarchical)
    - *Sign or Symptom diagnoses Pathologic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

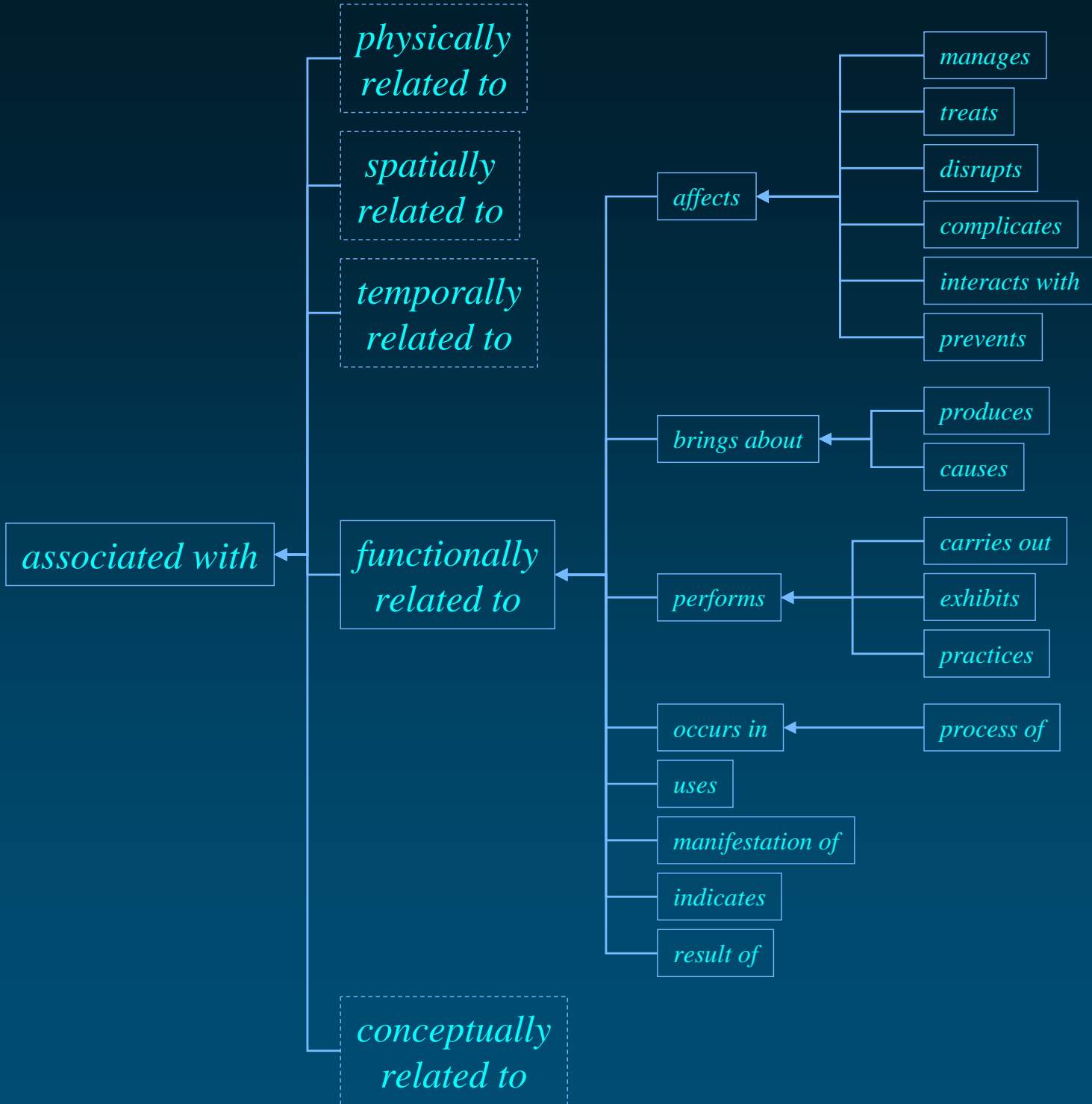


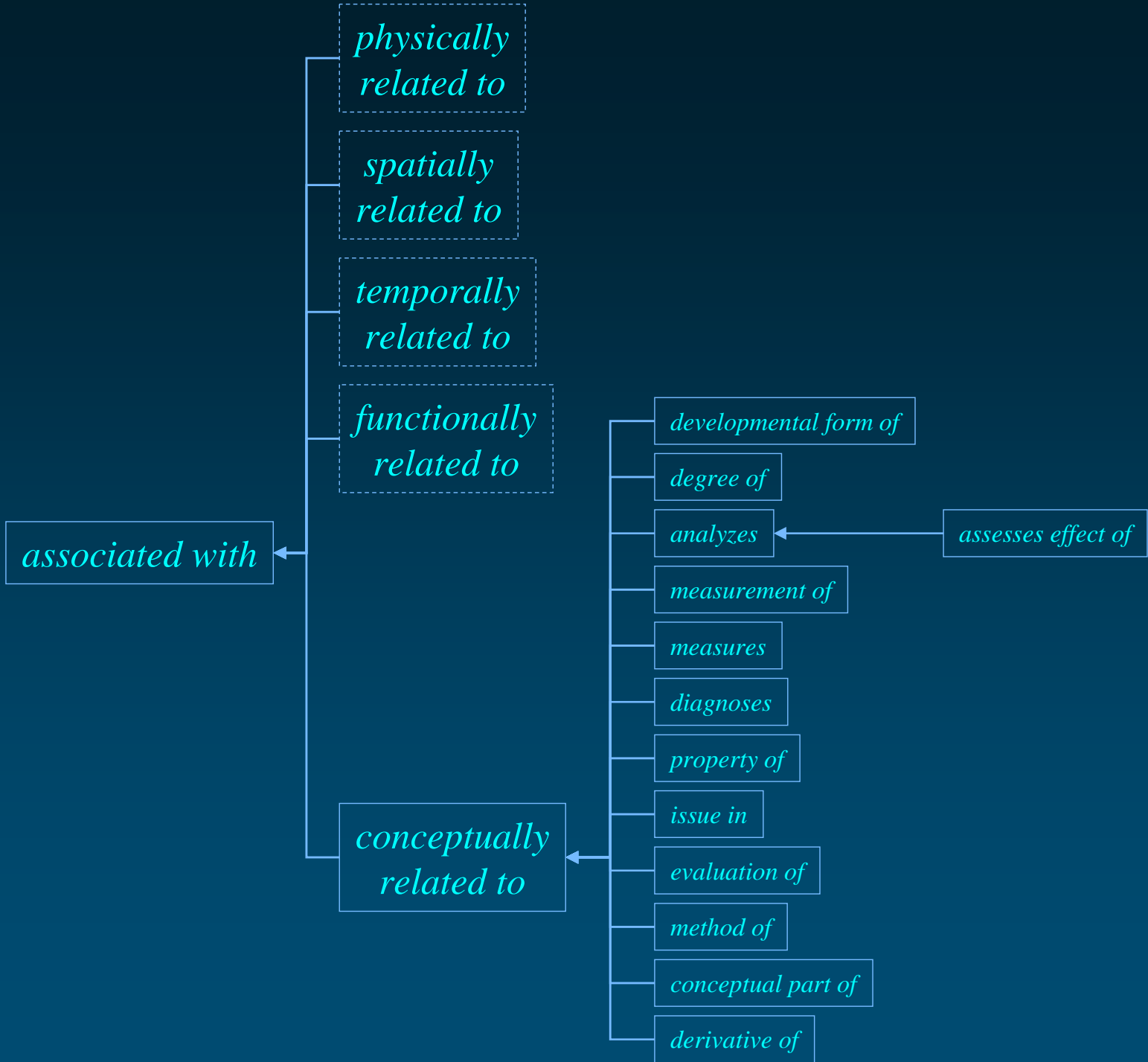


# Associative relationship hierarchy









# Semantic relationship information

ID	• T202
Name	• ingredient_of
Tree number	• R1.8
Definition	• <i>Is a component of, as in a constituent of a preparation.</i>
Abbreviation	• IG
Inverse rel.	• has_ingredient



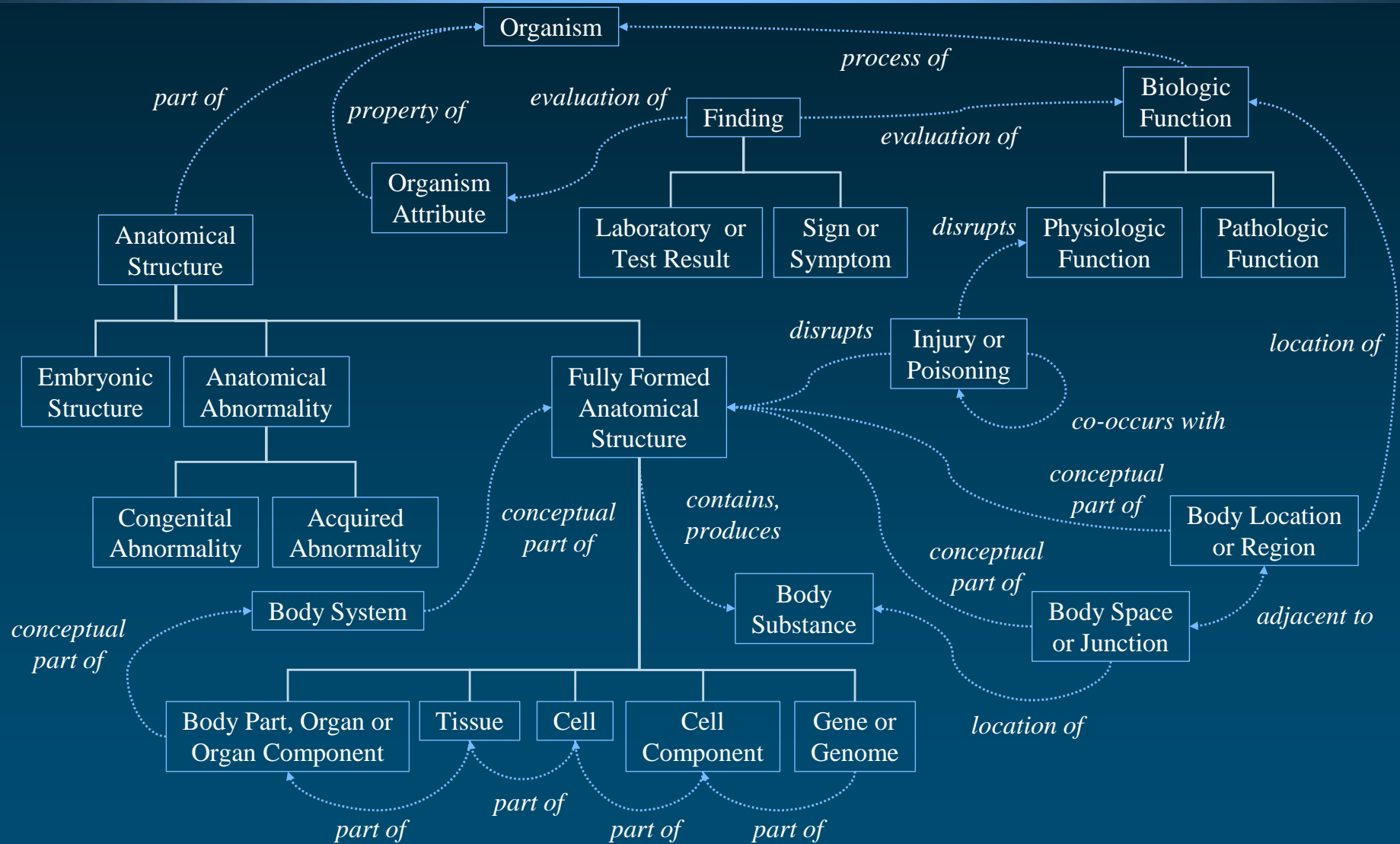
# Semantic relations

## ◆ 612 assertions

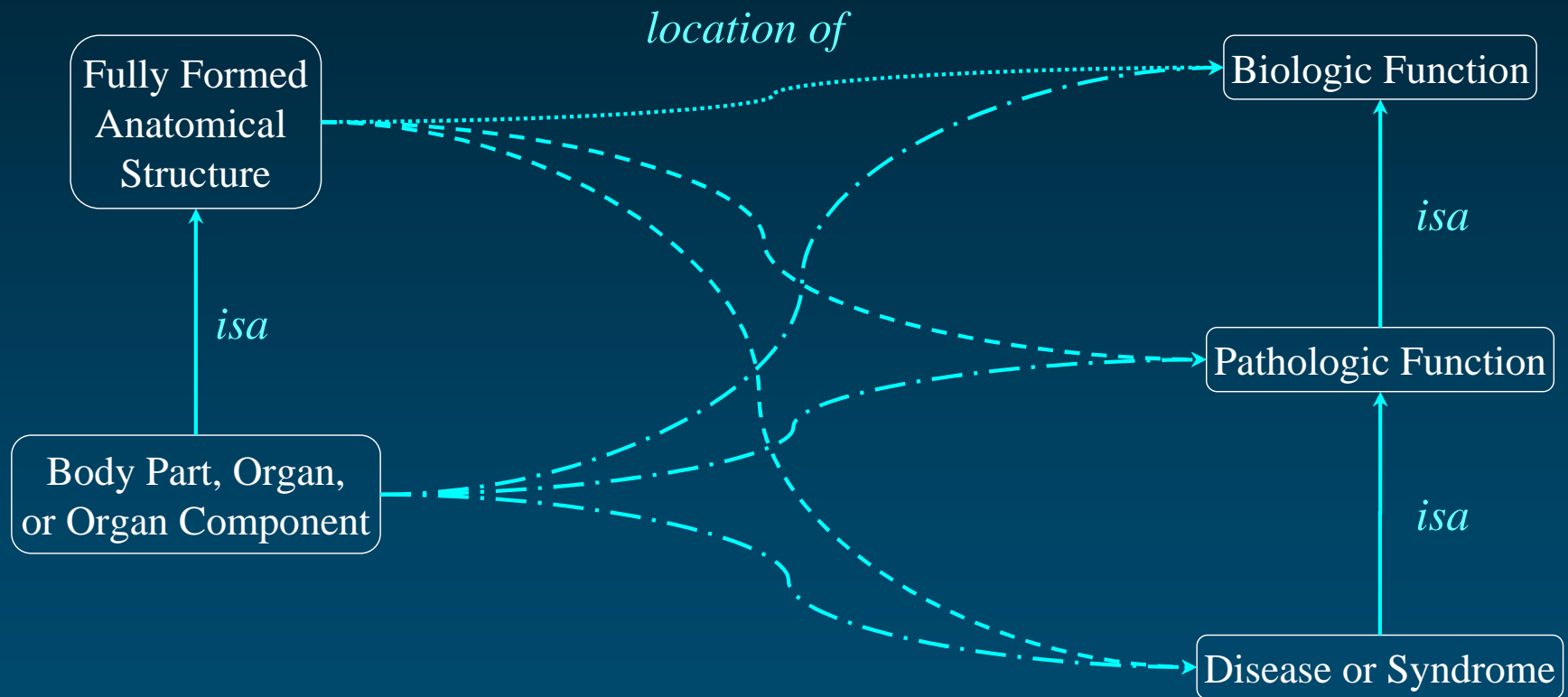
- 558 (type<sub>1</sub>, relationship, type<sub>2</sub>)
  - Hierarchical
    - *Animal* *isa* *Organism*
    - *Enzyme* *isa* *Biologically Active Substance*
  - Associative
    - *Sign or Symptom* *diagnoses* *Pathologic Function*
    - *Pharmacologic Substance* *treats* *Pathologic Function*
- 54 (relationship<sub>1</sub>, *isa*, relationship<sub>2</sub>)
  - *treats* *isa* *affects*



# Semantic relations (subset)



# Inheritance





# Semantic relations (inherited)

## ◆ 6864 assertions

- 6752 ( $\text{type}_1$ , relationship,  $\text{type}_2$ )
  - Hierarchical
    - **Mammal** *isa* **Organism** (from: *Animal isa Organism*)
    - **Enzyme** *isa* **Chemical**  
(from: *Enzyme isa Biologically Active Substance*)
  - Associative
    - **Body Part, Organ, or Organ Component**  
*location\_of* **Disease or Syndrome**  
(from: *Fully Formed Anatomical Structure*  
*location\_of* *Biologic Function*)
- 112 ( $\text{relationship}_1$ , *isa*,  $\text{relationship}_2$ )
  - *treats isa functionally\_related\_to* (from: *treats isa affects*)



# Semantic relation information

- |                        |                                     |
|------------------------|-------------------------------------|
| Sem. Type <sub>1</sub> | • Fully Formed Anatomical Structure |
| Relationship           | • location_of                       |
| Sem. Type <sub>2</sub> | • Biologic Function                 |
| Link status            | • D                                 |



# UMLS Semantic Network

*How to use it?*

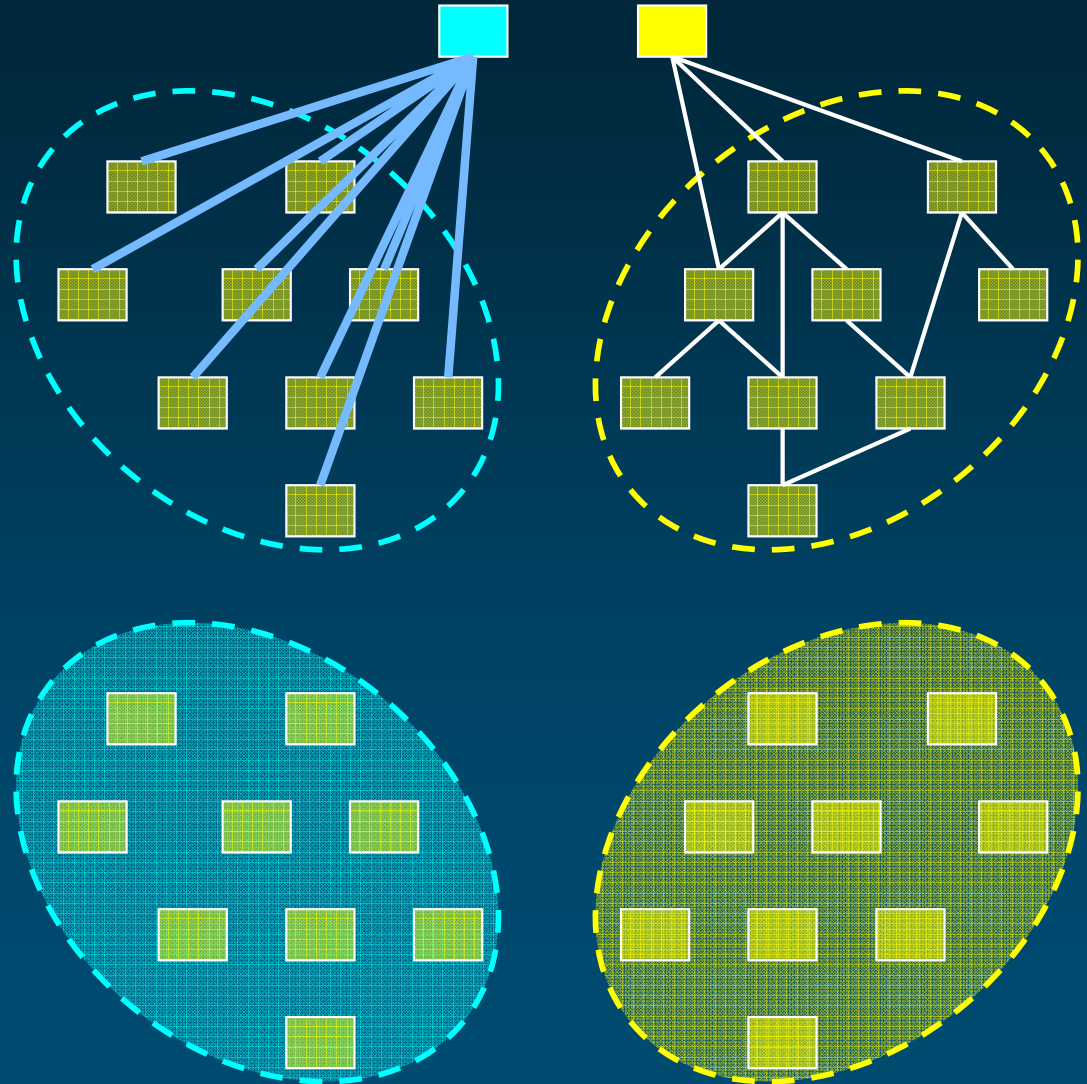
# Why a semantic network?

- ◆ Semantic Types serve as high level categories assigned to Metathesaurus concepts, *independently of their position in a hierarchy*
- ◆ A relation between 2 Semantic Types (ST) indicates 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

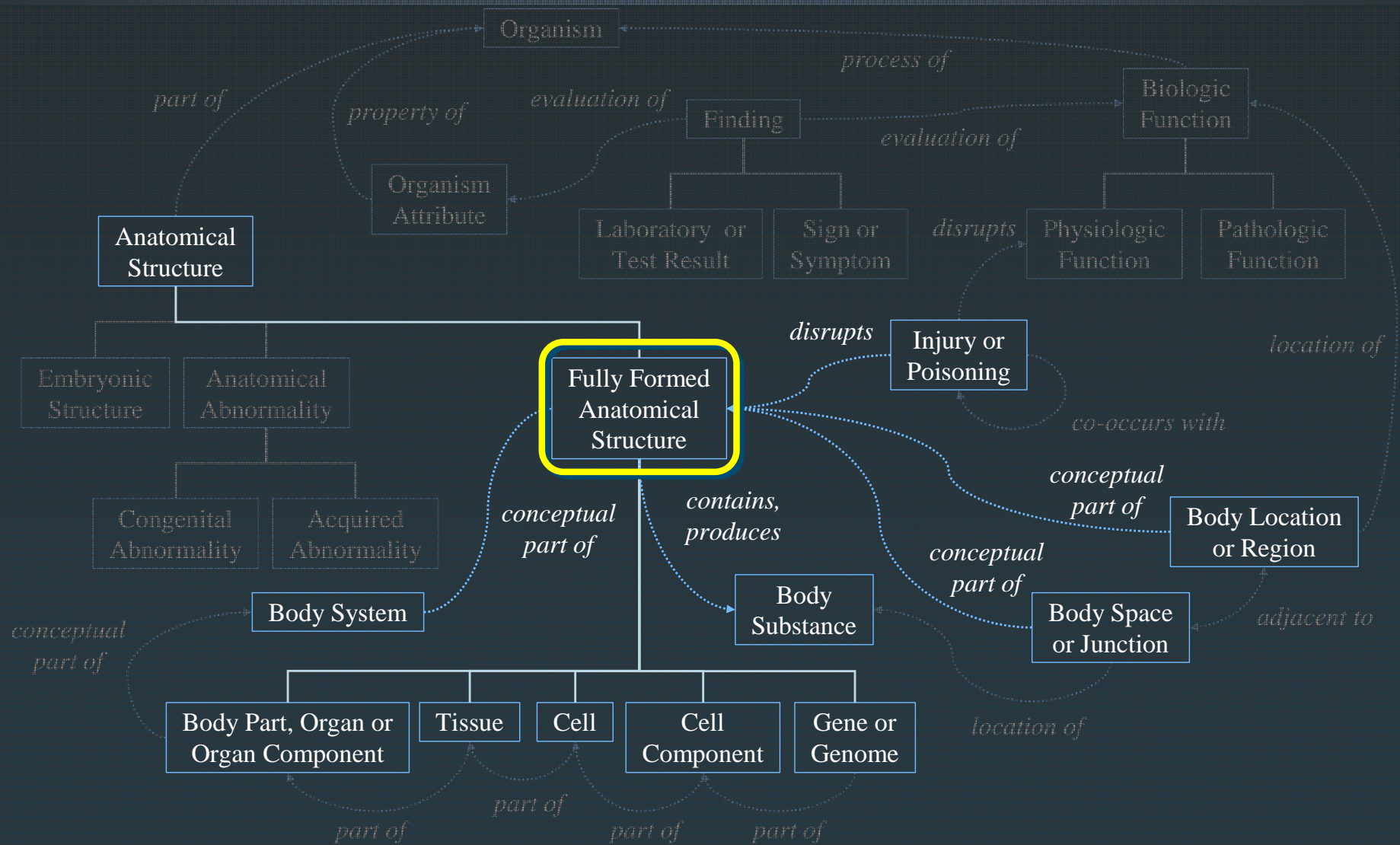


# Identify concepts for a given category

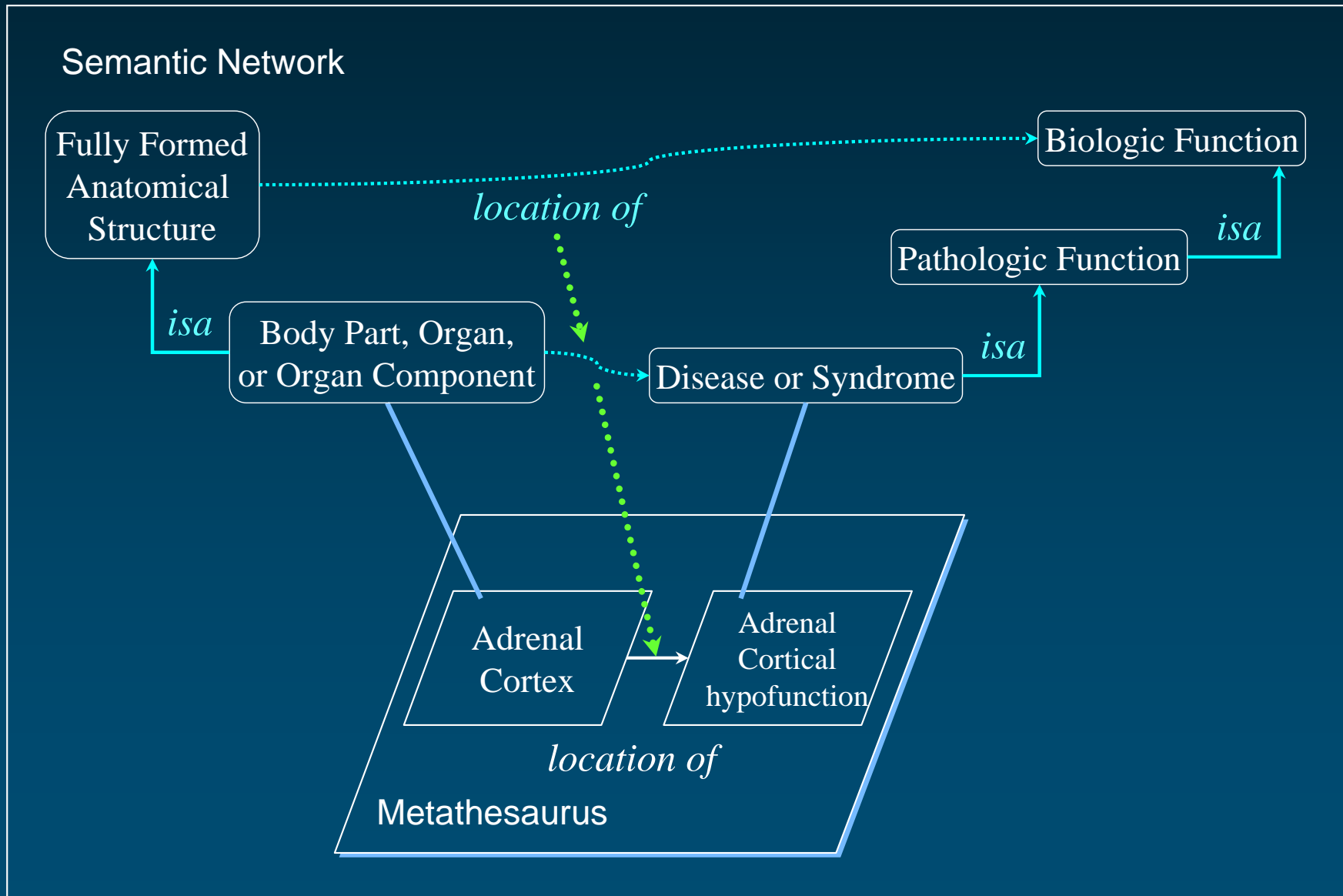
- ◆ Semantic type
  - List of all concepts having this semantic type
- ◆ Concept
  - List of all descendants



# Source of domain knowledge



# Semantics of Metathesaurus relations



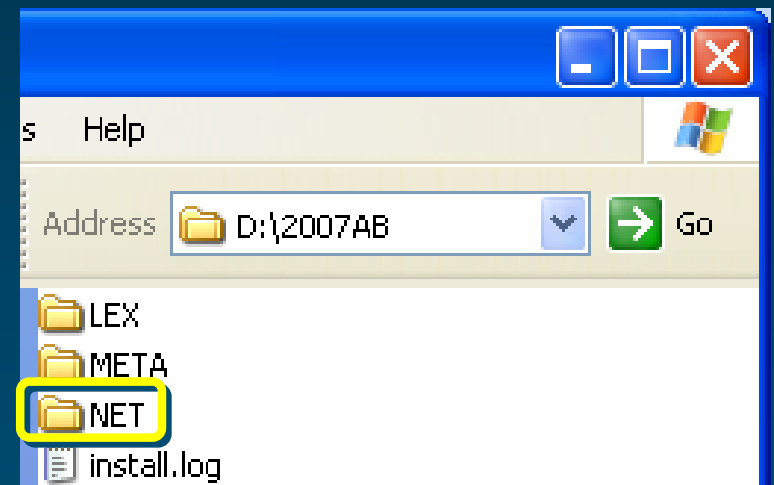
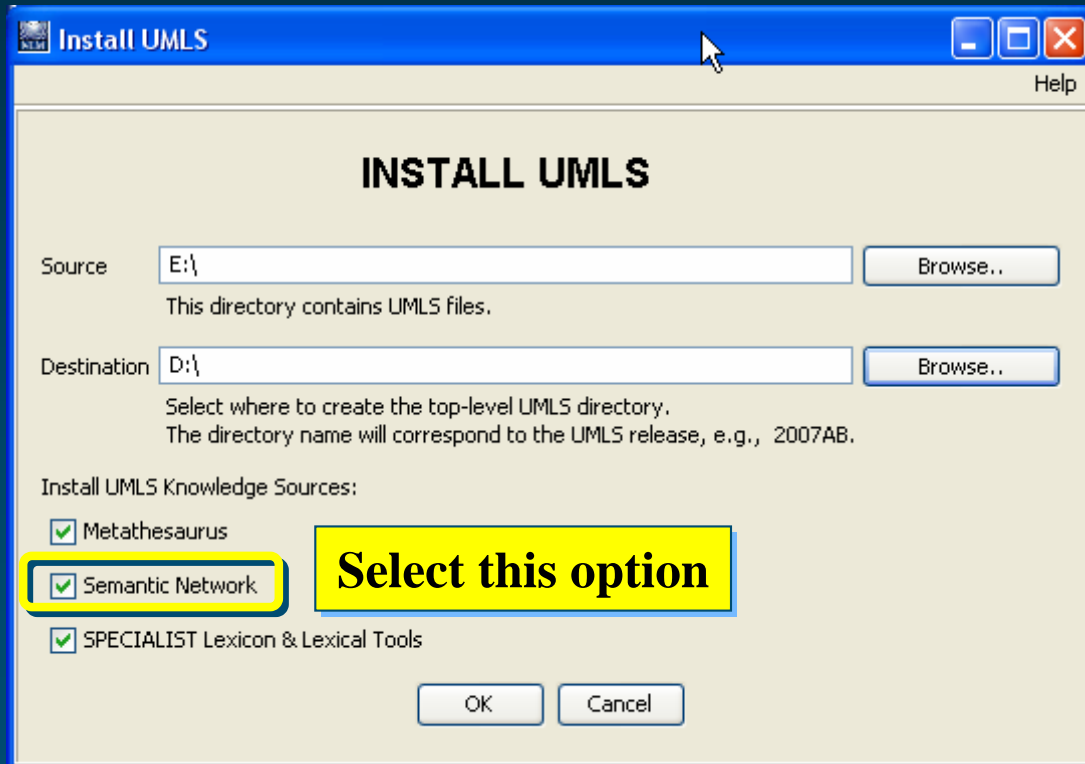
# UMLS Semantic Network

*Where to get it from?*



# UMLS distribution

- ◆ Part of the UMLS distribution
  - Installed by MetamorphoSys (option)



# UMLS Semantic Network website

- ◆ No intellectual property restrictions on the Semantic Network
- ◆ Available outside UMLS license agreement
- ◆ UMLS Semantic Network website  
<http://semanticnetwork.nlm.nih.gov/>



# UMLS Semantic Network website

## The UMLS Semantic Network



The UMLS Semantic Network is one of three UMLS Knowledge Sources developed as part of the Unified Medical Language System project. The network provides a consistent categorization of all concepts represented in the UMLS Metathesaurus.

The UMLS Semantic Network is available in unit record and relational format and is distributed subject to these [terms and conditions](#).

- [Download the UMLS Semantic Network files](#)

The UMLS semantic network reduces the complexity of the Metathesaurus by grouping concepts according to the semantic types that have been assigned to them. For certain purposes, however, an even smaller and coarser-grained set of semantic type groupings may be desirable. [The UMLS semantic groups](#) have been established for this purpose.

Further information about the UMLS Semantic Network can be found at:

- [UMLS Semantic Network Fact Sheet](#)
- [UMLS Semantic Network Documentation](#)

McCray AT. [An upper level ontology for the biomedical domain](#). *Comp Funct Genom* 2003; 4:80-4.

Updated: February 1, 2006

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# Semantic Network files

## The UMLS Semantic Network Files



The UMLS Semantic Network files are distributed subject to these [terms and conditions](#).

Download the current Semantic Network files:

- [The SRFIL file](#) (File Description)
- [The SRFLD file](#) (Field Description)
- [The SRDEF file](#) (Basic information about the Semantic Types and Relations)
- [The SRSTR file](#) (Structure of the Network)
- [The SRSTRE1 file](#) (Fully inherited set of Relations (UIs))
- [The SRSTRE2 file](#) (Fully inherited set of Relations (Names))
  
- [The SU file](#) (Unit Record)

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## Unified Medical Language System

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<http://www.nlm.nih.gov/research/umls/meta3.html>

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### Section 3 SEMANTIC NETWORK

#### 3.0 Introduction

The Semantic Network consists of (1) a set of broad subject categories, or **Semantic Types**, that provide a consistent categorization of all concepts represented in the UMLS Metathesaurus, and (2) a set of useful and important relationships, or **Semantic Relations**, that exist between Semantic Types. This section of the documentation provides an overview of the Semantic Network, and describes the files of the Semantic Network. Sample records illustrate structure and content of these files.

#### 3.1 Overview

The purpose of the Semantic Network is to provide a consistent categorization of all concepts represented in the UMLS Metathesaurus and to provide a set of useful relationships between these concepts. All information about specific concepts is found in the Metathesaurus. The Network provides information about the set of basic semantic types, or categories, which may be assigned to these concepts, and it defines the set of relationships that may hold between the semantic types. The Semantic Network contains 135 semantic types and 54 relationships. The Semantic Network serves as an authority for the semantic types that are assigned to concepts in the Metathesaurus. The Network defines these types, both with textual descriptions and by means of the information inherent in its hierarchies.

The semantic types are the nodes in the Network, and the relationships between them are the links. There are major groupings of semantic types for organisms, anatomical structures, biologic function, chemicals, events, physical objects, and concepts or ideas. The current scope of the UMLS semantic types is quite broad, allowing for the semantic categorization of a wide range of terminology in multiple domains.



# UMLS Semantic Network

*Future directions*

# Future directions

- ◆ Harmonize with other top-level ontologies of biomedicine
  - Semantic types (e.g., with BioTop)
  - Semantic relationships (e.g., with the OBO Relation Ontology, with relationships in GALEN)



# BioTop

<http://www.ifomis.org/biotop>

## BioTop

A Biomolecular Top-Domain Ontology

University Medical Center Freiburg - Medical Informatics Research Group  
Institute for Formal Ontology and Medical Information Science (IFOMIS)  
Jena University Language & Information Engineering (JULIE) Lab

[Overview](#) | [Implementation](#) | [Users and Examples](#) | [Publications](#) | [Feedback](#)

### Overview

The increasing need for advanced ontology-based knowledge management in the life sciences and the description of its basic ontological entities is generally being acknowledged but, up until now, the development of biological ontologies lacks adherence to foundational principles of ontology design.

In light of this we introduce *BioTop*, a description-logic-based upper ontology of the most important entities in the domain of molecular biology, founded upon the formal principles mentioned above.

*BioTop* was originally based on [GENIA](#), an ontology applied for corpus annotation in text mining contexts. In contrast to this, a major desideratum for *BioTop* is to describe as many classes as possible in terms of necessary and sufficient conditions.

To further ensure ontological validity we employ [BFO](#) as the top layer for the actual *BioTop* classes.

We use [OWL-DL](#) as a formal knowledge representation language and can thus use a terminological reasoner for classification in order to maintain consistency during the ontology engineering and also to automatically infer its full multihierarchical structure.

### Users and Examples

- ◆ [BOOTStrep](#): Bootstrapping Of Ontologies and Terminologies Strategic REsearch Project
- ◆ [UMLS-Mapping](#): A mapping of the Universal Medical Language System (UMLS) Semantic Network (SN) to *BioTop*
- ◆ [taxdemo](#): A *small* and *demonstrative* ontology of how biological taxa can be based on *BioTop* and described in the (submitted) ISMB paper (You can download the implementation or get either a [HTML view](#) or a [graphical view](#) of it.)





# OBO relations

Summary Table

name	transitive	symmetric	reflexive	anti-symmetric	documentation
is_a	+		+	+	<a href="#">View detailed summary</a>
part_of	+		+	+	<a href="#">View detailed summary</a>
integral_part_of	+		+	+	<a href="#">View detailed summary</a>
proper_part_of	+				<a href="#">View detailed summary</a>
located_in	+		+		<a href="#">View detailed summary</a>
contained_in					<a href="#">View detailed summary</a>
adjacent_to					<a href="#">View detailed summary</a>
transformation_of	+				<a href="#">View detailed summary</a>
derives_from	+				<a href="#">View detailed summary</a>
preceded_by	+				<a href="#">View detailed summary</a>
has_participant					<a href="#">View detailed summary</a>
has_agent					<a href="#">View detailed summary</a>
instance_of					<a href="#">View detailed summary</a>

<http://www.obofoundry.org/ro/>



# GALEN relations *isIngredientOf*

<http://opengalen.org>

- + topLinkClass
- Attribute
  - DomainAttribute
    - ConstructiveAttribute
      - InversePartitiveAttribute
        - InverseStructuralPartitiveAttribute
          - + IsDivisionOf
          - makesUp
            - x specificallyMakesUp
            - isIngredientOf
            - x isSpecificIngredientOf
            - x isMultipleIngredientOf
            - x isNotIngredientOf
            - x isNotChemicalSubgroupOf
      - + inverseProcessPartitiveAttribute
    - + InverseStructuralAttribute
    - + involves
    - + FunctionalAttribute
    - x hasReference
    - x hasFrameOfReference
    - + hasPhysicalMeans
    - + hasPersonPerforming
    - x records
    - x hasEmployer
  - + WrapperAttribute
  - + TemporalAttribute
  - + ModifierAttribute
  - + SpecificationLevelAttribute
  - + ApplicationAttribute

Name:	isIngredientOf
Inverse:	hasIngredient
Cardinality:	manyMany
Properties:	inverse, transitive
Transits:	makesUp
Transitive orbits:	makesUp

Attribute    Pretty: on    Filter: on

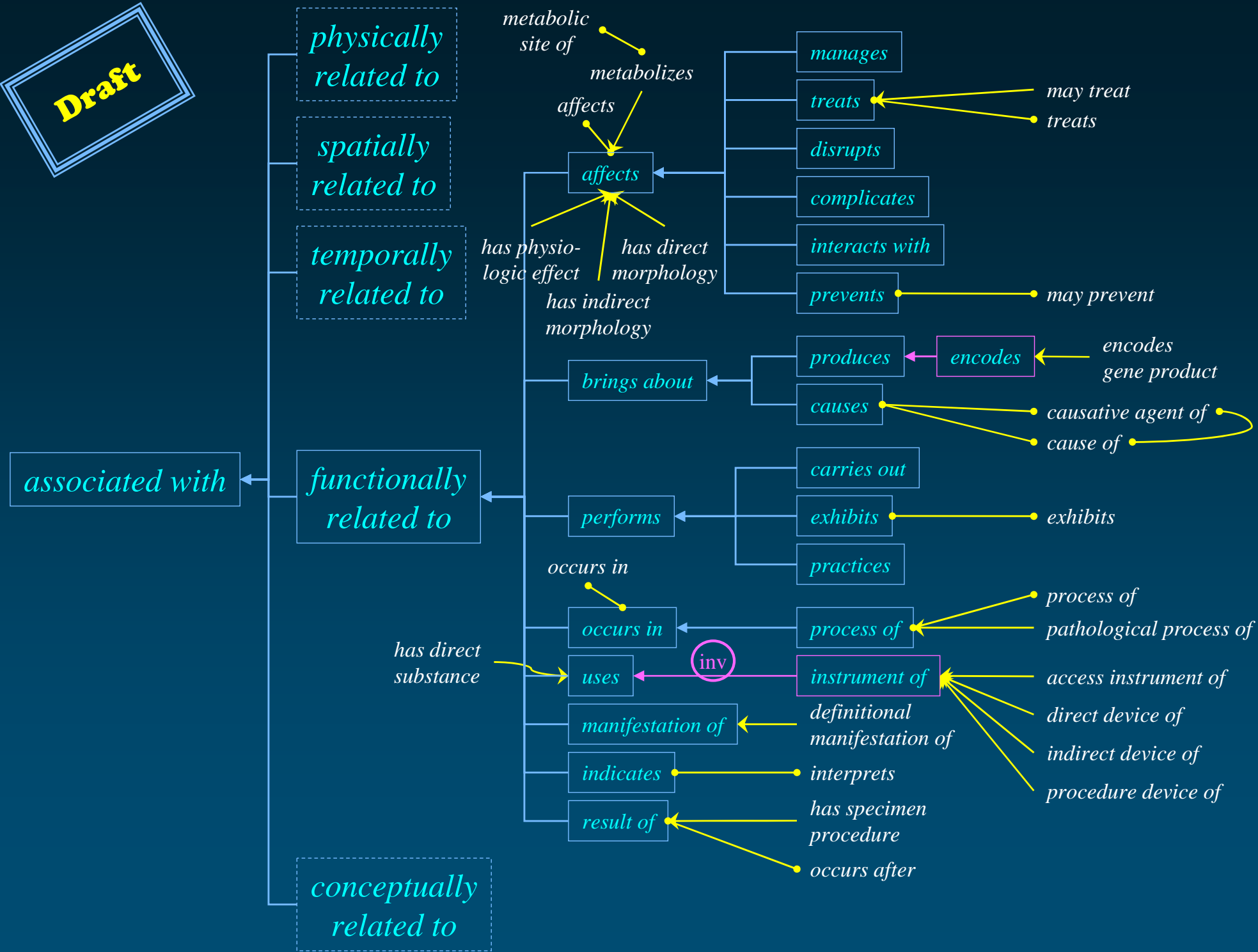


# Future directions

- ◆ Harmonize with other top-level ontologies of biomedicine
  - Semantic types (e.g., with BioTop)
  - Semantic relationships (e.g., with the OBO Relation Ontology, with relationships in GALEN)
- ◆ Align relationships between Metathesaurus and Semantic Network



**Draft**

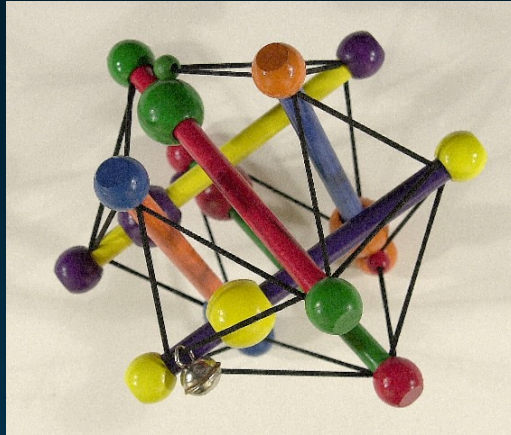


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- ◆ McCray AT, Bodenreider O.  
*A conceptual framework for the biomedical domain.*  
In: Green R, Bean CA, Myaeng SH, editors. *The semantics of relationships: an interdisciplinary perspective.*  
Dordrecht; Boston: Kluwer Academic Publishers; 2002. p. 181-198.

<http://semanticnetwork.nlm.nih.gov/>





# Medical Ontology Research

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