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UMLS Webcasts series
February 29, 2008

UMLS Semantic Network

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Acknowledgments

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  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

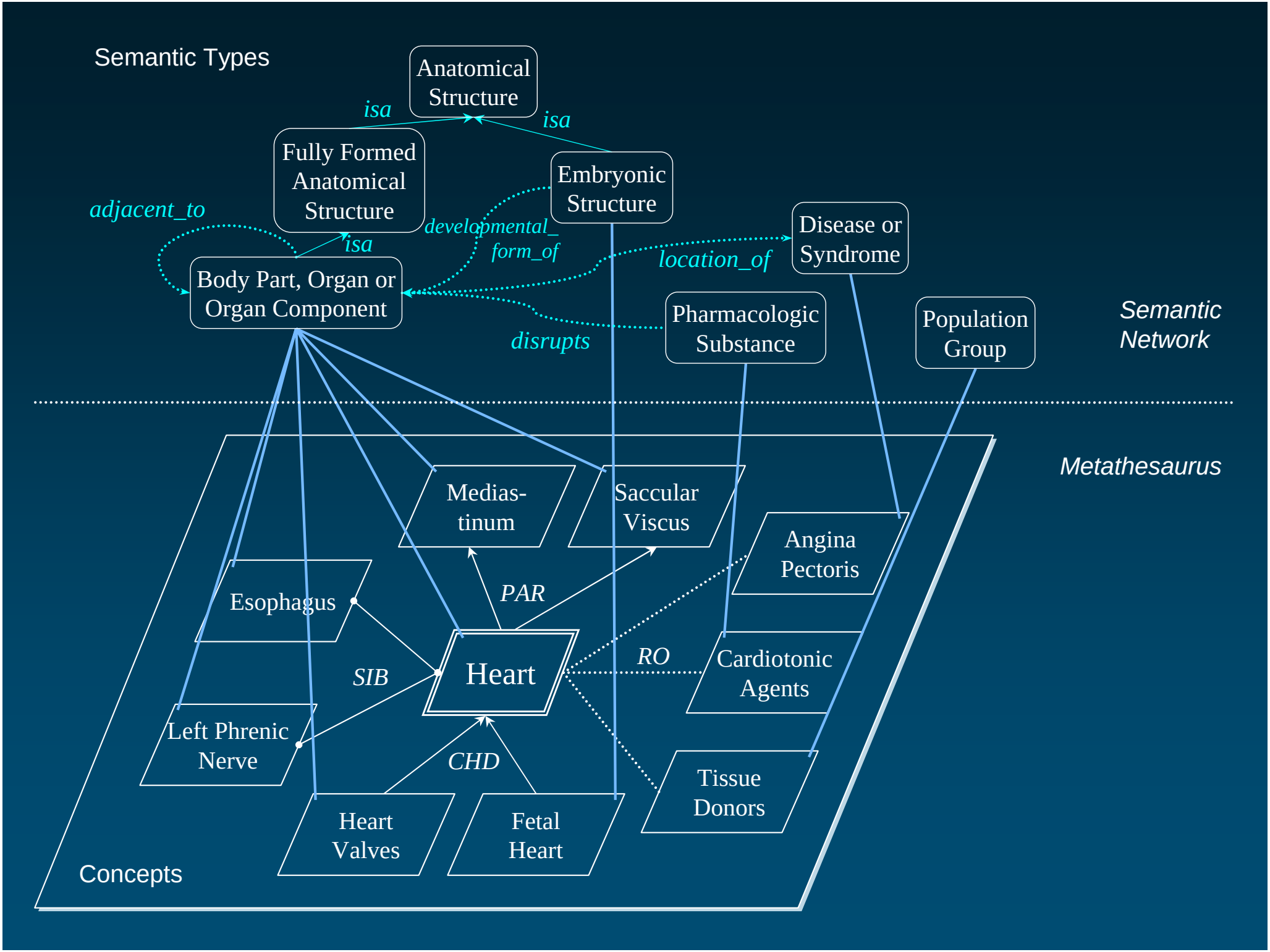
Lexical resources

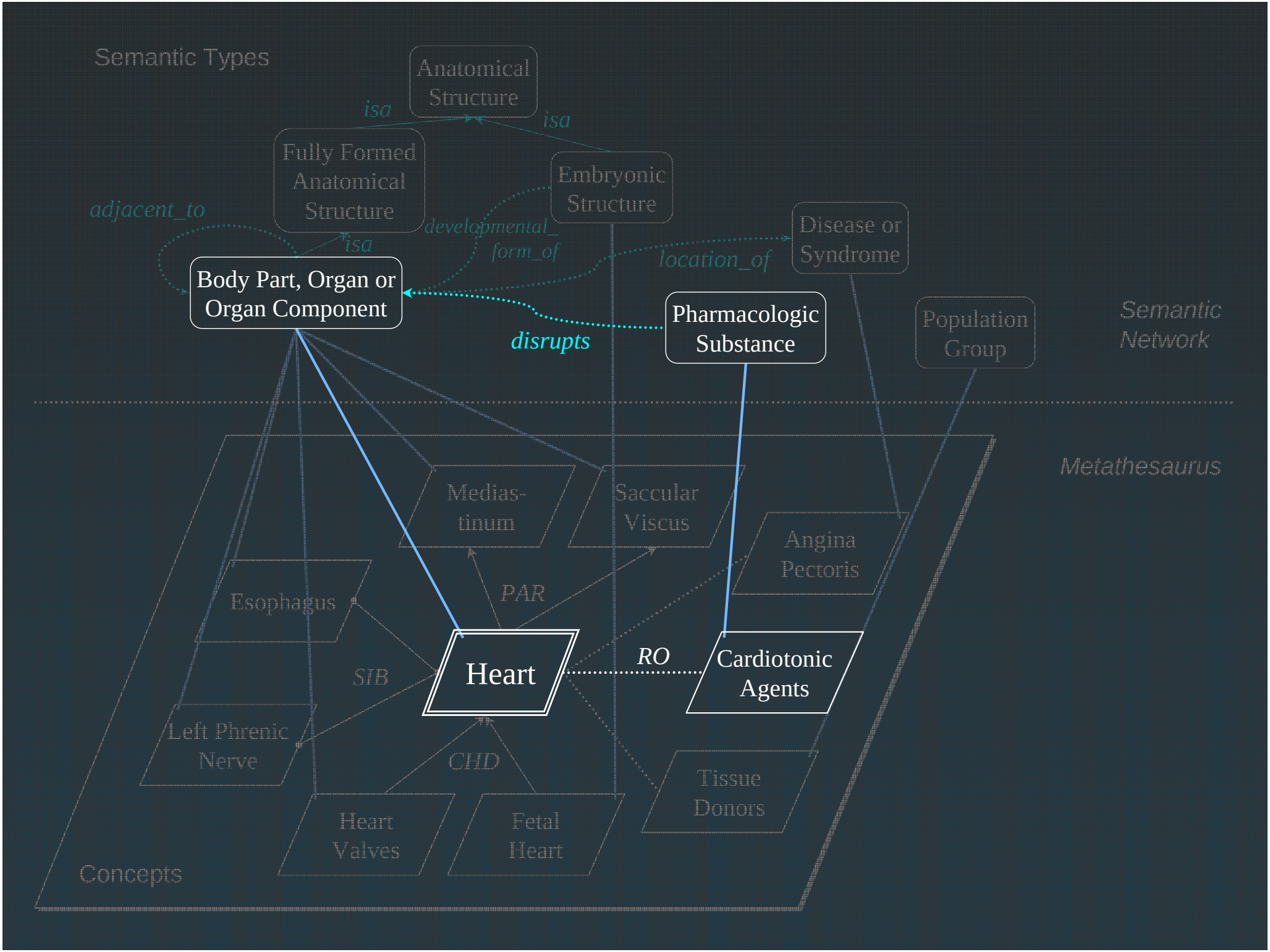
Terminological resources

Ontological resources

The UMLS

A two-level structure


Semantic Types
Anatomical Structure
isa
isa
Fully Formed Anatomical Structure
Embryonic Structure
adjacent_to
isa
developmental_ form_of
Disease or Syndrome
location_of
Body Part, Organ or Organ Component
Pharmacologic Substance
Population Group
disrupts
Semantic Network
Metathesaurus
Medias- tinum
Saccular Viscus
Angina Pectoris
Esophagus
PAR
Heart
RO
Cardiotonic Agents
SIB
Left Phrenic Nerve
CHD
Tissue Donors
Heart Valves
Fetal Heart
Concepts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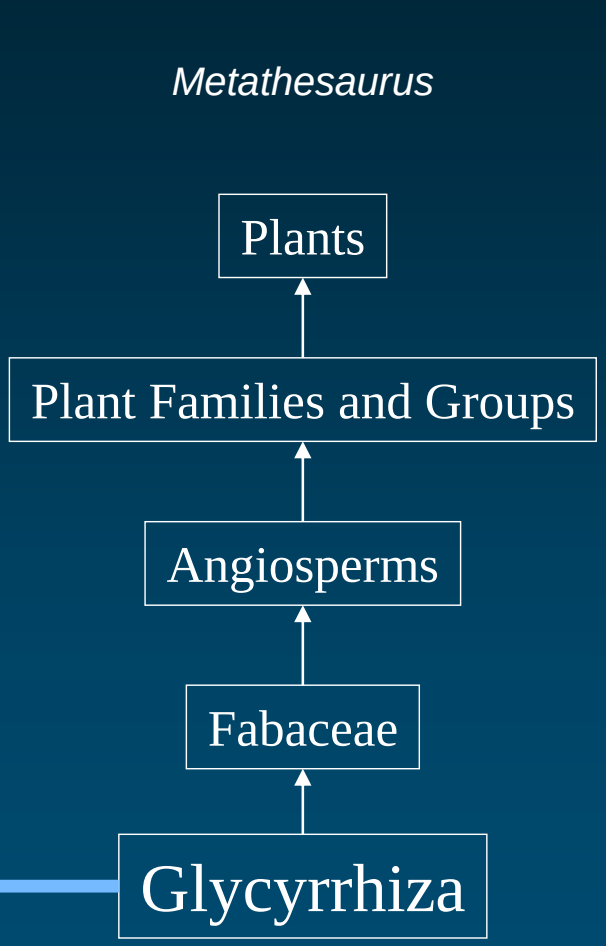
Cardiotonic Agents
Left Phrenic Nerve
CHD
Tissue Donors
Heart Valves
Fetal Heart
Concepts

Concept categorization

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

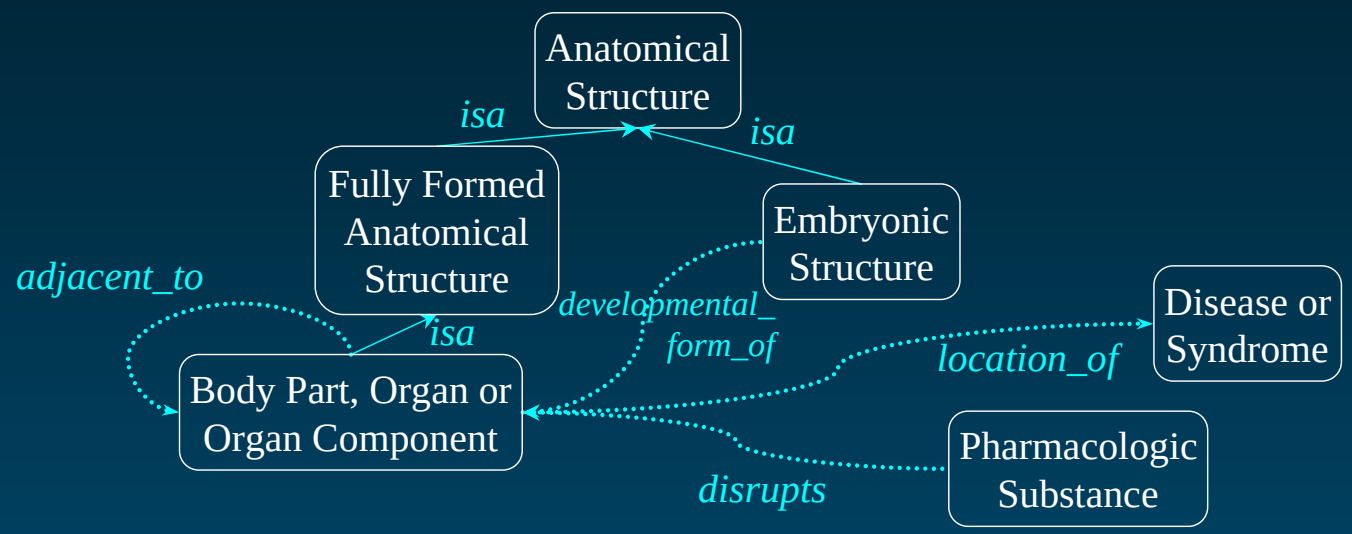
http://en.wikipedia.org/wiki/Glycyrrhiza



UMLS Semantic Network

What is it?

Semantic network



- Semantic *types*: Nodes
- Semantic *relationships*: Links
- Semantic *relations*: Triples ($node_1$, link, $node_2$)

Semantic types

- Semantic types (135)
  - tree structure
  - 2 major hierarchies
    - Entity
      - Physical Object
      - Conceptual Entity
    - Event
      - Activity
      - Phenomenon or Process

Entity

- Physical Object
  - Organism
    - Plant
      - Alga
    - Fungus
    - Virus
    - Rickettsia or Chlamydia
    - Bacterium
    - Archaeon
    - Animal
      - Invertebrate
      - Vertebrate
        - Amphibian
        - Bird
        - Fish
        - Reptile
        - Mammal
          - Human
  - Anatomical Structure
    - Embryonic Structure
    - Anatomical Abnormality
      - Congenital Abnormality
      - Acquired Abnormality
    - Fully Formed Anatomical Structure
      - Body Part, Organ, or Organ Component
      - Tissue
      - Cell
      - Cell Component
      - Gene or Genome

Physical Object [continued]

  - Manufactured Object
    - Medical Device
    - Research Device
    - Clinical Drug
  - Substance
    - Chemical
      - Chemical Viewed Functionally
        - Pharmacologic Substance
          - Antibiotic
        - Biomedical or Dental Material
        - Biologically Active Substance
          - Neuroreactive Substance or Biogenic Amine
          - Hormone
          - Enzyme
          - Vitamin
          - Immunologic Factor
          - Receptor
        - Indicator, Reagent, or Diagnostic Aid
        - Hazardous or Poisonous Substance
      - Chemical Viewed Structurally
        - Organic Chemical
          - Nucleic Acid, Nucleoside, or Nucleotide
          - Organophosphorus Compound
          - Amino Acid, Peptide, or Protein
          - Carbohydrate
          - Lipid
            - Steroid
            - Eicosanoid
        - Inorganic Chemical
        - Element, Ion, or Isotope
    - Body Substance
    - Food

Entity [continued]

- Conceptual Entity
  - Idea or Concept
    - Temporal Concept
    - Qualitative Concept
    - Quantitative Concept
    - Functional Concept
      - Body System
    - Spatial Concept
      - Body Space or Junction
      - Body Location or Region
      - Molecular Sequence
        - Nucleotide Sequence
        - Amino Acid Sequence
        - Carbohydrate Sequence
      - Geographic Area
  - Finding
    - Laboratory or Test Result
    - Sign or Symptom
  - Organism Attribute
    - Clinical Attribute
  - Intellectual Product
    - Classification
    - Regulation or Law
  - Language
  - Occupation or Discipline
    - Biomedical Occupation or Discipline
  - Organization
    - Health Care Related Organization
    - Professional Society
    - Self-help or Relief Organization
  - Group Attribute
  - Group
    - Professional or Occupational Group
    - Population Group
    - Family Group
    - Age Group
    - Patient or Disabled Group

Hierarchical structure Entity

Event

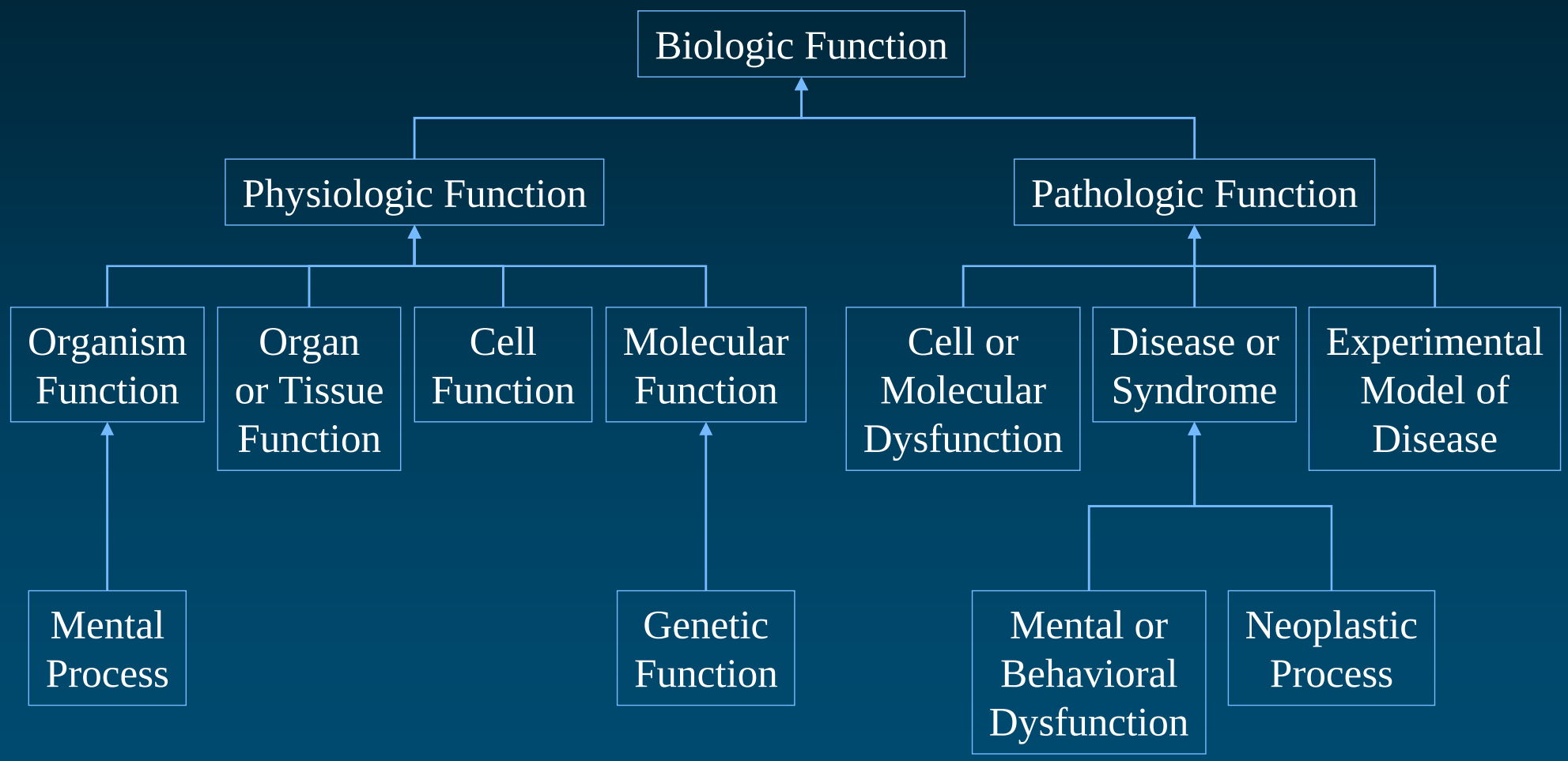
- Activity
  - Behavior
    - Social Behavior
    - Individual Behavior
  - Daily or Recreational Activity
  - Occupational Activity
    - Health Care Activity
      - Laboratory Procedure
      - Diagnostic Procedure
      - Therapeutic or Preventive Procedure
    - Research Activity
      - Molecular 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)



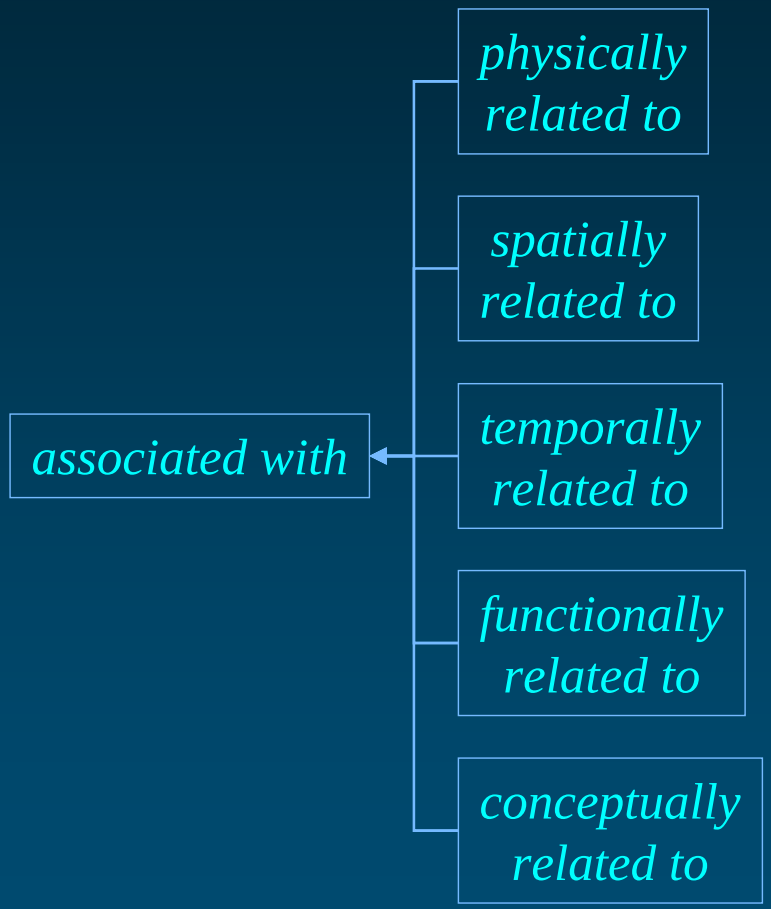
Semantic type information

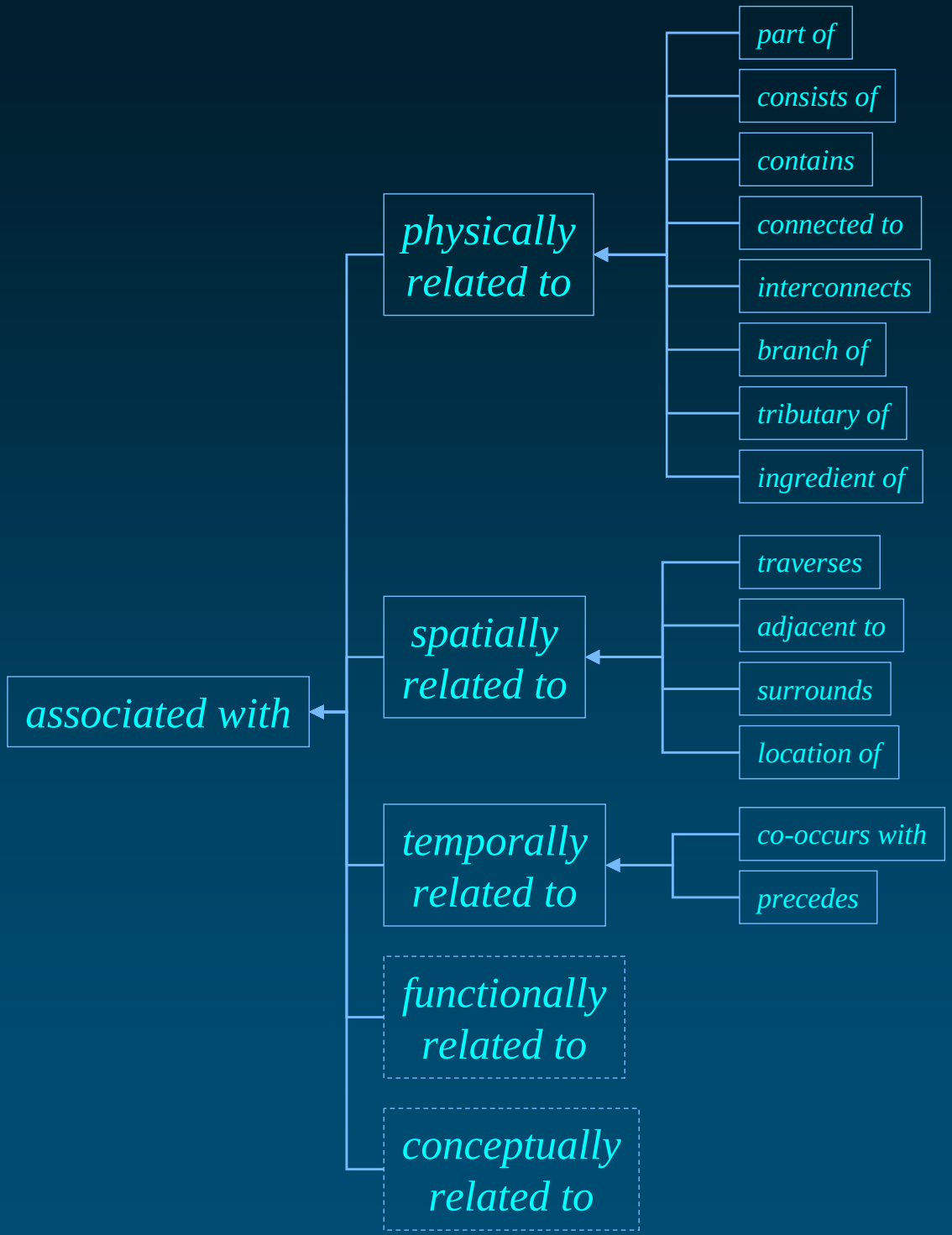
ID	• T047
Name	• Disease or Syndrome
Tree number	• B2.2.1.2.1
Definition	• *A condition which alters or interferes with a normal process, state, or activity of an organism. It is usually characterized by the abnormal functioning of one or more of the host's systems, parts, or organs. Included here is 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

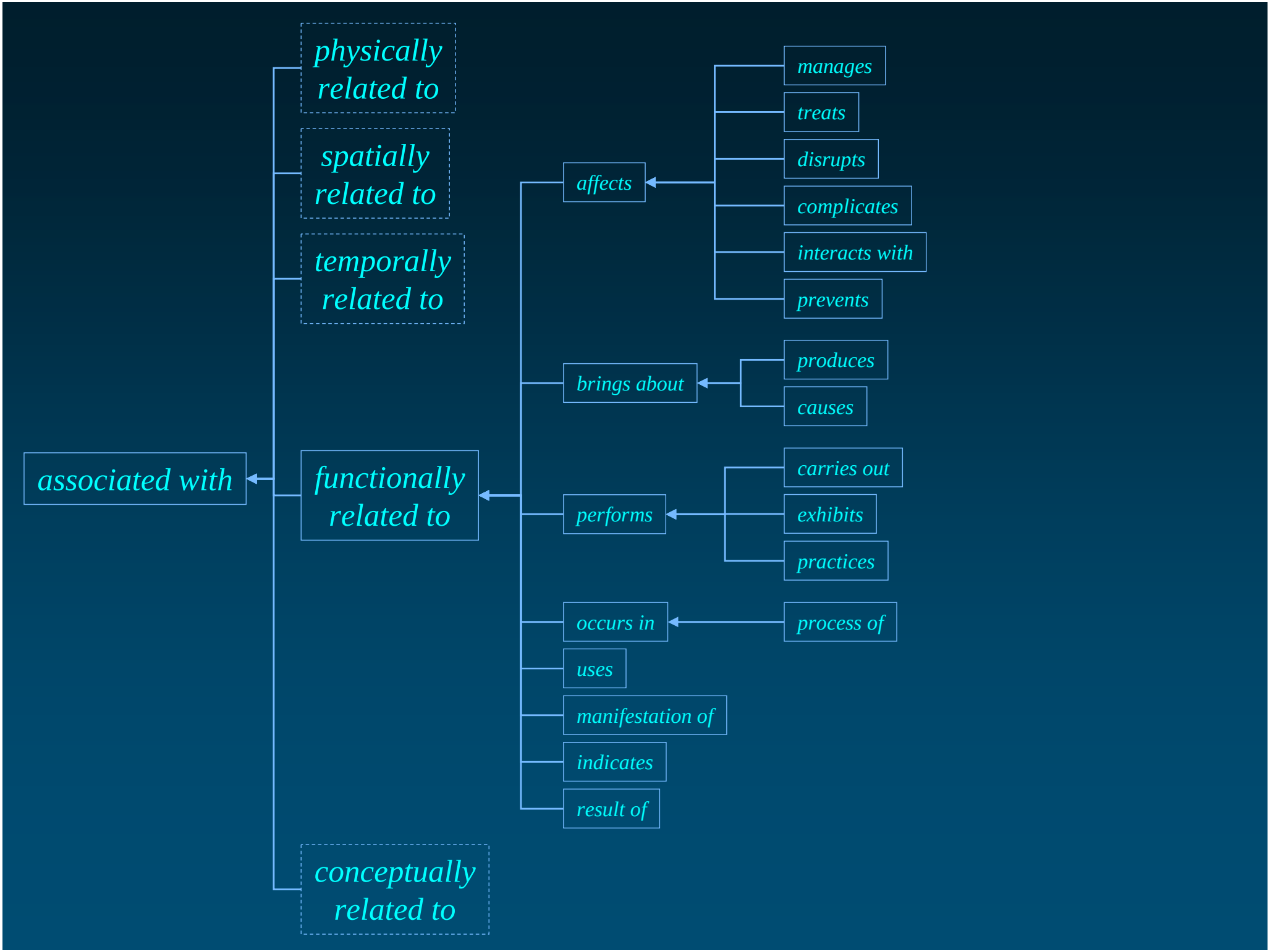
Semantic relationships

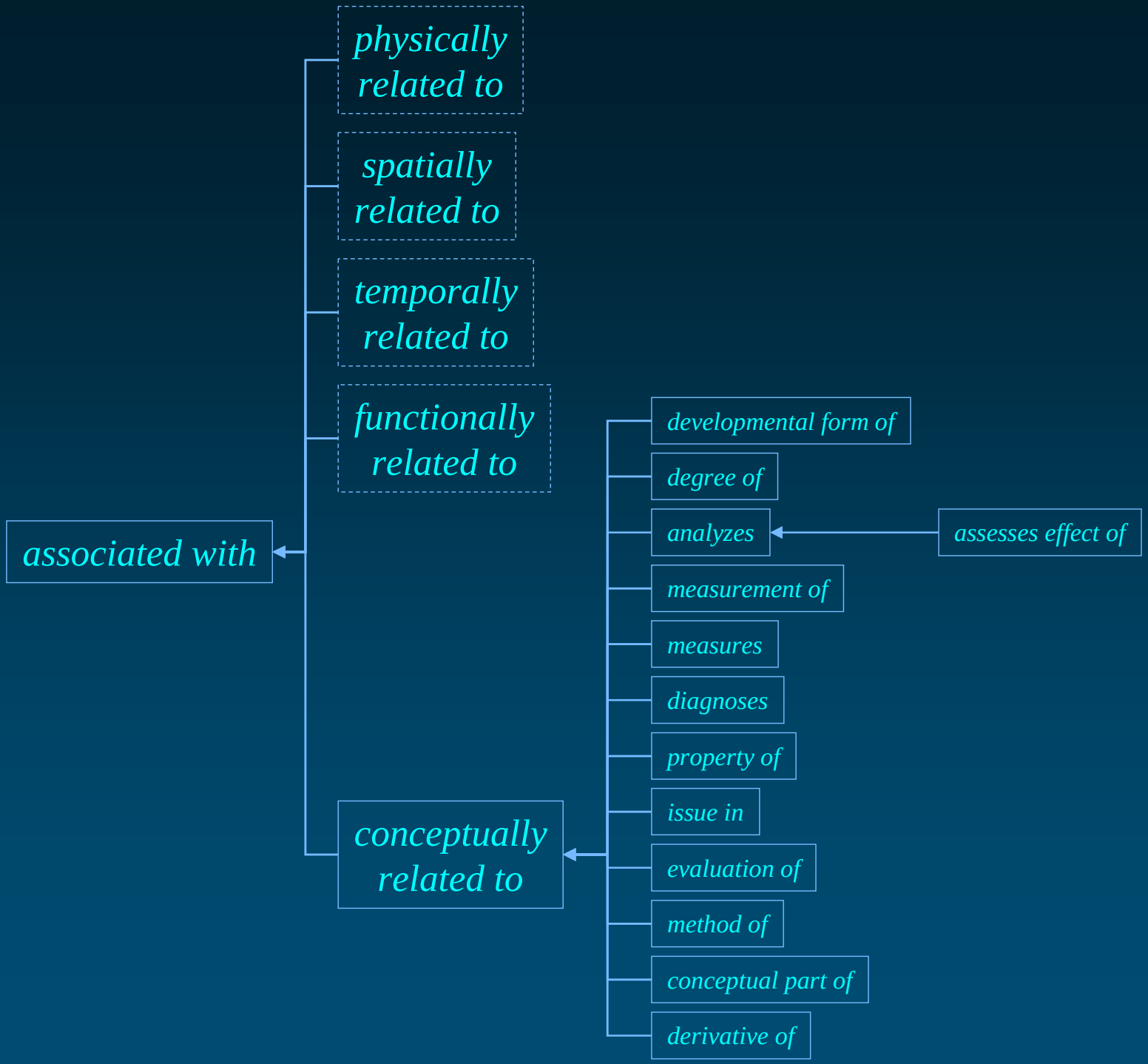
- Semantic network relationships (54)
  - Hierarchical (isa = is a kind of)
    - among types
      - Animal *isa* Organism
      - Enzyme *isa* Biologically Active Substance
    - among relations
      - treats *isa* affects
  - Associative (non-hierarchical)
    - Sign or Symptom *diagnoses* Pathologic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

Associative relationship hierarchy




associated with
physically related to
part of
consists of
contains
connected to
interconnects
branch of
tributary of
ingredient of
spatially related to
traverses
adjacent to
surrounds
location of
temporally related to
co-occurs with
precedes
functionally related to
conceptually related to


associated with
physically related to
spatially related to
temporally related to
functionally related to
conceptually related to
affects
manages
treats
disrupts
complicates
interacts with
prevents
brings about
produces
causes
performs
carries out
exhibits
practices
occurs in
process of
uses
manifestation of
indicates
result of


associated with
physically related to
spatially related to
temporally related to
functionally related to
conceptually related to
developmental form of
degree of
analyzes
assesses effect of
measurement of
measures
diagnoses
property of
issue in
evaluation of
method of
conceptual part of
derivative of

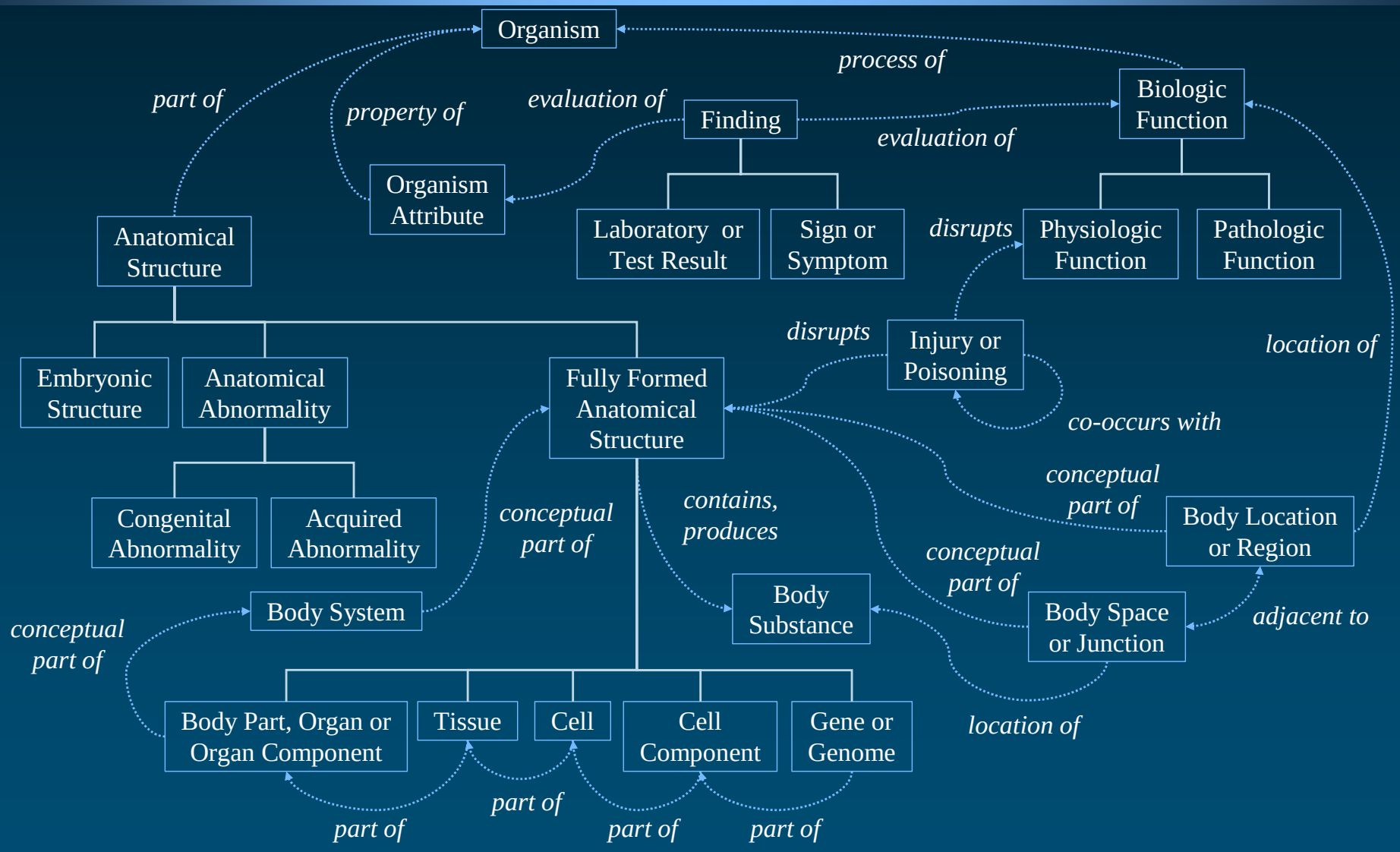
Semantic relationship information

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

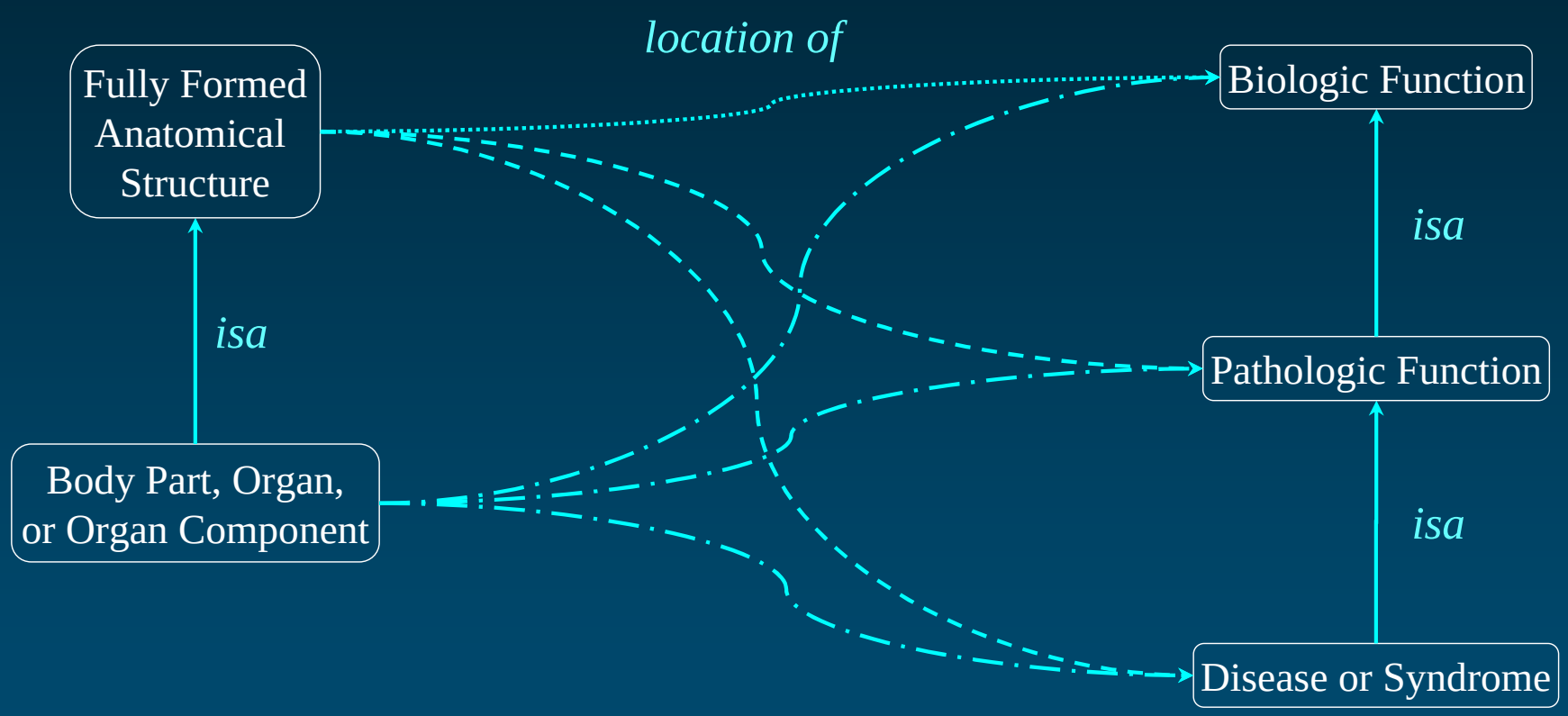
Semantic relations

- 612 assertions
  - 558 ($type_1$, relationship, $type_2$)
    - Hierarchical
      - Animal *isa* Organism
      - Enzyme *isa* Biologically Active Substance
    - Associative
      - Sign or Symptom *diagnoses* Pathologic Function
      - Pharmacologic Substance *treats* Pathologic Function
  - 54 ($relationship_1$, *isa*, $relationship_2$)
      - treats *isa* affects

Semantic relations (subset)



Inheritance



Semantic relations (inherited)

- 6864 assertions
  - 6752 ($type_1$, relationship, $type_2$)
    - 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 ($relationship_1$, *isa*, $relationship_2$)
    - treats *isa* functionally_related_to (from: treats *isa* affects)

Semantic relation information

Sem. $Type_1$	Fully Formed Anatomical Structure
Relationship	location_of
Sem. $Type_2$	Biologic Function
Link status	D

UMLS Semantic Network

How to use it?

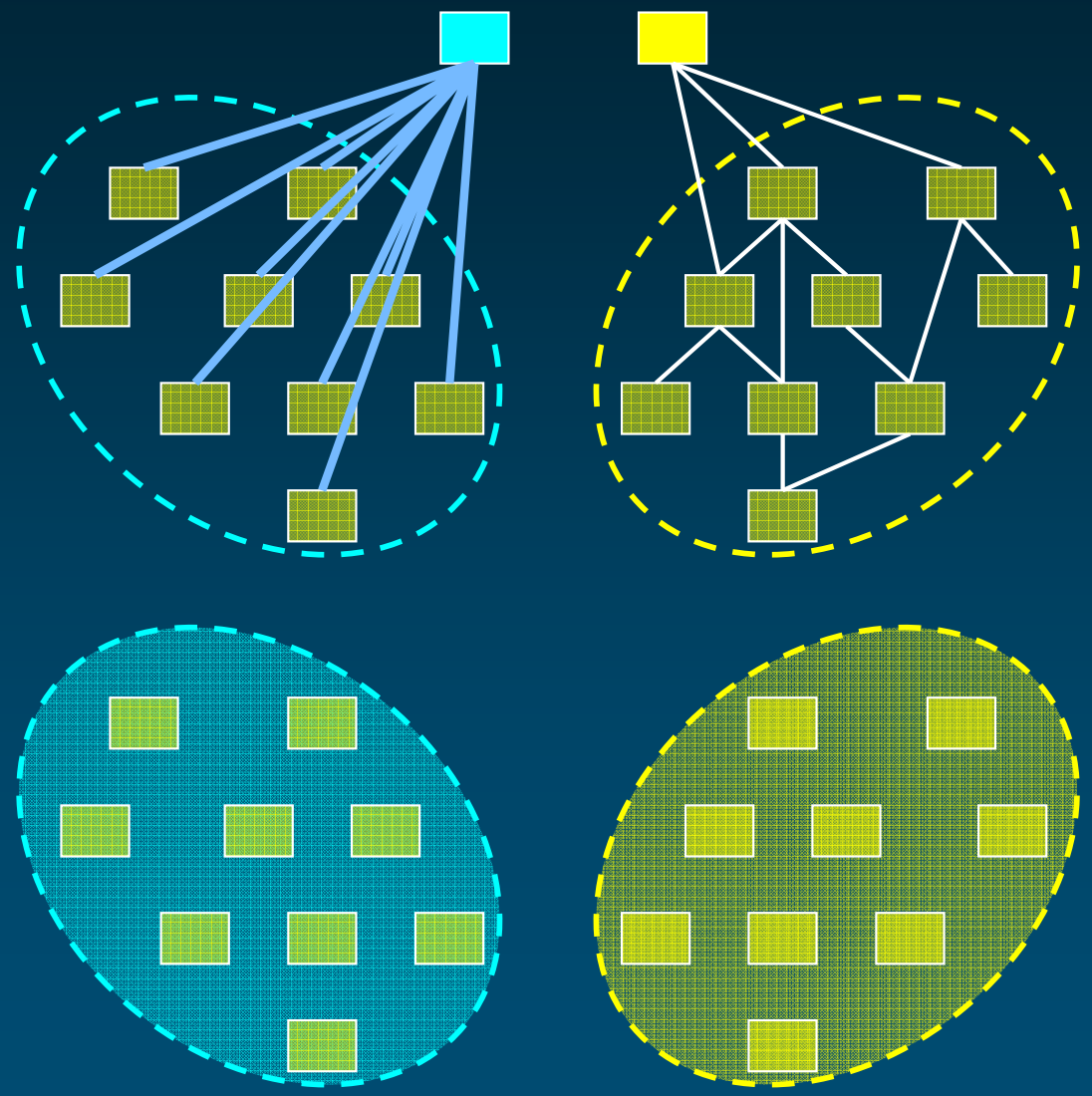
Why a semantic network?

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

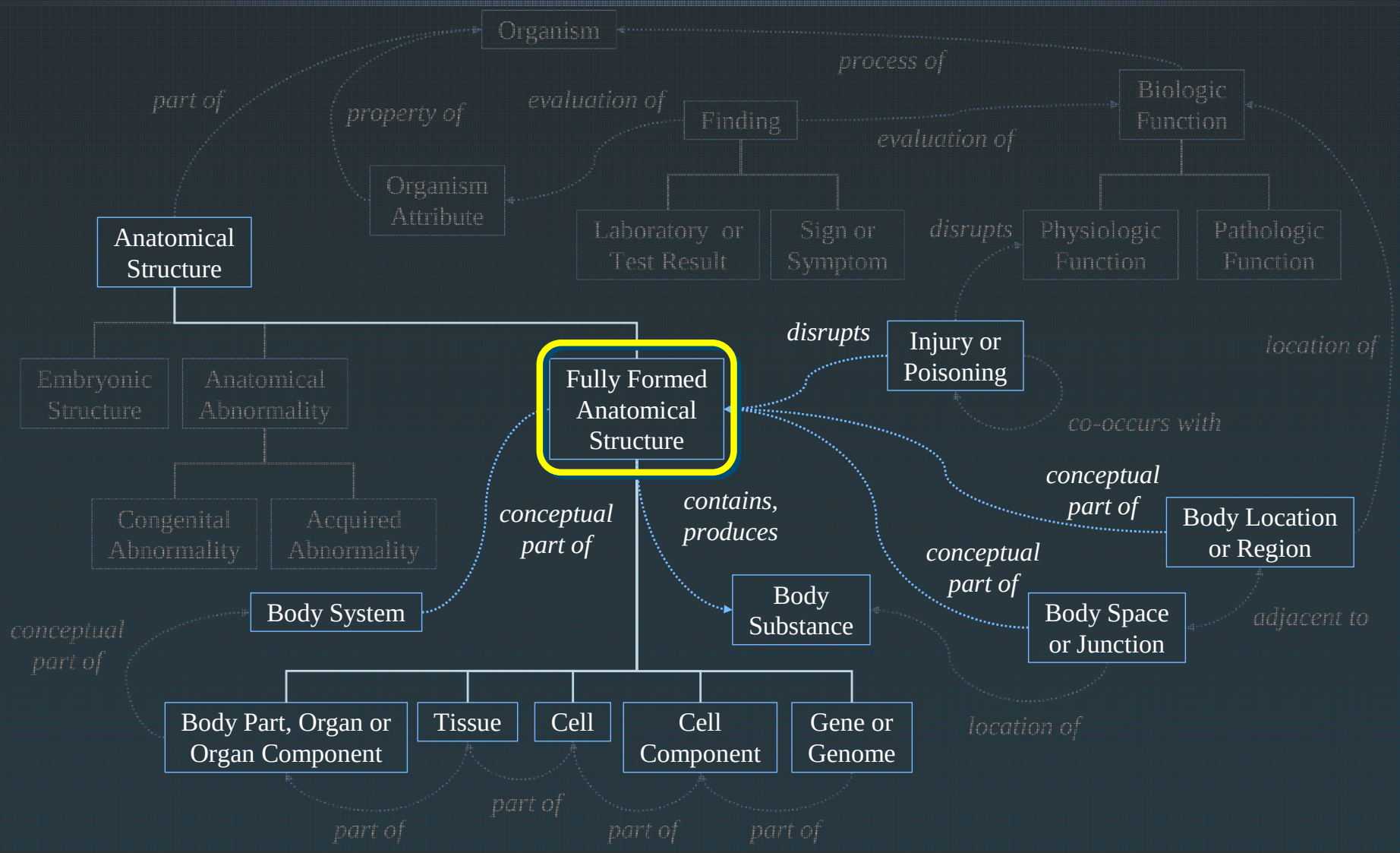
- A relation between 2 Semantic Types (ST) indicates a possible link between 2 concepts that have been assigned to those STs
  - The relationship may or may not hold at the concept level
  - Other relationships may apply at the concept level

Identify concepts for a given category

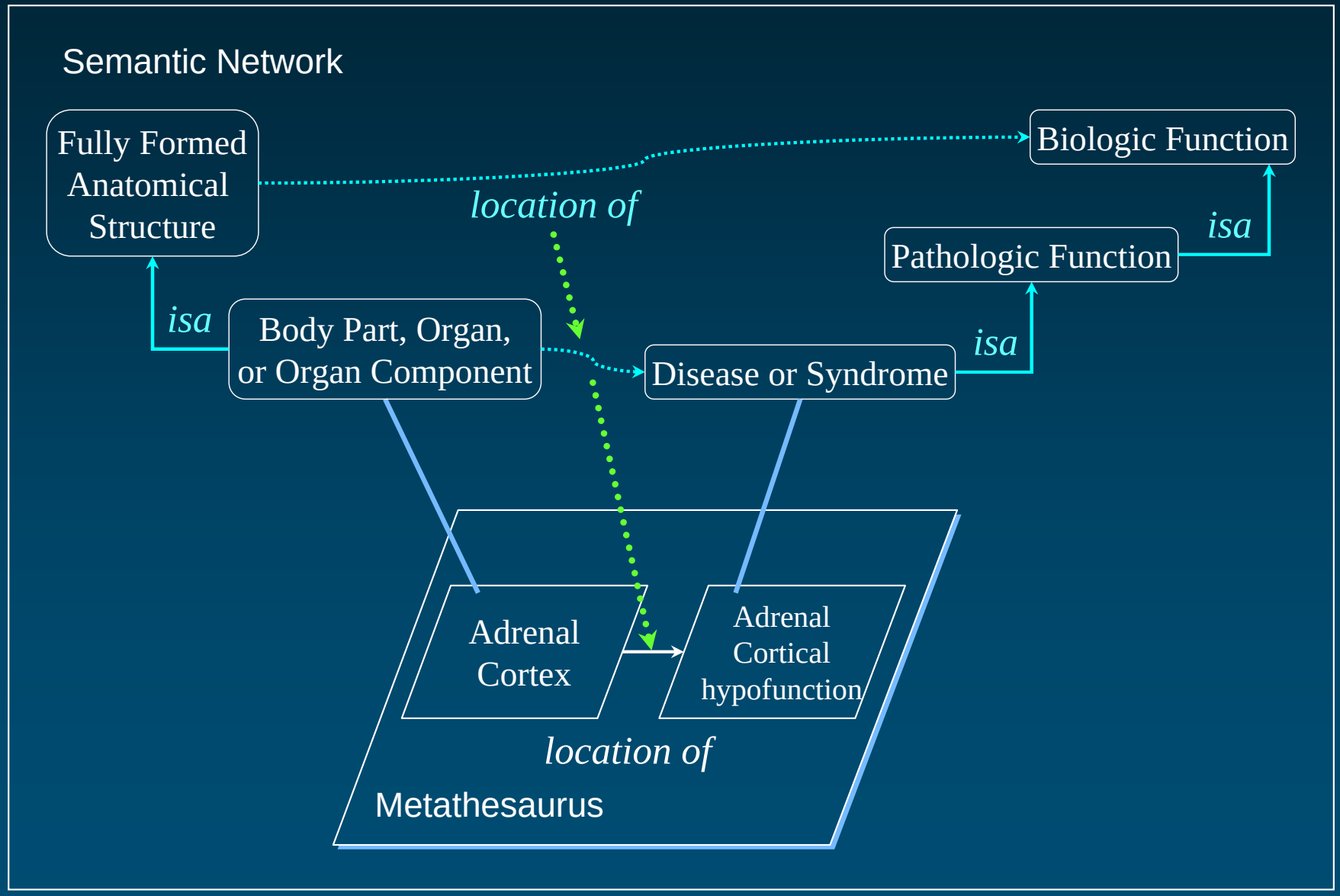
- Semantic type
  - List of all concepts having this semantic type
- Concept
  - List of all descendants



Source of domain knowledge



Semantics of Metathesaurus relations

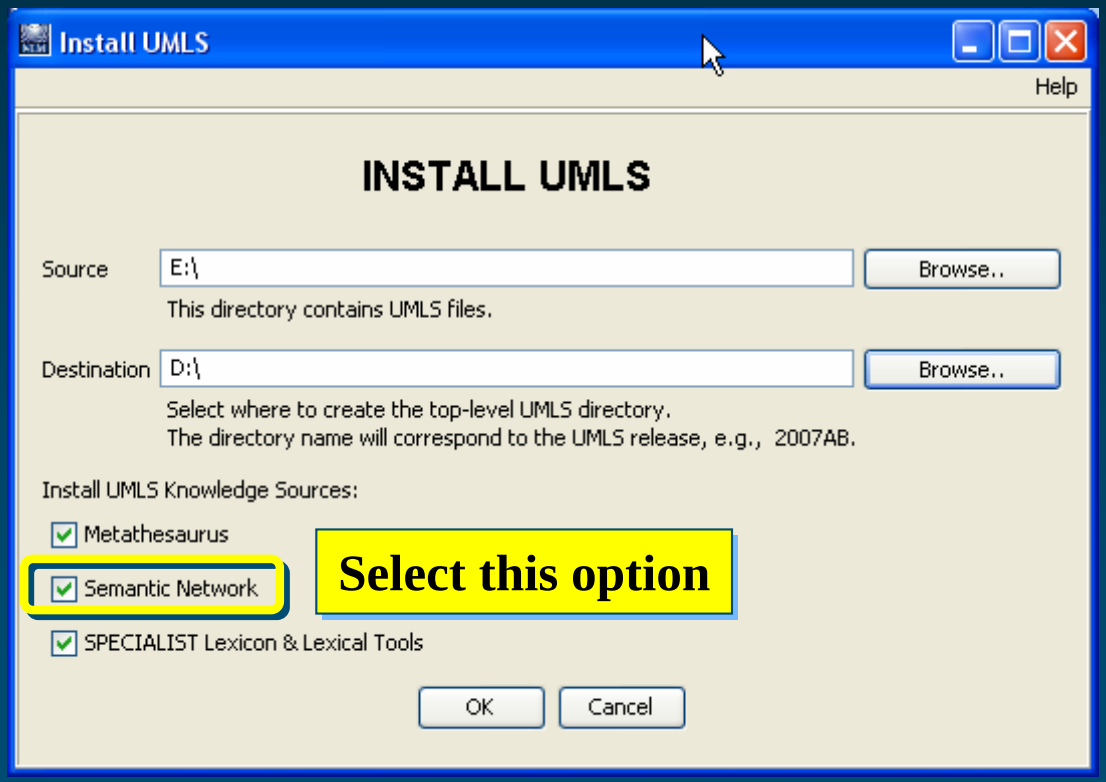


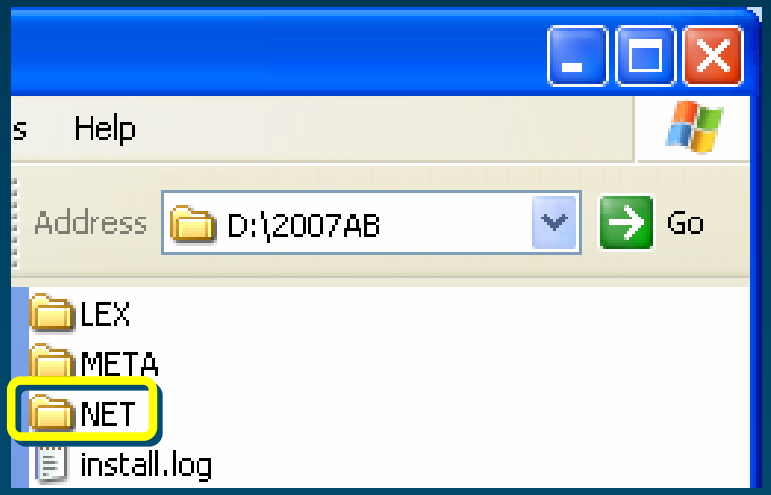
UMLS Semantic Network

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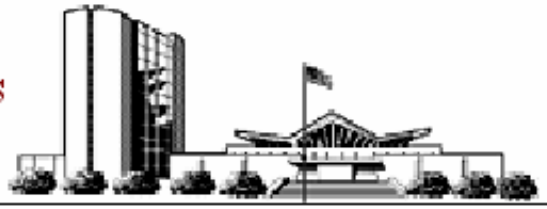
http://semanticnetwork.nlm.nih.gov/

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*
- NEW 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	+		+	+	View detailed summary
part_of	+		+	+	View detailed summary
integral_part_of	+		+	+	View detailed summary
proper_part_of	+				View detailed summary
located_in	+		+		View detailed summary
contained_in					View detailed summary
adjacent_to					View detailed summary
transformation_of	+				View detailed summary
derives_from	+				View detailed summary
preceded_by	+				View detailed summary
has_participant					View detailed summary
has_agent					View detailed summary
instance_of					View detailed summary

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

GALEN relations *isIngredientOf*

http://opengalen.org

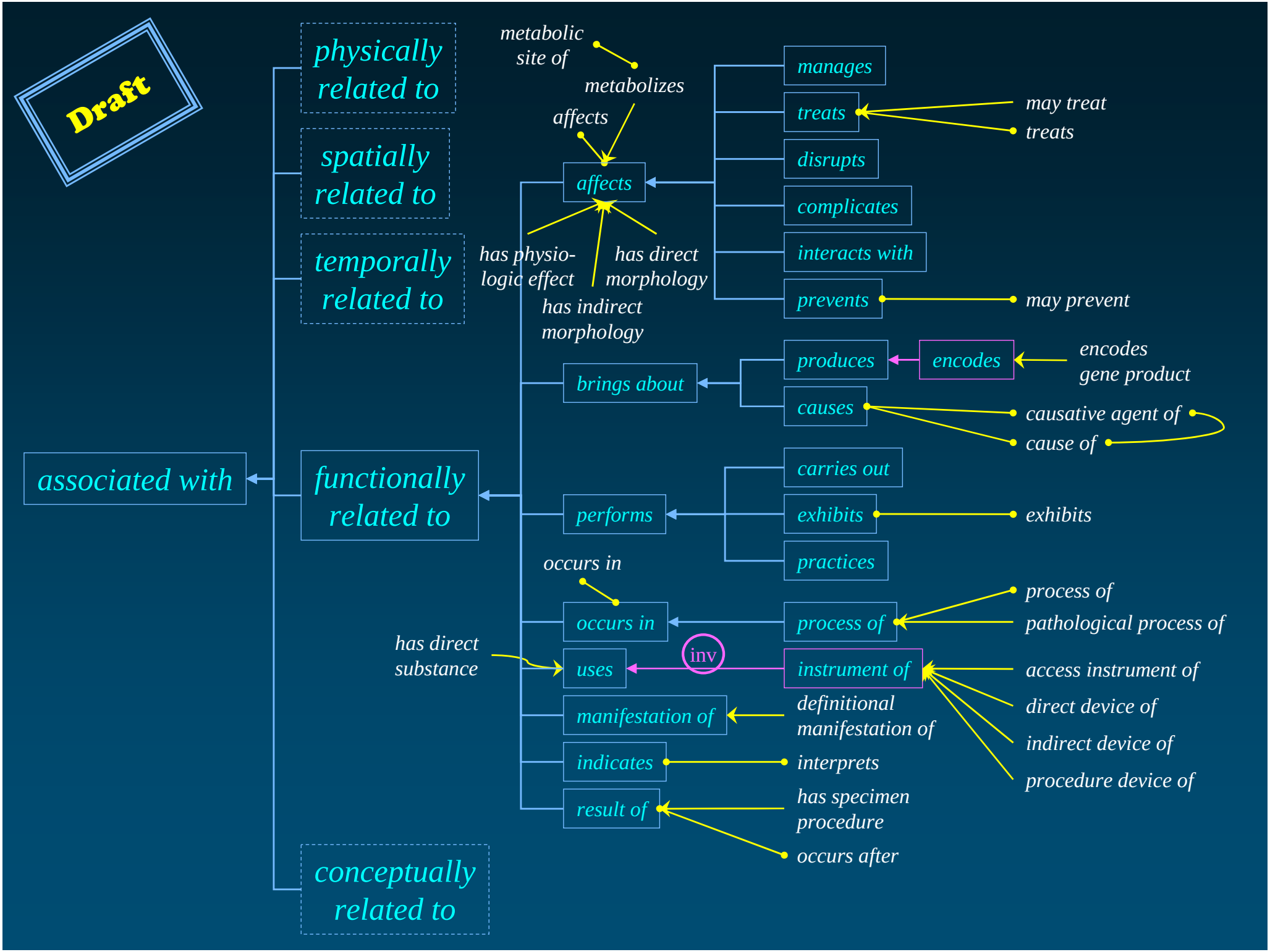
```
+ topLinkClass
Attribute
- DomainAttribute
  - ConstructiveAttribute
    - InversePartitiveAttribute
      - InverseStructuralPartitiveAttribute
        + IsDivisionOf
        - makesUp
          x specificallyMakesUp
          - isIngredientOf
            x isSpecificIngredientOf
            x isMultipleIngredientOf
          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
associated with
physically related to
spatially related to
temporally related to
functionally related to
conceptually related to
metabolic site of
metabolizes
affects
affects
has physio-logic effect
has direct morphology
has indirect morphology
manages
treats
may treat
treats
disrupts
complicates
interacts with
prevents
may prevent
brings about
produces
encodes
encodes gene product
causes
causative agent of
cause of
performs
carries out
exhibits
exhibits
practices
occurs in
occurs in
process of
process of
pathological process of
has direct substance
uses
inv
instrument of
access instrument of
direct device of
indirect device of
procedure device of
manifestation of
definitional manifestation of
indicates
interprets
result of
has specimen procedure
occurs after

References

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  Methods Inf Med 1995;34(1-2):193-201
- McCray AT.
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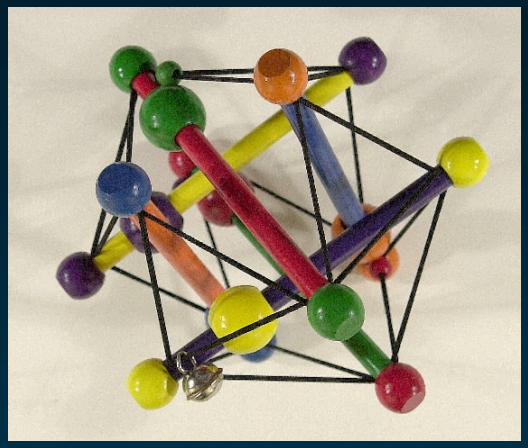
  A conceptual framework for the biomedical domain.
  In: Green R, Bean CA, Myaeng SH, editors. The semantics of relationships: an interdisciplinary perspective.
  Dordrecht; Boston: Kluwer Academic Publishers; 2002. p. 181-198.

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



Medical Ontology Research

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