Associate Fellowship Program
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Unified Medical Language System
Overview

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Outline

- Introduction
- Overview through an example
  Addison's disease
- The three UMLS Knowledge Sources
  - UMLS Metathesaurus
  - UMLS Semantic Network
  - SPECIALIST Lexicon and lexical tools

Introduction

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"
- Complementary to IAIMS

(Integrated Academic Information Management Systems)

«[…] 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

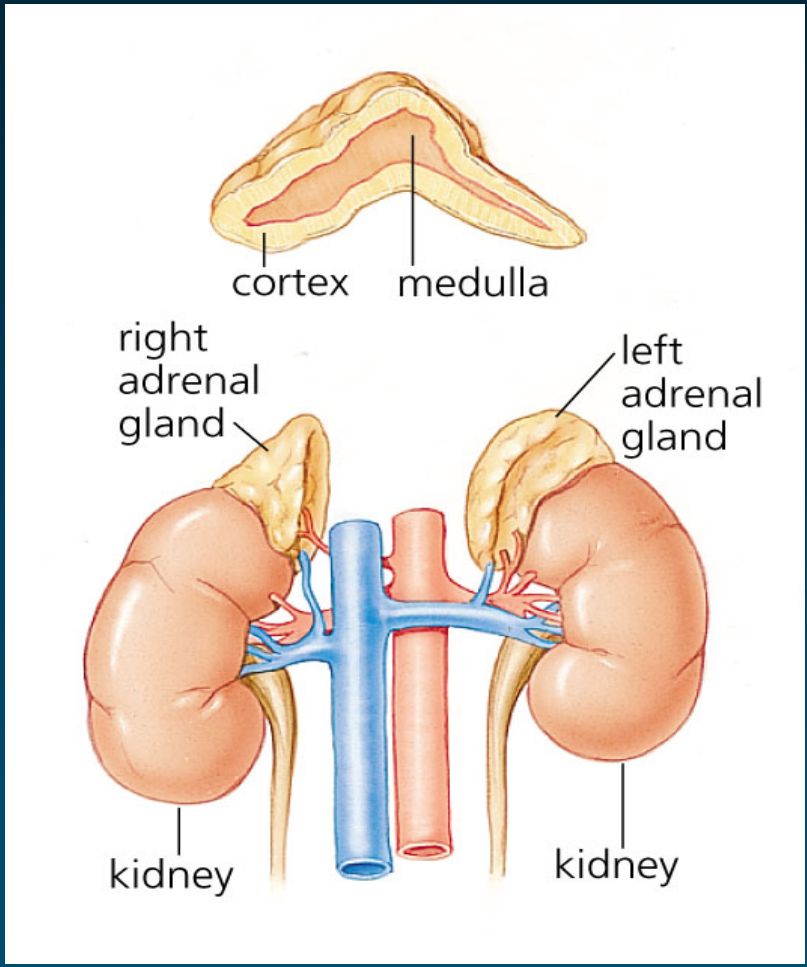
- Database
  - Series of relational files
- Interfaces
  - Web interface: UMLS Terminology Services (UTS)
  - Application programming interfaces (Java and web services)
- Applications
  - lvg (lexical programs)
  - MetamorphoSys (installation and customization)
  - RRF browser (browsing subsets)

The UMLS is *not* an end-user application

Overview through an example

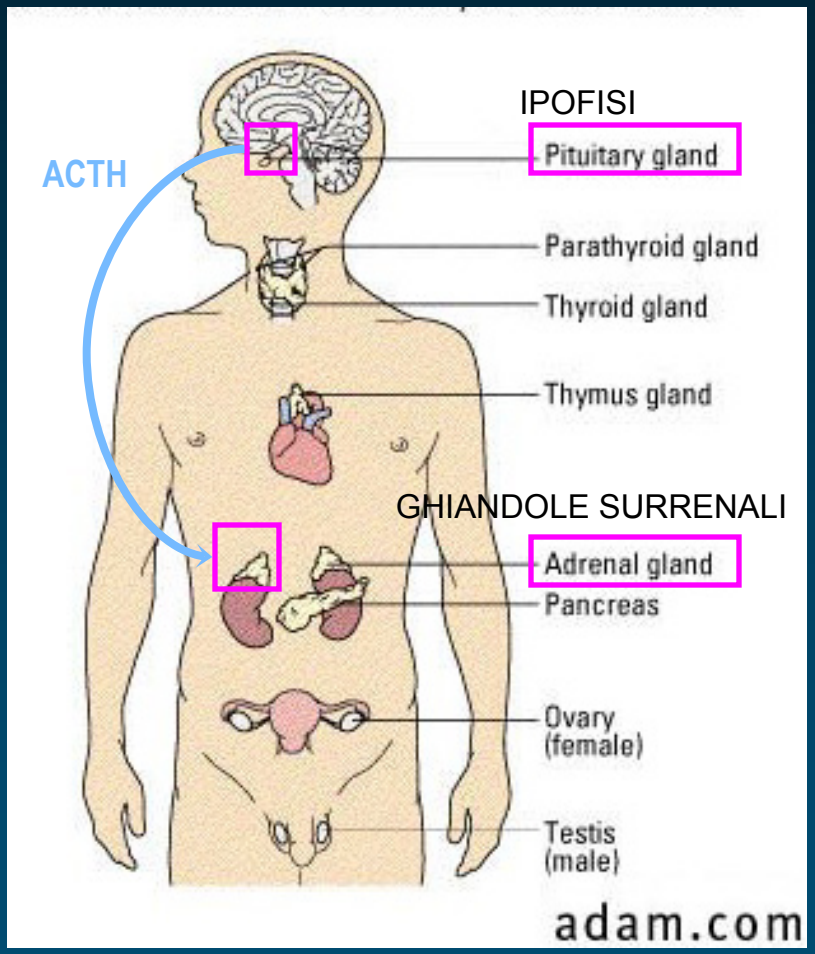
Addison's disease

- Addison's disease is a rare endocrine disorder
- Addison's disease occurs when the adrenal glands do not produce enough of the hormone cortisol
- For this reason, the disease is sometimes called chronic adrenal insufficiency, or hypocortisolism



Adrenal insufficiency Clinical variants

- Primary / Secondary
  - Primary: lesion of the adrenal glands themselves
  - Secondary: inadequate secretion of ACTH by the pituitary gland
- Acute / Chronic
- Isolated / Polyendocrine deficiency syndrome



L'ormone adrenocorticotropo (ACTH) stimola la corteccia surrenale a produrre gli ormoni legati al metabilismo dei grassi

Addison's disease: Symptoms

- Fatigue
- Weakness
- Low blood pressure
- Pigmentation of the skin (exposed and non-exposed parts of the body)
- …

AD in medical vocabularies

- Synonyms: different terms
  - Addisonian syndrome — eponym
  - Bronzed disease — symptoms
  - Melasma addisonii — symptoms
  - Asthenia pigmentosa — symptoms
  - Primary adrenal deficiency — clinical variants
  - Primary adrenal insufficiency — clinical variants
  - Primary adrenocortical insufficiency — clinical variants
  - Chronic adrenocortical insufficiency — clinical variants
- Contexts: different hierarchies

Organize terms

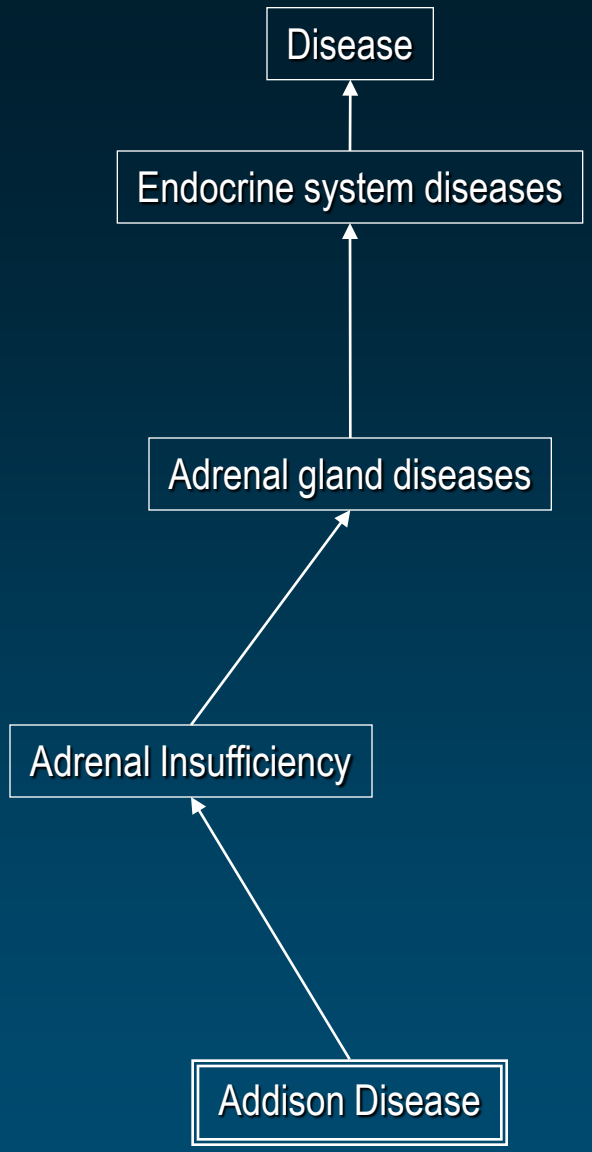
- Synonymous terms clustered into a concept
- Preferred term
- Unique identifier (CUI)

Addison Disease	MeSH	D000224
Primary hypoadrenalism	MedDRA	10036696
Primary adrenocortical insufficiency	ICD-10	E27.1
Addison's disease (disorder)	SNOMED CT	363732003

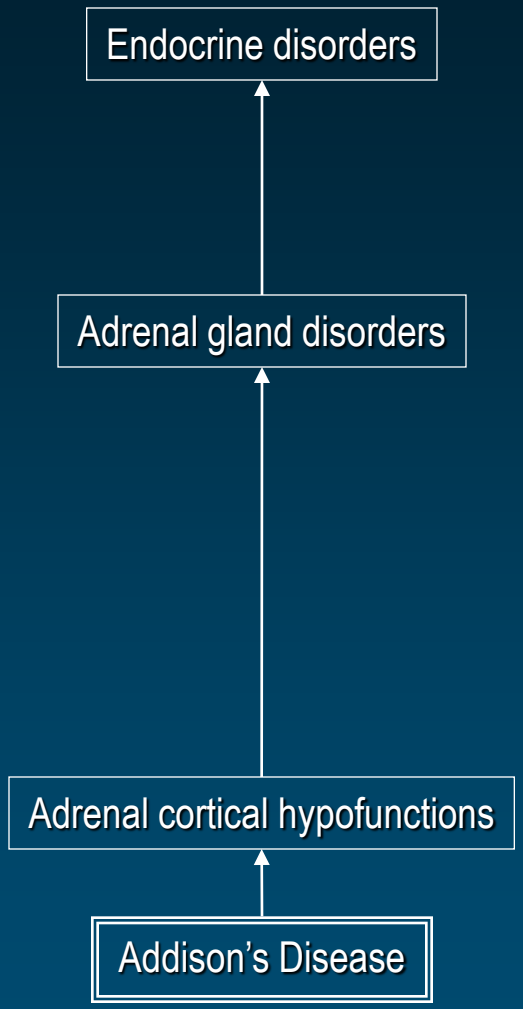
C0001403

Addison's disease

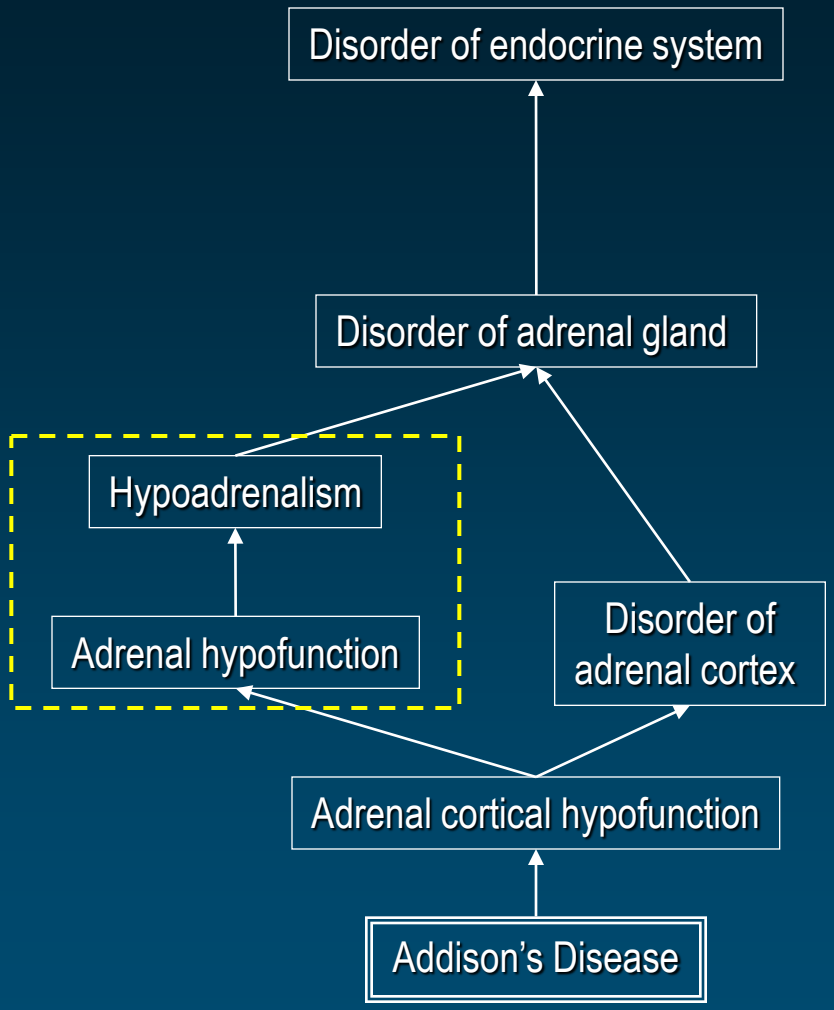
MeSH


Disease
Endocrine system diseases
Adrenal gland diseases
Adrenal Insufficiency
Addison Disease

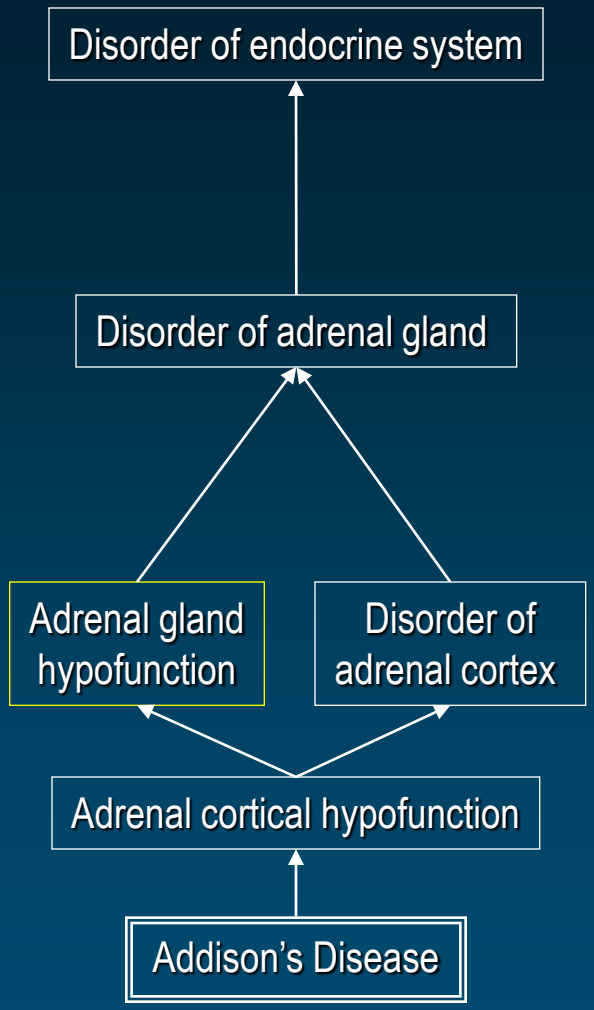
MedDRA


Endocrine disorders
Adrenal gland disorders
Adrenal cortical hypofunctions
Addison's Disease

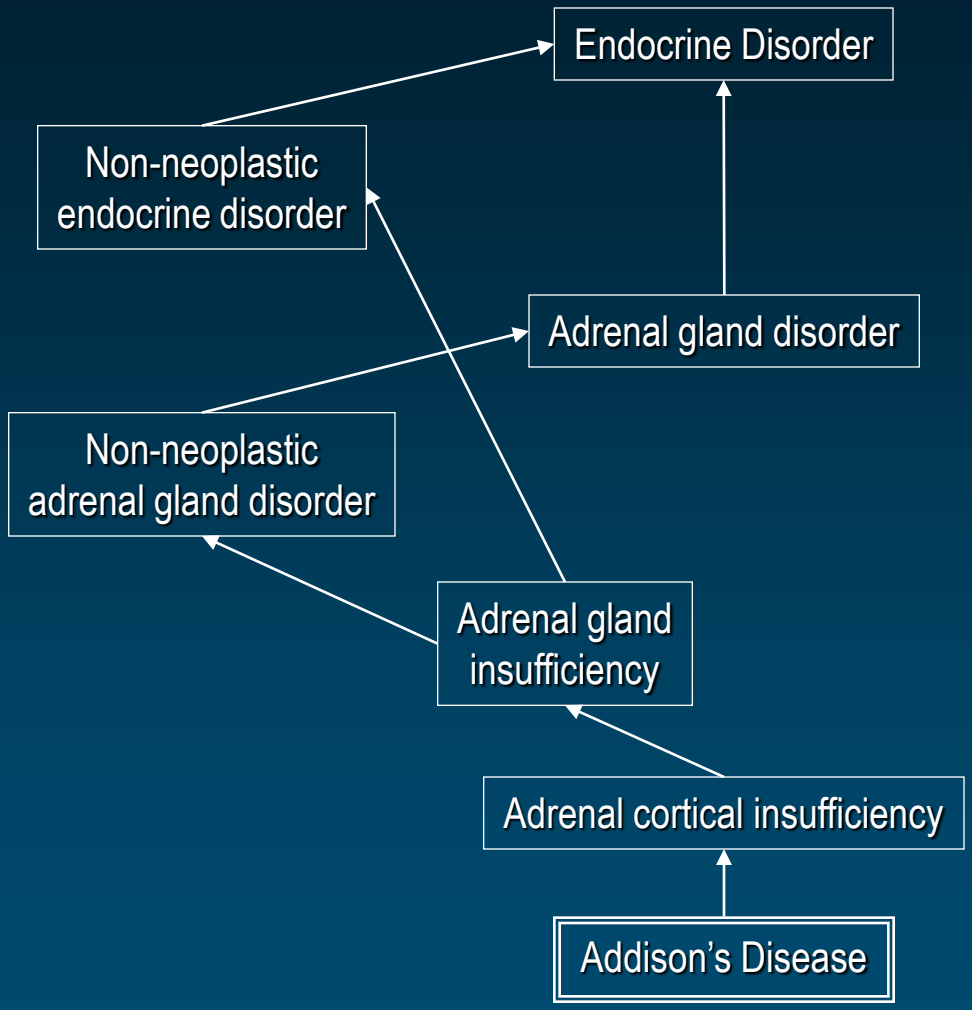
SNOMED CT (native)



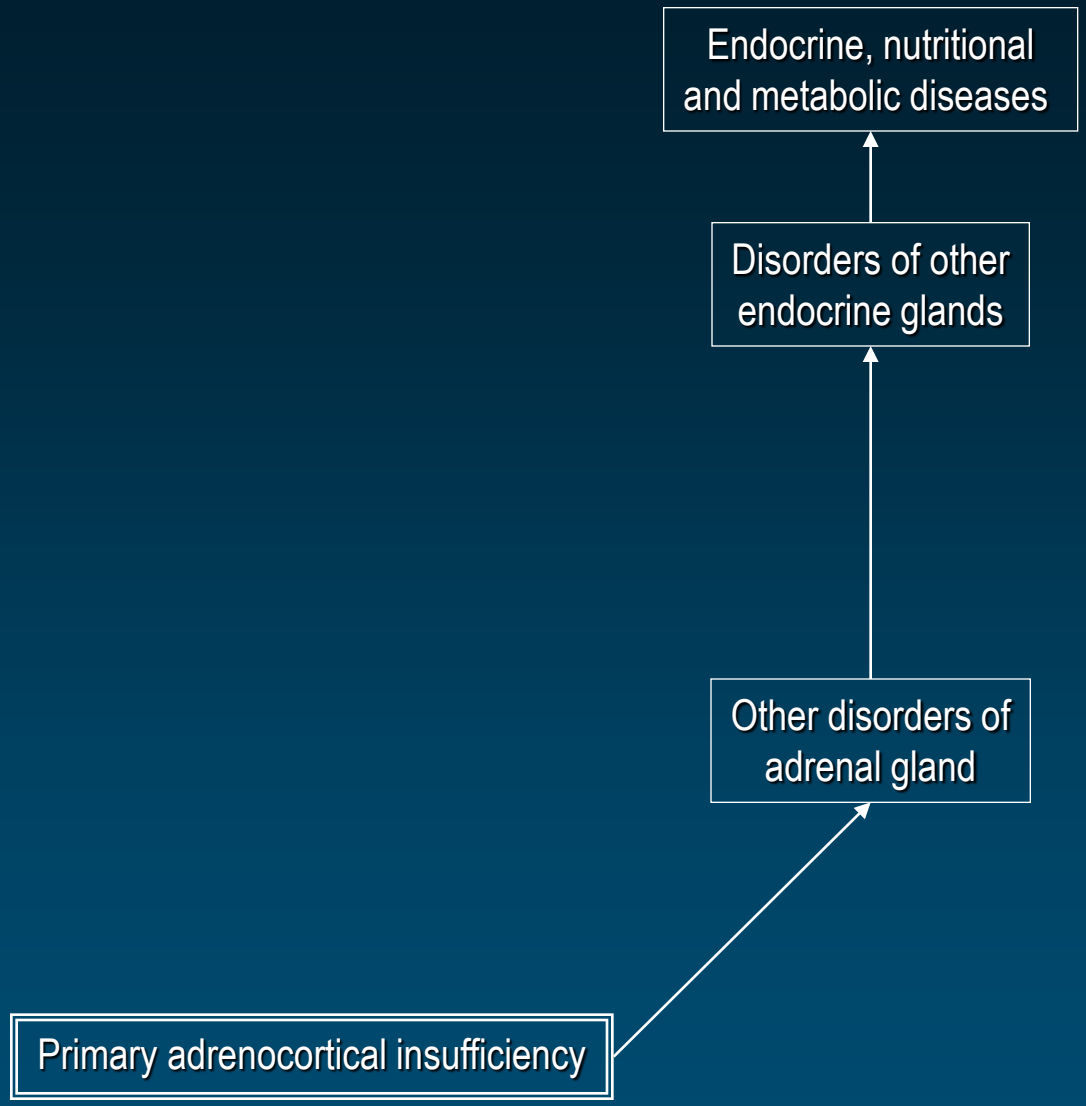
SNOMED CT (UMLS view)



NCI Thesaurus

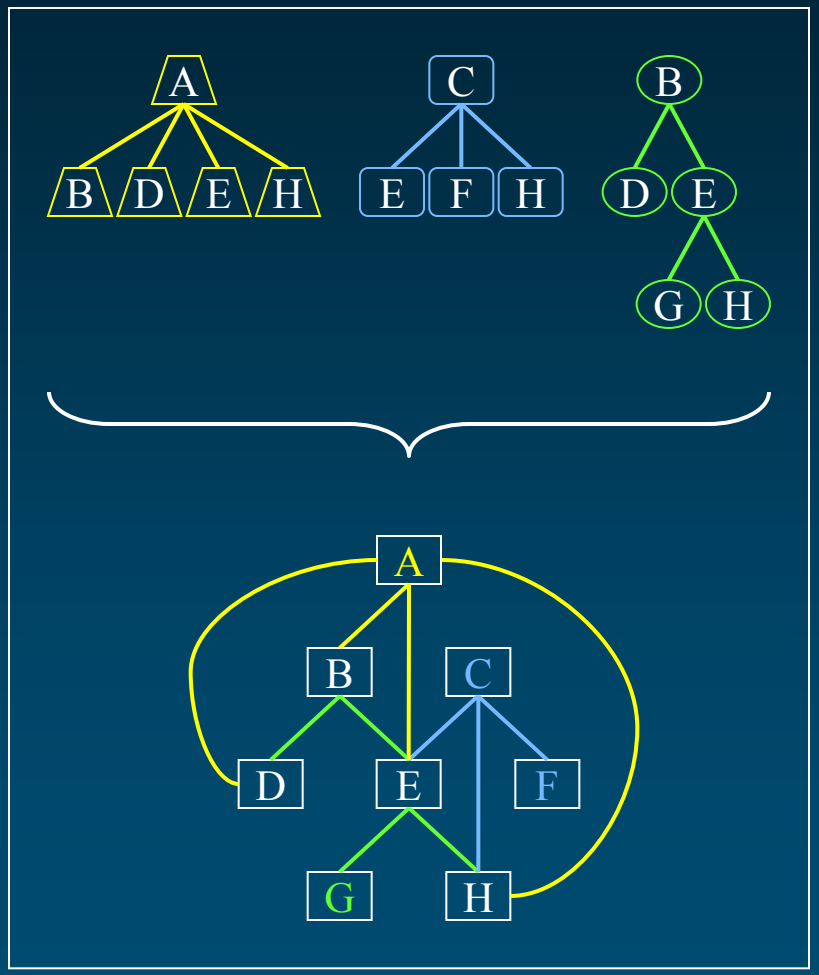


ICD-10

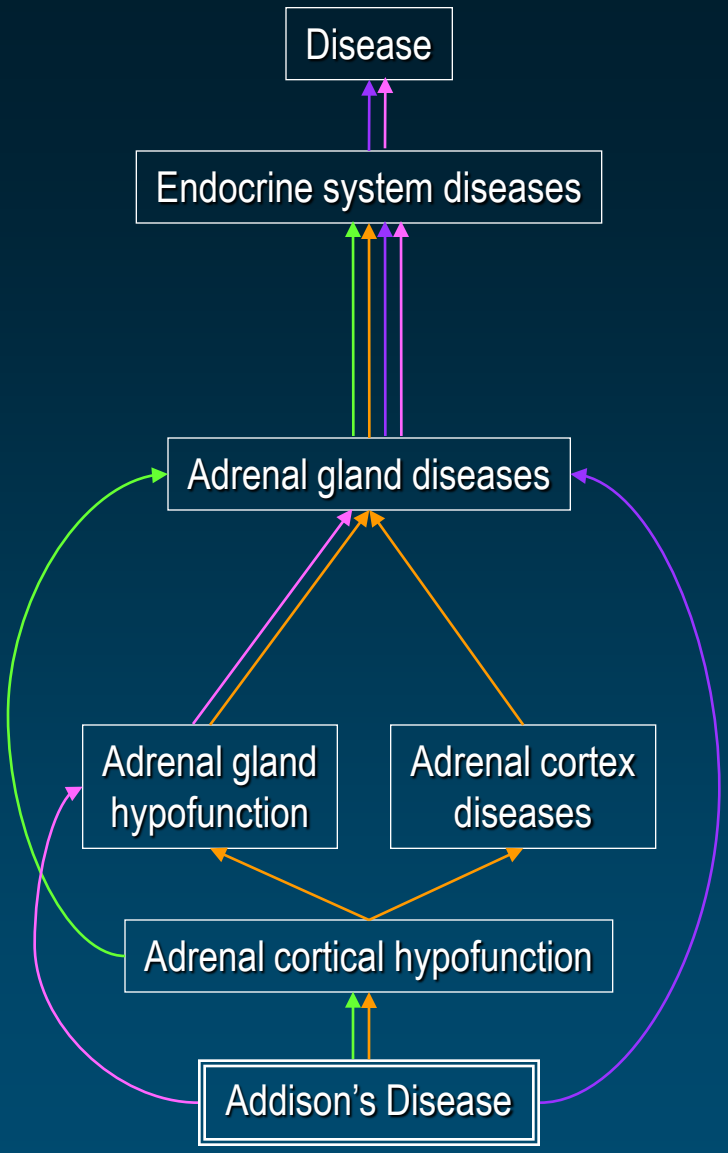

Endocrine, nutritional and metabolic diseases
Disorders of other endocrine glands
Other disorders of adrenal gland
Primary adrenocortical insufficiency

Organize concepts

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

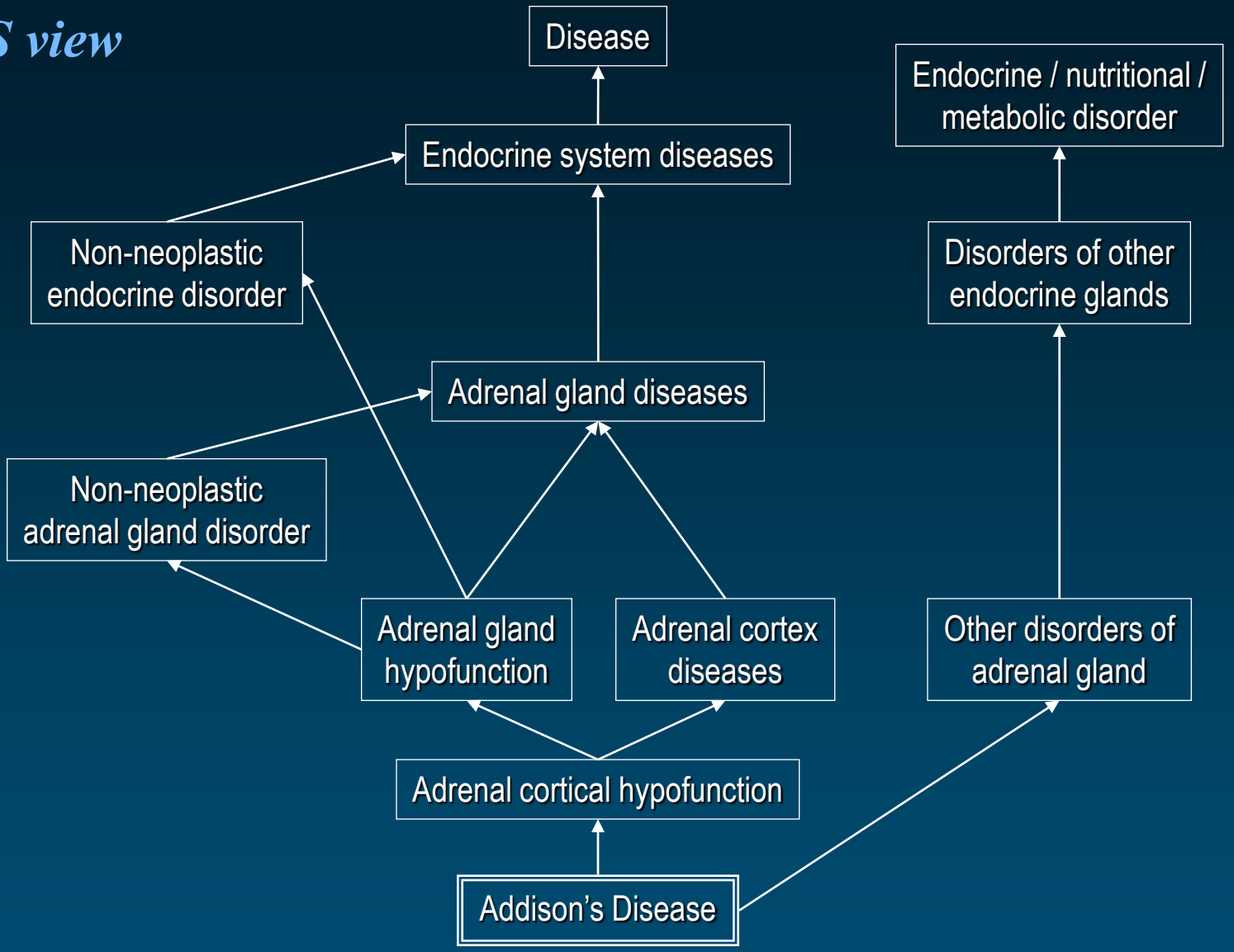


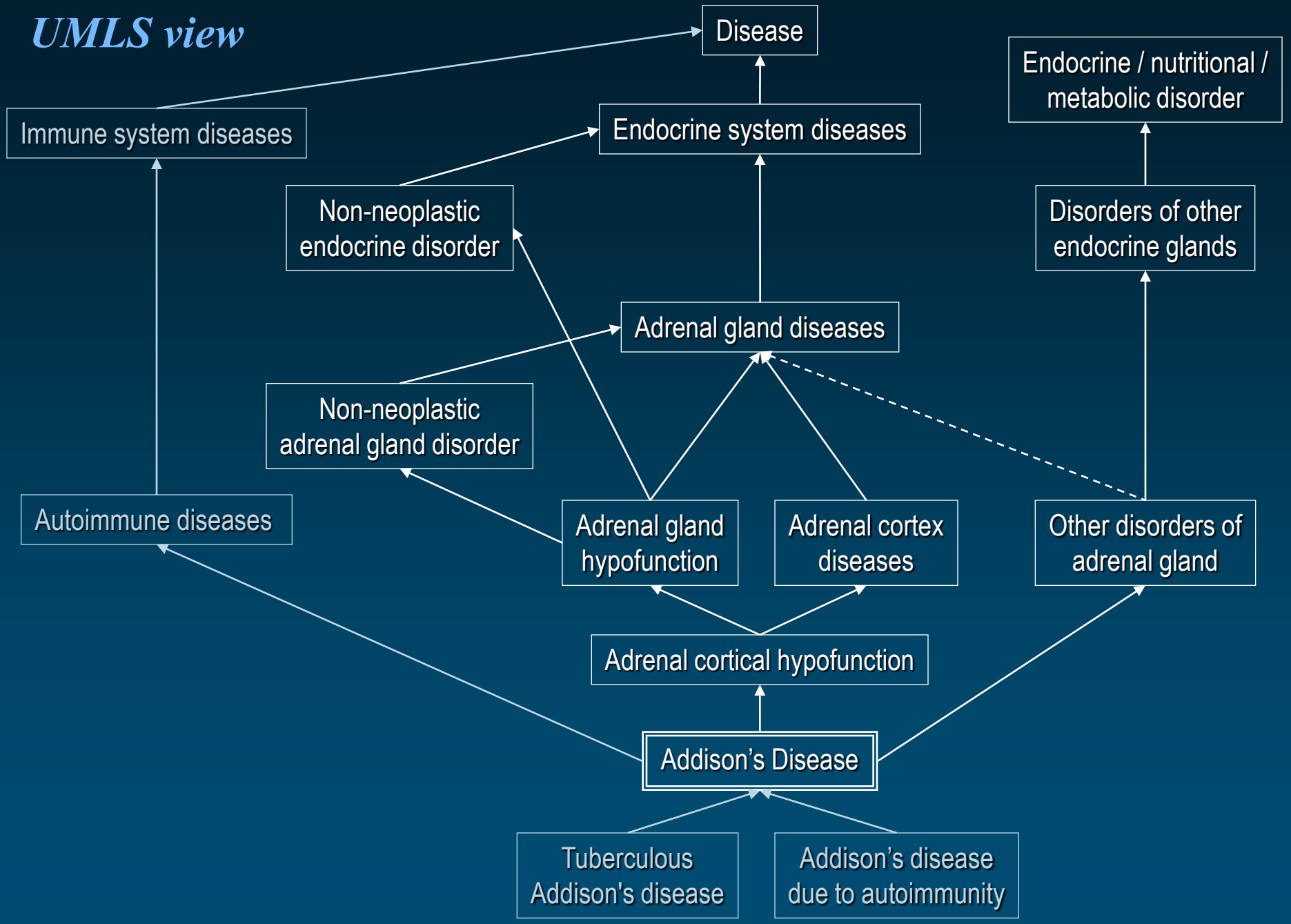
organize concepts



SNOMED CT
SNOMED Intl
MeSH
MedDRA

UMLS view


Disease
Endocrine / nutritional / metabolic disorder
Endocrine system diseases
Non-neoplastic endocrine disorder
Disorders of other endocrine glands
Adrenal gland diseases
Non-neoplastic adrenal gland disorder
Adrenal gland hypofunction
Adrenal cortex diseases
Other disorders of adrenal gland
Adrenal cortical hypofunction
Addison's Disease

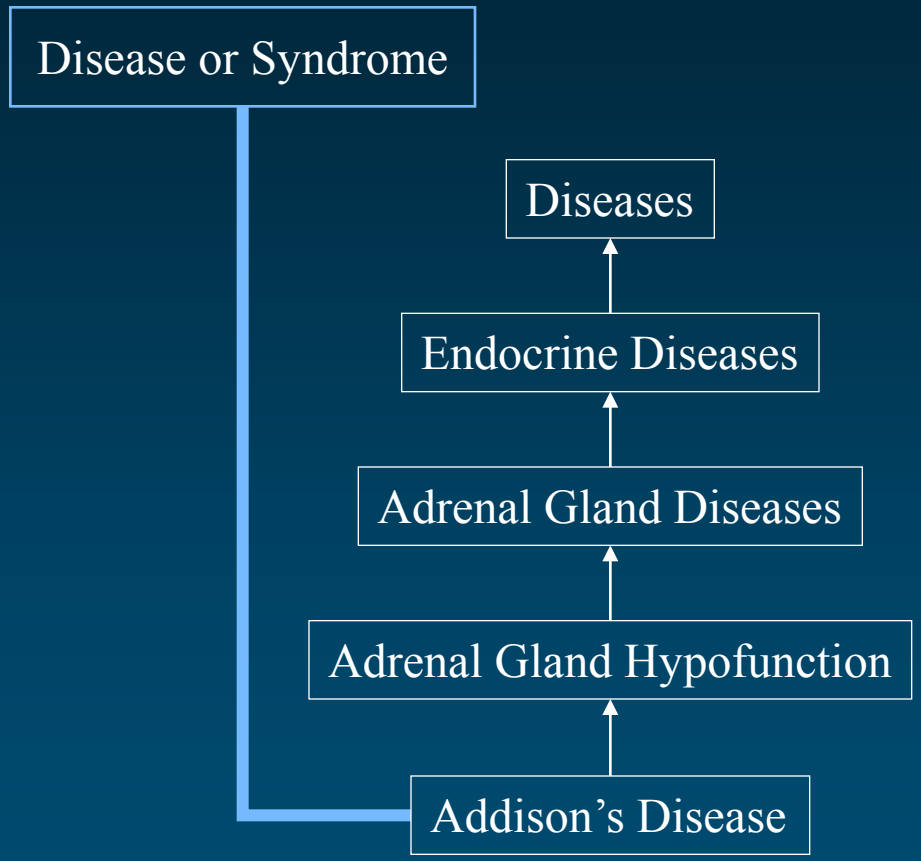

UMLS view
Disease
Immune system diseases
Endocrine system diseases
Endocrine / nutritional / metabolic disorder
Non-neoplastic endocrine disorder
Disorders of other endocrine glands
Adrenal gland diseases
Non-neoplastic adrenal gland disorder
Autoimmune diseases
Adrenal gland hypofunction
Adrenal cortex diseases
Other disorders of adrenal gland
Adrenal cortical hypofunction
Addison's Disease
Tuberculous Addison's disease
Addison's disease due to autoimmunity

Relate to other concepts

- Additional hierarchical relations
  - link to other trees
  - make relationships explicit
- Non-hierarchical relations
- Co-occurring concepts
- Mapping relations

Categorize concepts

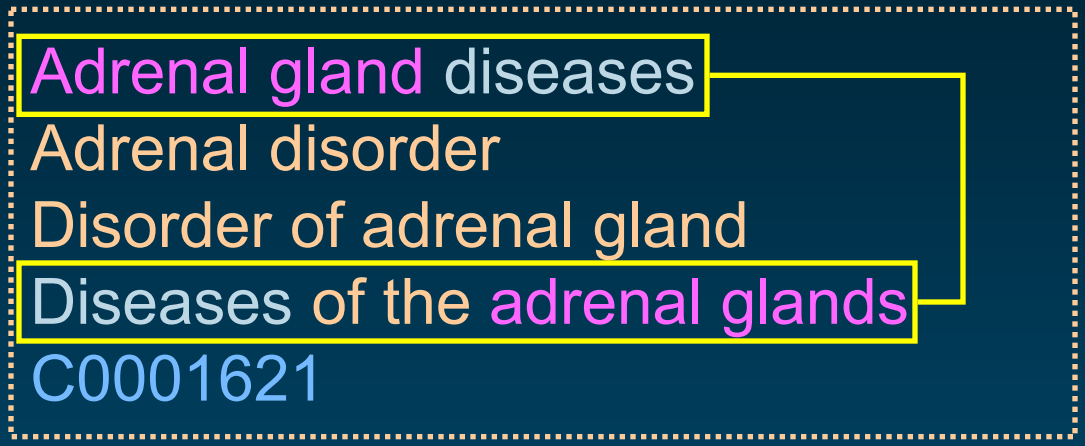
- High-level categories (semantic types)
- Assigned by the Metathesaurus editors
- Independently of the hierarchies in which these concepts are located



How do they do that?

- Lexical knowledge
- Semantic pre-processing
- UMLS editors

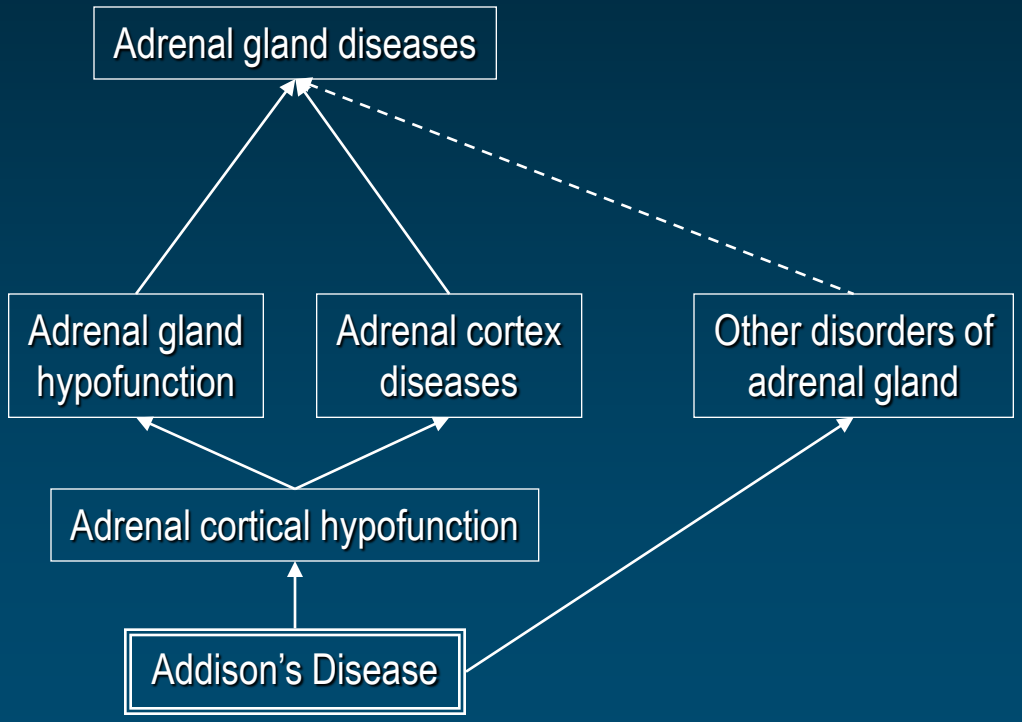
Lexical knowledge



Semantic pre-processing

- Metadata in the source vocabularies
- Tentative categorization
- Positive (or negative) evidence for tentative synonymy relations based on lexical features

Additional knowledge: UMLS editors



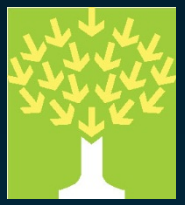
UMLS Summary

- Synonymous terms clustered into concepts
- Unique identifier

- Finer granularity
- Broader scope
- Additional hierarchical relationships
- Semantic categorization

UMLS Knowledge Sources

UMLS 3 components



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

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 (2014AA)

- 139 families of source vocabularies
  - Not counting translations
- 21 languages
- Broad coverage of biomedicine
  - 8.6M names (normalized)
  - ~3M concepts
  - >10M relations
- Common presentation

Biomedical terminologies

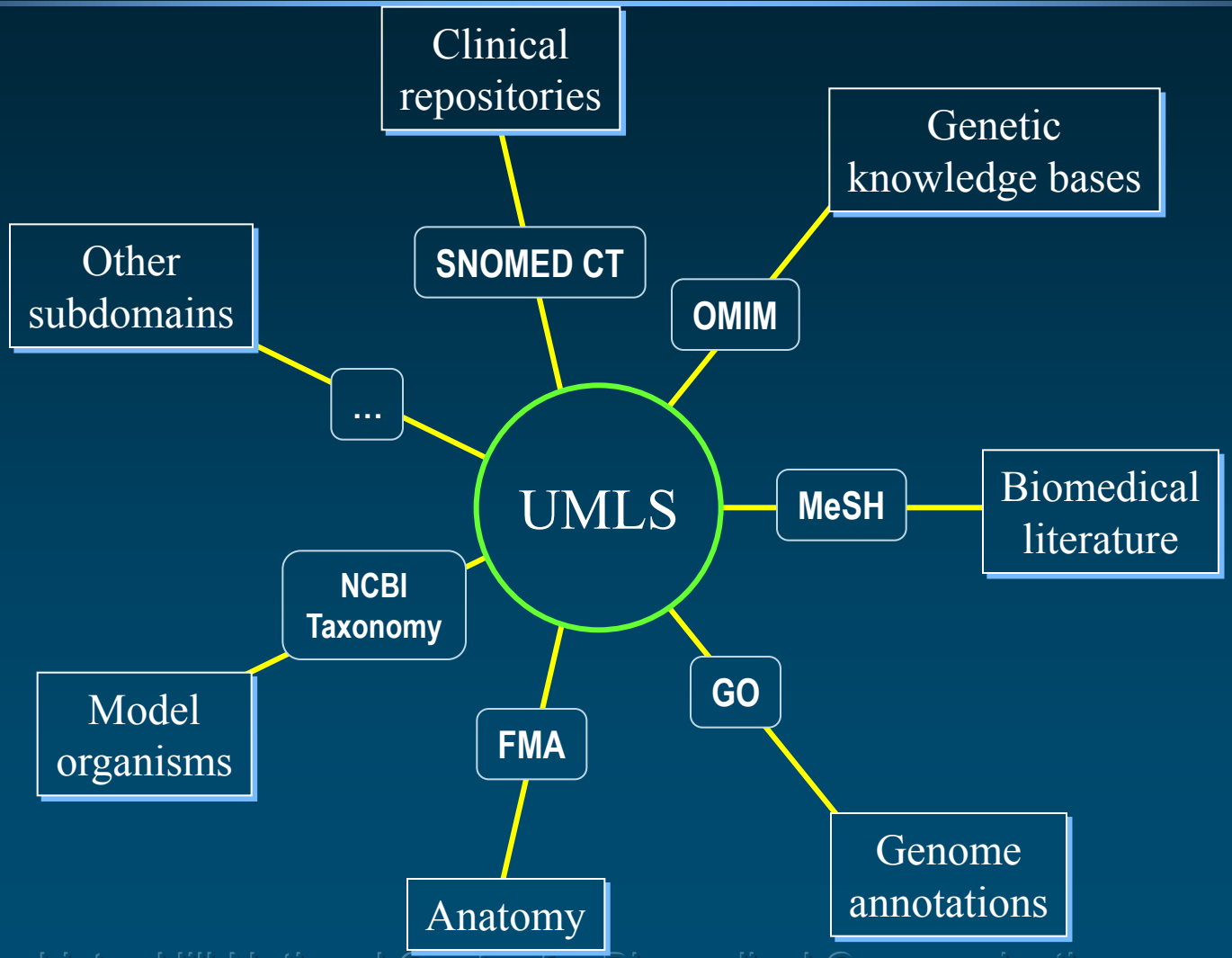
- General vocabularies
  - anatomy (FMA, Neuronames)
  - drugs (RxNorm, ATC, First DataBank, Micromedex)
  - medical devices (UMD, SPN)
- Several perspectives
  - clinical terms (SNOMED CT)
  - information sciences (MeSH)
  - administrative terminologies (ICD-9/10-CM, CPT-4)
  - data exchange terminologies (HL7, LOINC)

Biomedical terminologies (cont'd)

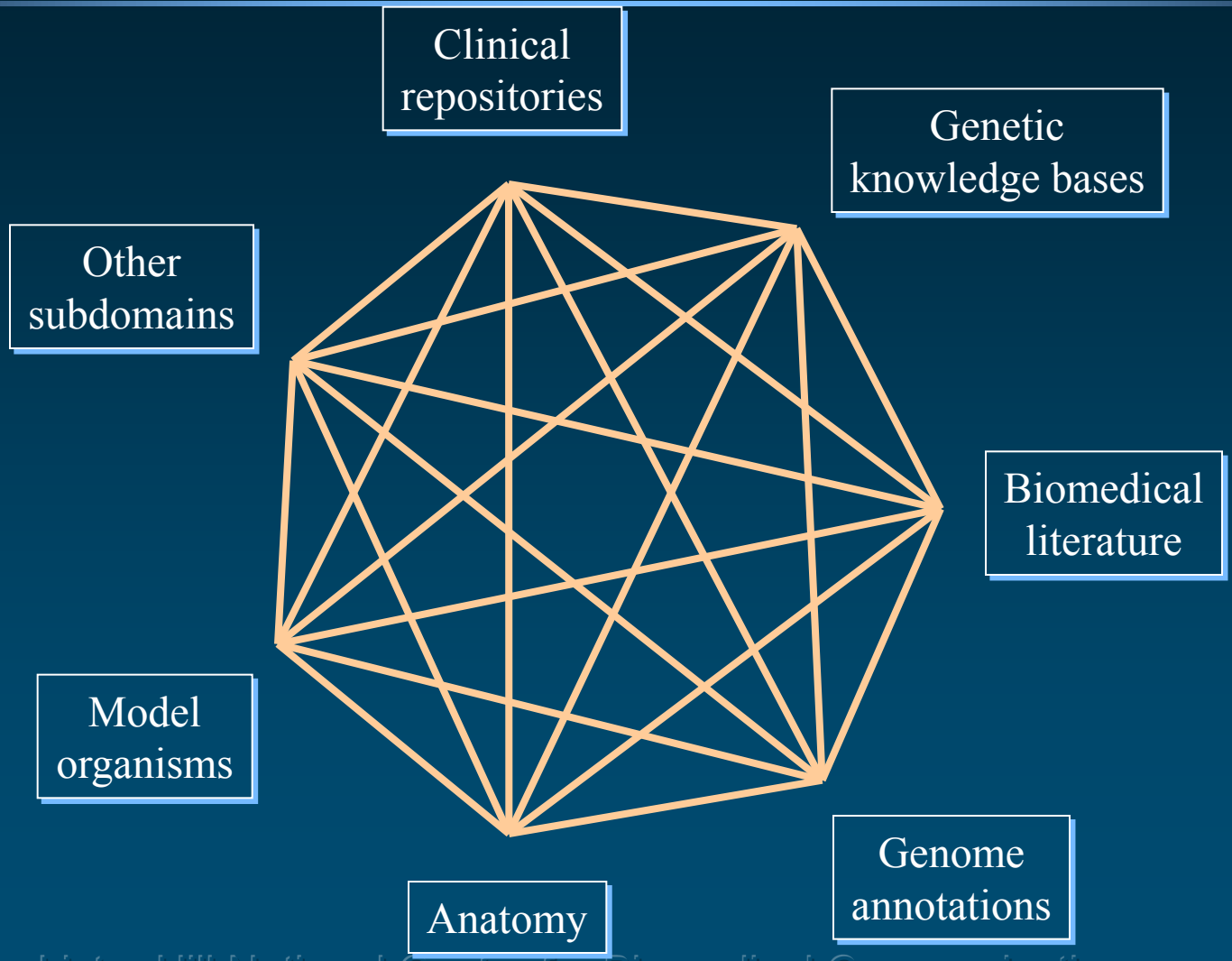
- Specialized vocabularies
  - nursing (NIC, NOC, NANDA, Omaha, ICNP)
  - dentistry (CDT)
  - oncology (PDQ)
  - psychiatry (DSM, APA)
  - adverse reactions (MedDRA, WHO ART)
  - primary care (ICPC)
- Terminology of knowledge bases (AI/Rheum, DXplain, QMR)

The UMLS serves as a vehicle for the regulatory standards (HIPAA, HITSP, Meaningful Use)

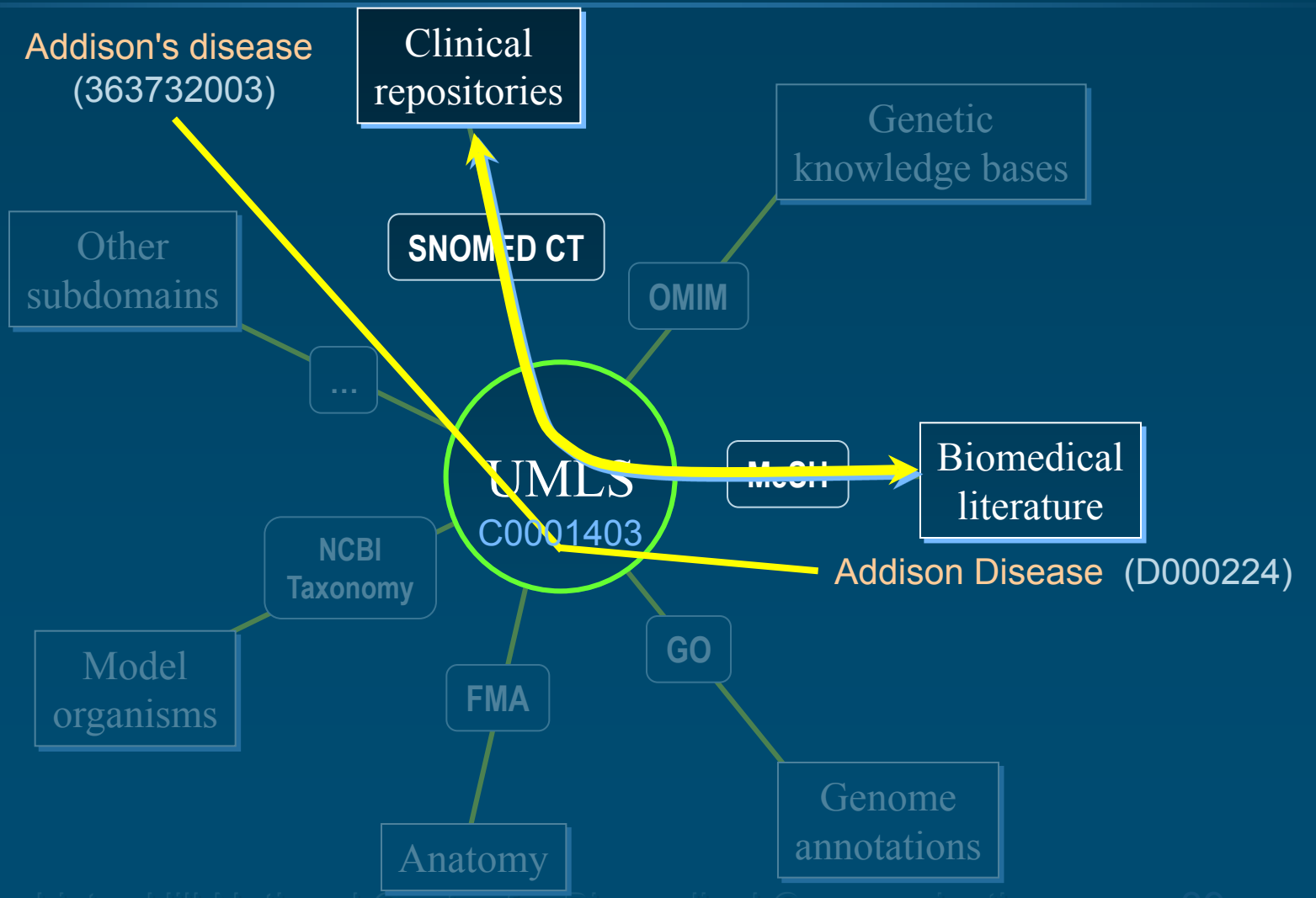
Integrating subdomains



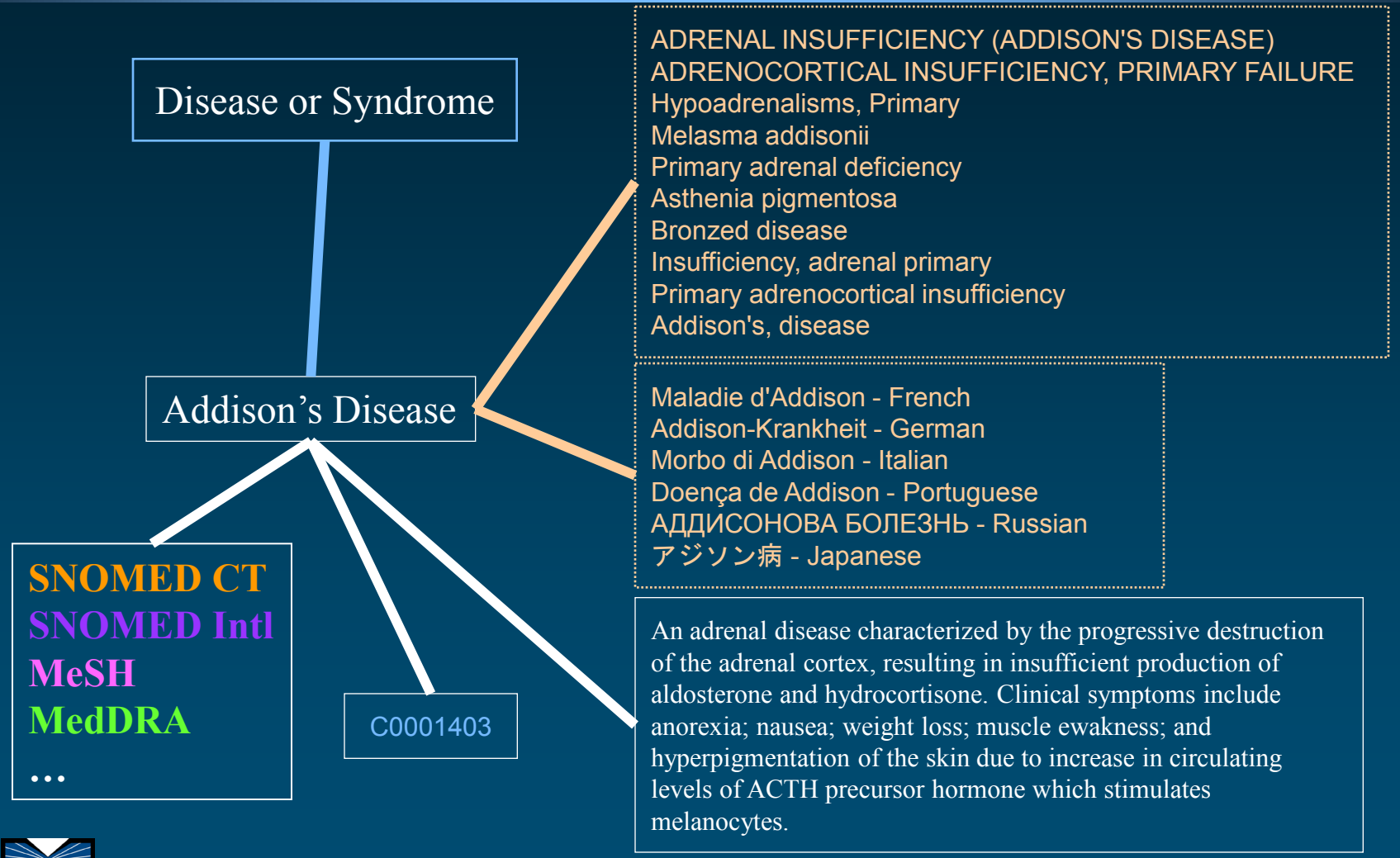
Integrating subdomains



Trans-namespace integration

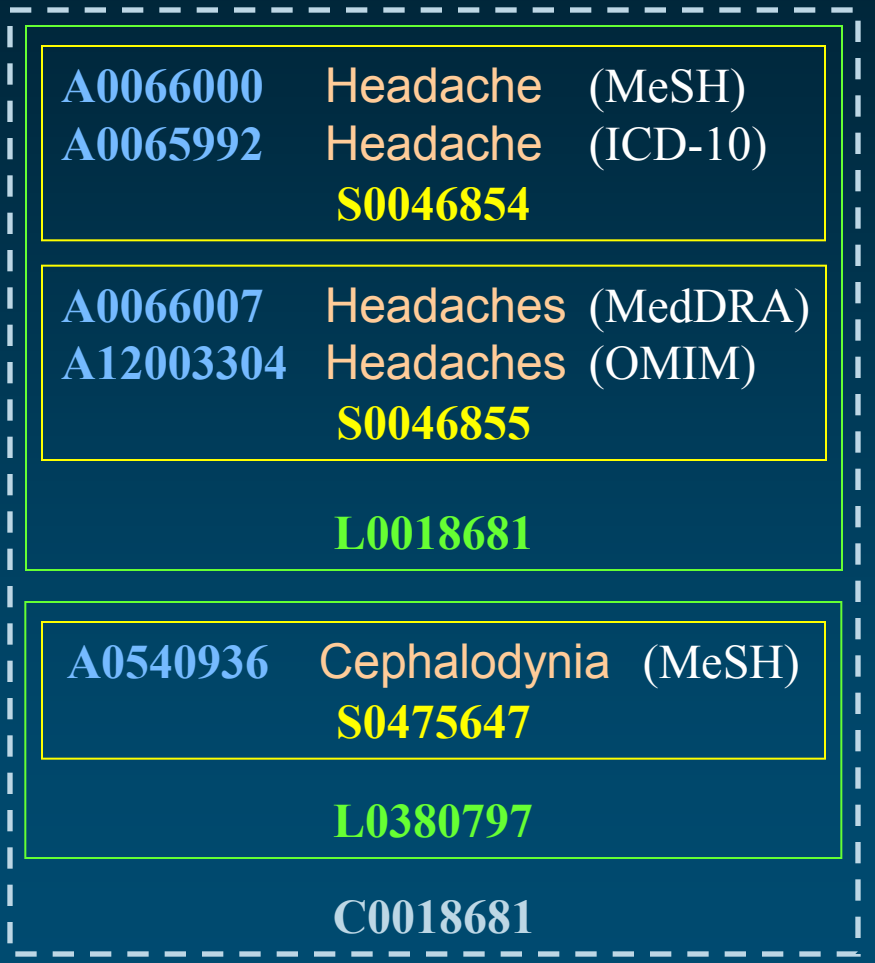


Addison's Disease: Concept



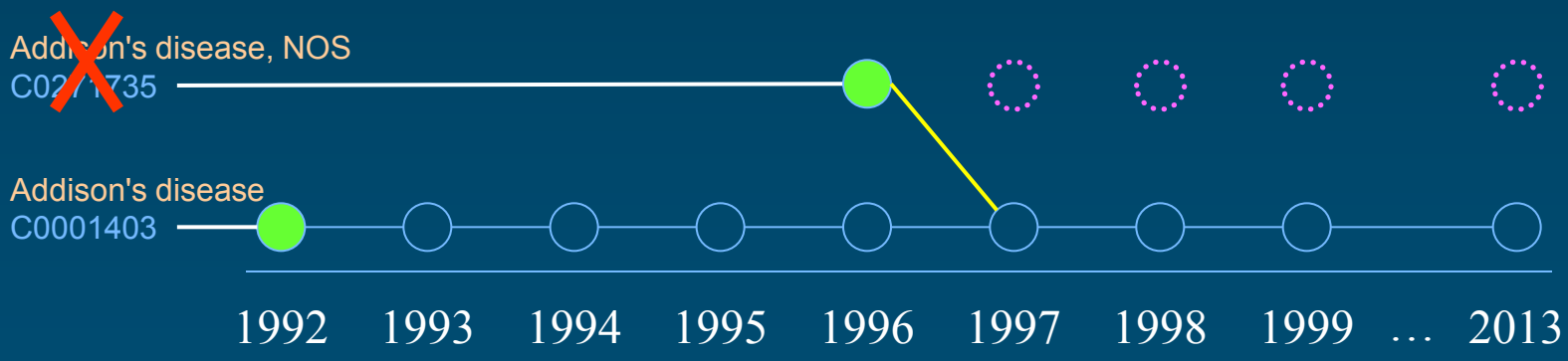
Metathesaurus Concepts (2014AA)

- Concept (2.9M) CUI
  - Set of synonymous concept names
- Term (8.6M) LUI
  - Set of normalized names
- String (9.7M) SUI
  - Distinct concept name
- Atom (11.6M) AUI
  - Concept name in a given source



Metathesaurus Evolution over time

- Concepts never die (in principle)
  - CUIs are permanent identifiers
- What happens when they do die (in reality)?
  - Concepts can merge or split
  - Resulting in new concepts and deletions



Metathesaurus Relations

- 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

Symbolic relations

- Relation
  - Pair of "atom" identifiers
  - Type
  - Attribute (if any)
  - List of sources (for type and attribute)
- Semantics of the relationship: defined by its type [and attribute]

Source transparency: the information is recorded at the "atom" level

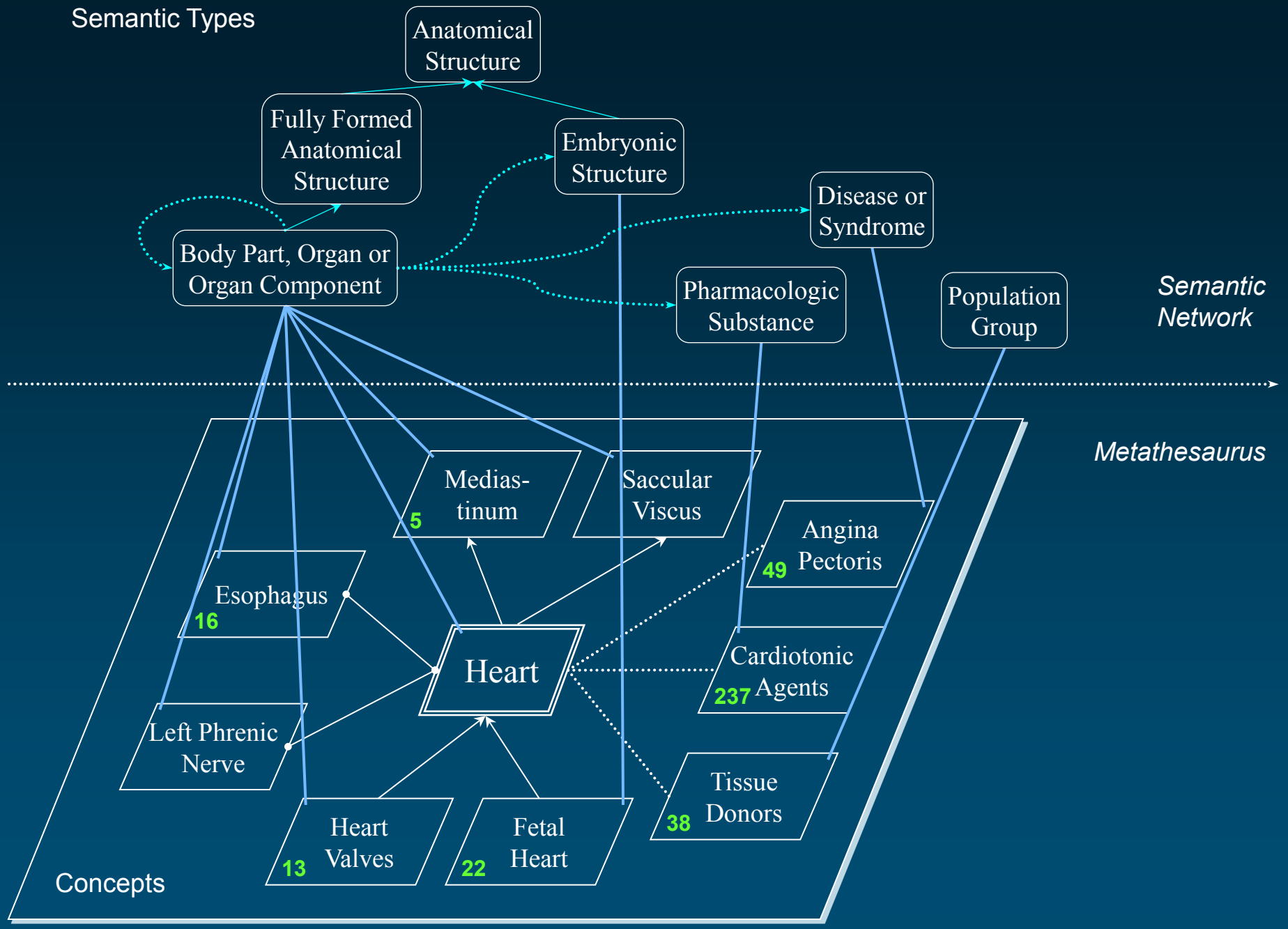
Mapping relations

- Simple mappings
  - <atom 1> mapped_to <atom 2>
  - e.g.,
    - SNOMED CT to ICD-9-CM
- Complex mappings
  - <atom 1> mapped_to <boolean expression>
  - e.g.,
    - ICD-9-CM to MeSH (search strategies)

NB: partially redundant with relations in MRREL

Everything else

- Source-specific attributes (MRSAT)
  - Legacy identifiers, external cross-references
    - SNOMED International legacy codes (SNOMED CT)
    - RxNorm to NDC
  - Concept status in a particular source (SNOMED CT)
  - Frequency of occurrence in MEDLINE (MeSH)
  - MedlinePlus URL (MeSH)
  - …
- Co-occurrence information
  - Co- occurrence of MeSH descriptors in MEDLINE for the most part
  - No longer part of the UMLS Metathesaurus


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 Semantic Network

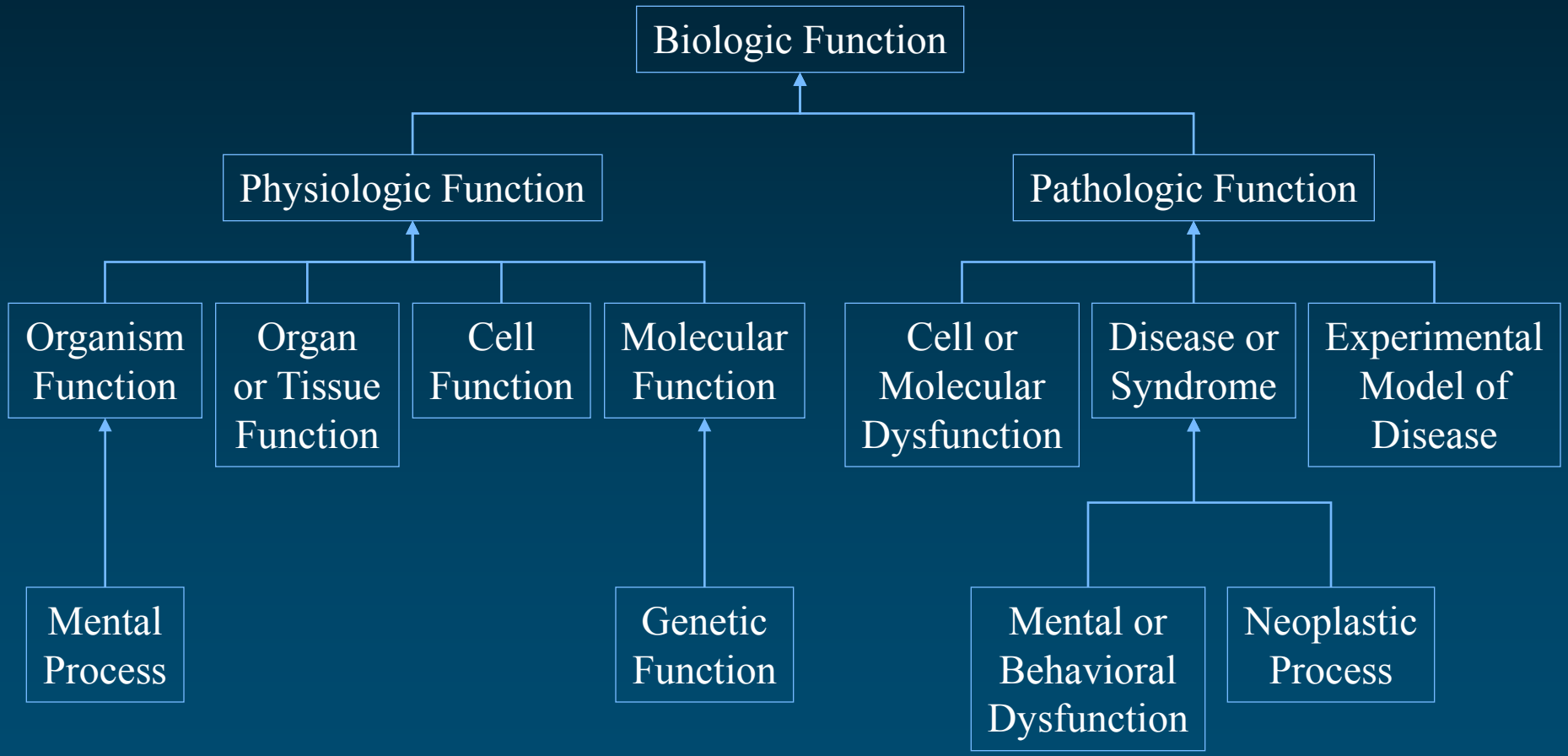
Semantic Network

- Semantic types (133)
  - tree structure
  - 2 major hierarchies
    - Entity
      - Physical Object
      - Conceptual Entity
    - Event
      - Activity
      - Phenomenon or Process

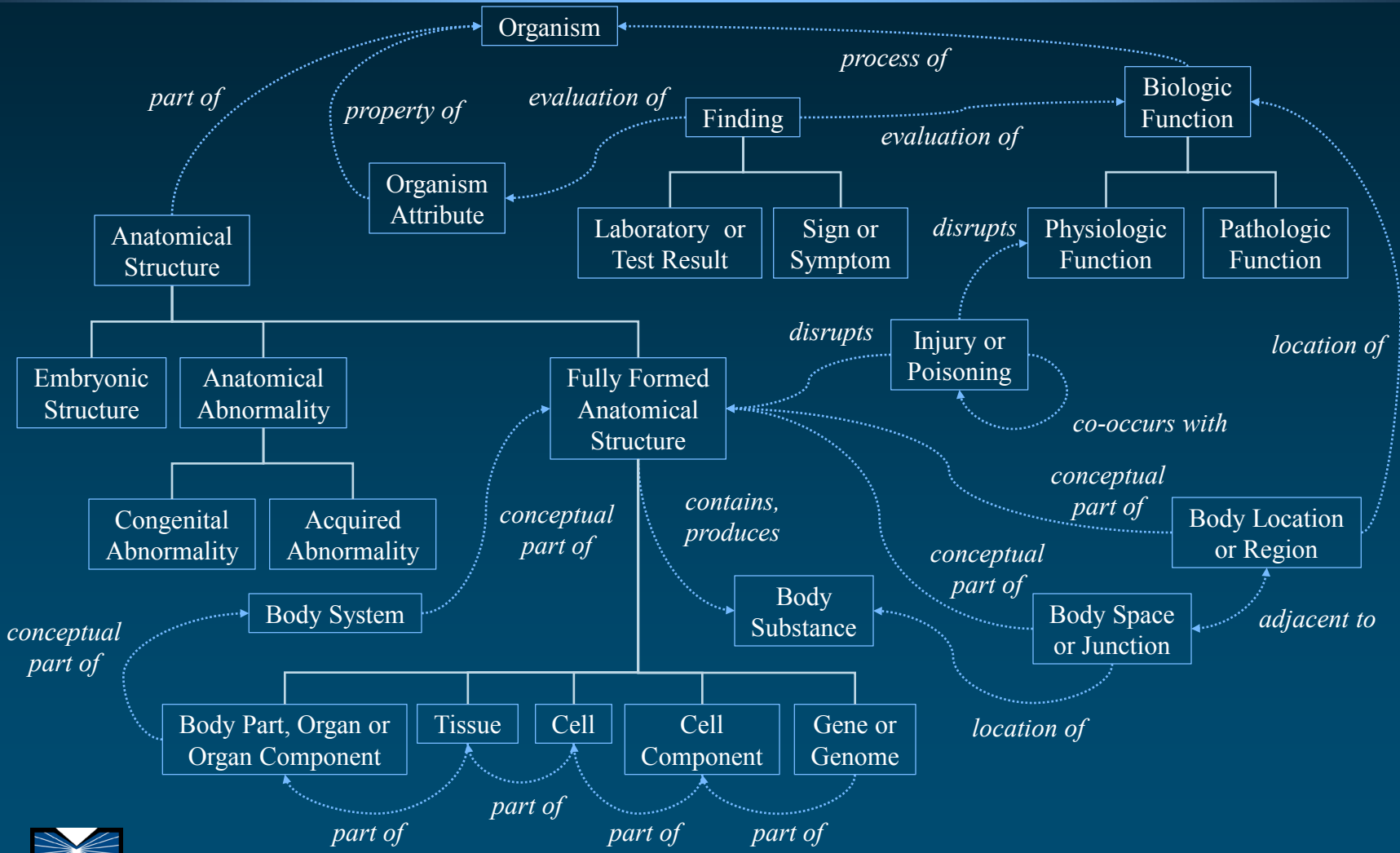
Semantic Network

- Semantic network
  - 54 relationships
  - 603 asserted relations
  - 6101 inferred relations
- Asserted semantic network relations (603)
  - hierarchical (isa = is a kind of)
    - among types (133)
      - Animal *isa* Organism
      - Enzyme *isa* Biologically Active Substance
    - among relations (54)
      - treats *isa* affects
  - non-hierarchical (416)
    - Sign or Symptom *diagnoses* Pathologic Function
    - Pharmacologic Substance *treats* Pathologic Function

“Biologic Function” hierarchy (isa)



Associative (non-isa) relationships

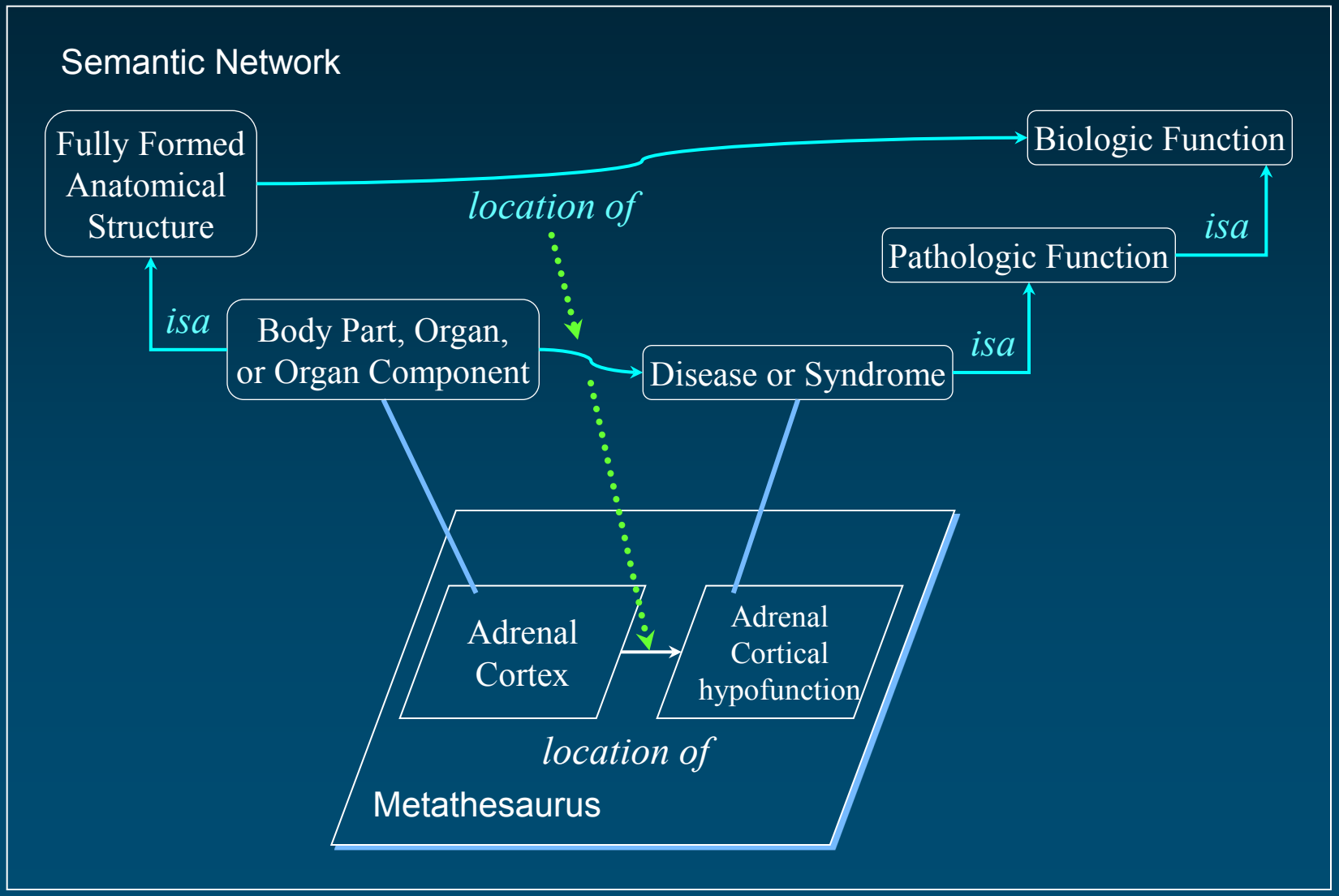


Why a semantic network?

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

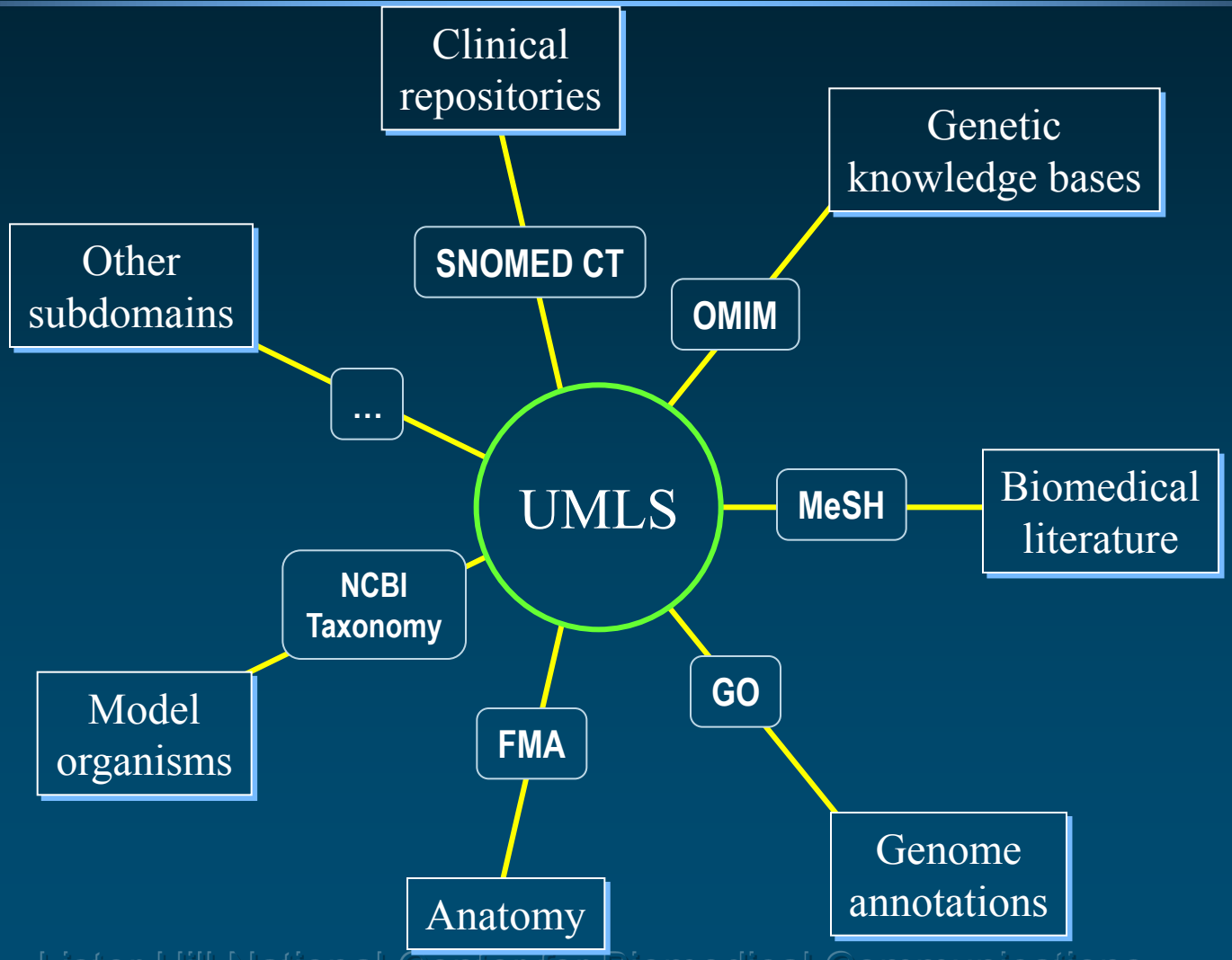
- A relationship between 2 Semantic Types (ST) is a possible link between 2 concepts that have been assigned to those STs
  - The relationship may or may not hold at the concept level
  - Other relationships may apply at the concept level

Relationships *may* inherit semantics



Conclusions

Integrating subdomains



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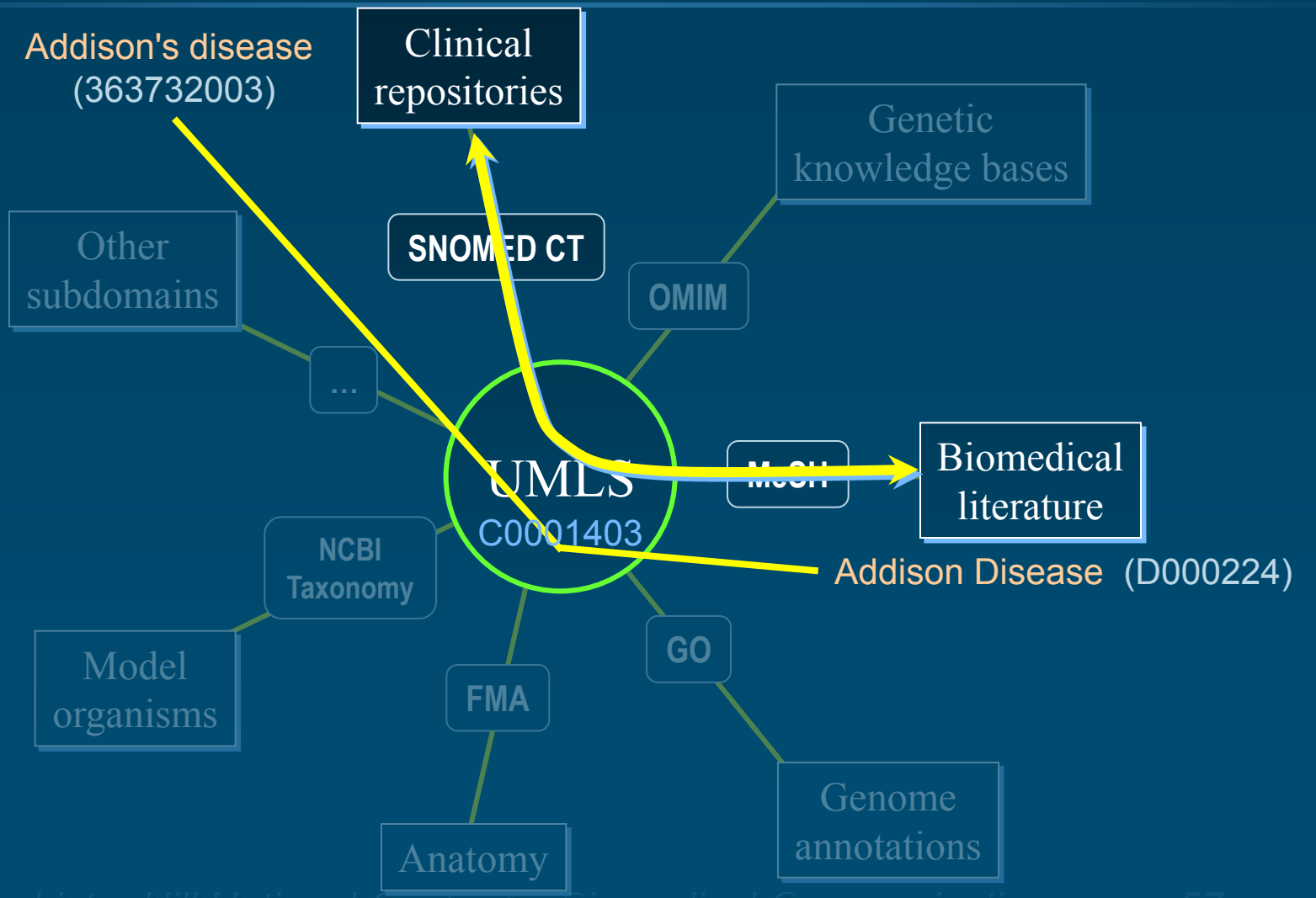
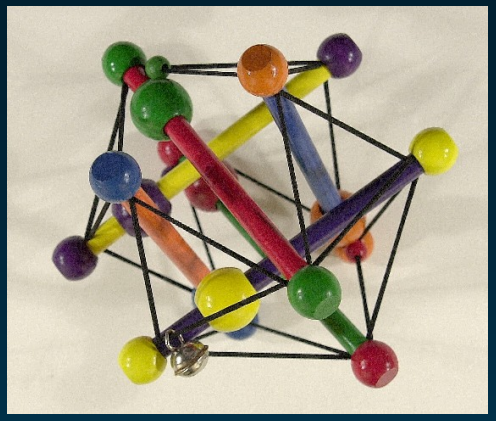


Other things you would need to know

- UMLS license agreement
  - http://wwwcf.nlm.nih.gov/umlslicense/snomed/license.cfm
- MetamorphoSys
  - http://www.nlm.nih.gov/research/umls/mmsys_doc.html
- UMLS Terminology Services (UTS)
  (formerly, UMLS Knowledge Source Server)
  - https://uts.nlm.nih.gov/



Medical Ontology Research

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Web: mor.nlm.nih.gov

NATIONAL LIBRARY OF MEDICINE

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References

References: UMLS home page

- UMLS home page
  - http://www.nlm.nih.gov/research/umls/
- UMLS documentation
  - Formerly know as the "Green Book"
  - Now online documentation
  - http://www.nlm.nih.gov/research/umls/UMLSDOC.HTML
- UMLS online tutorials
  - http://www.nlm.nih.gov/research/umls/online%20learning/index.htm

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  - 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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  - Bodenreider O. & McCray A. T. (2003). Exploring semantic groups through visual approaches. *Journal of Biomedical Informatics*, 36(6), 414-432.