Manufacturing Engineering Laboratory
NIST
National Institute of Standards and Technology

July 24, 2006

Ontologies for Data Integration:
The Unified Medical Language System

NATIONAL LIBRARY OF MEDICINE

Olivier Bodenreider

Lister Hill National Center
for Biomedical Communications
Bethesda, Maryland - USA

Outline

- From terminology integration
  to information integration
  Unified Medical Language System (UMLS)

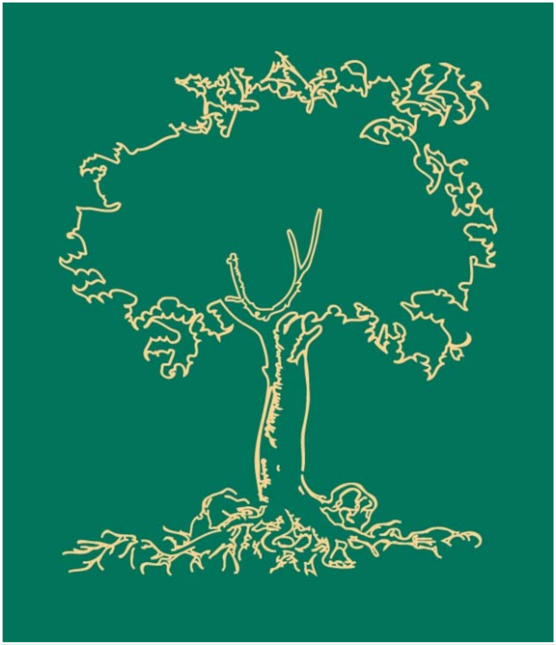
- UMLS in use:
  Mapping across terminologies

From terminology integration to information integration

Unified Medical Language System (UMLS)

What does UMLS stand for?

- Unified
- Medical
- Language
- System



UMLS®
Unified Medical Language System®
UMLS Metathesaurus®

Motivation

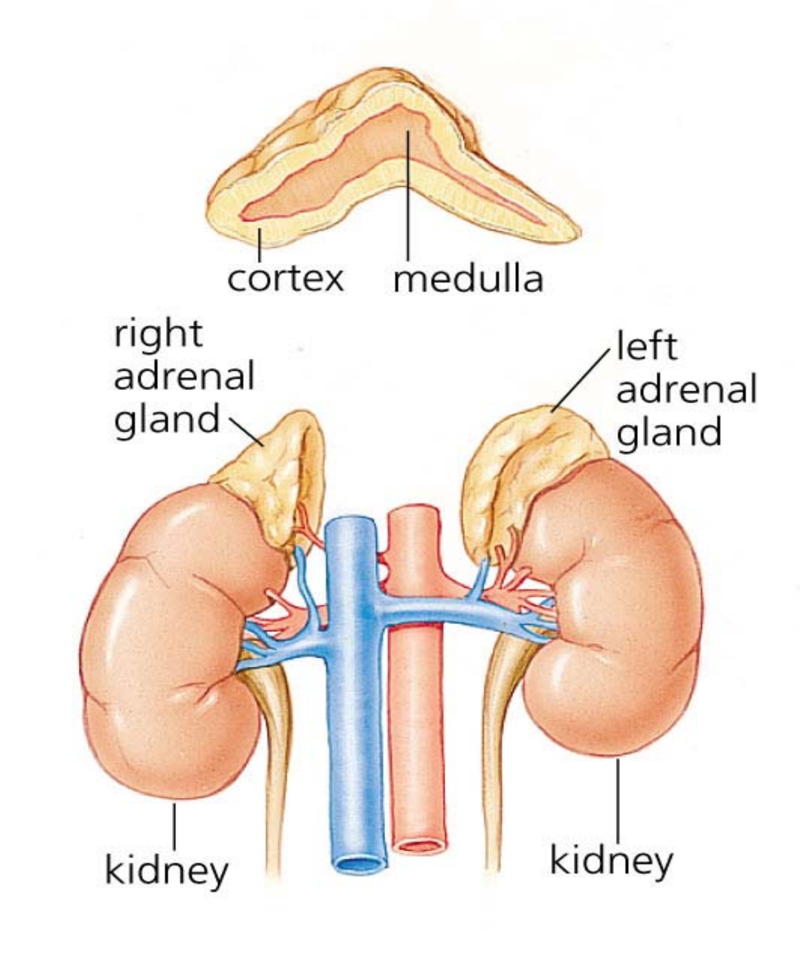
- Started in 1986
- National Library of Medicine
- “Long-term R&D project”

> «[…] 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.»

Overview through an example

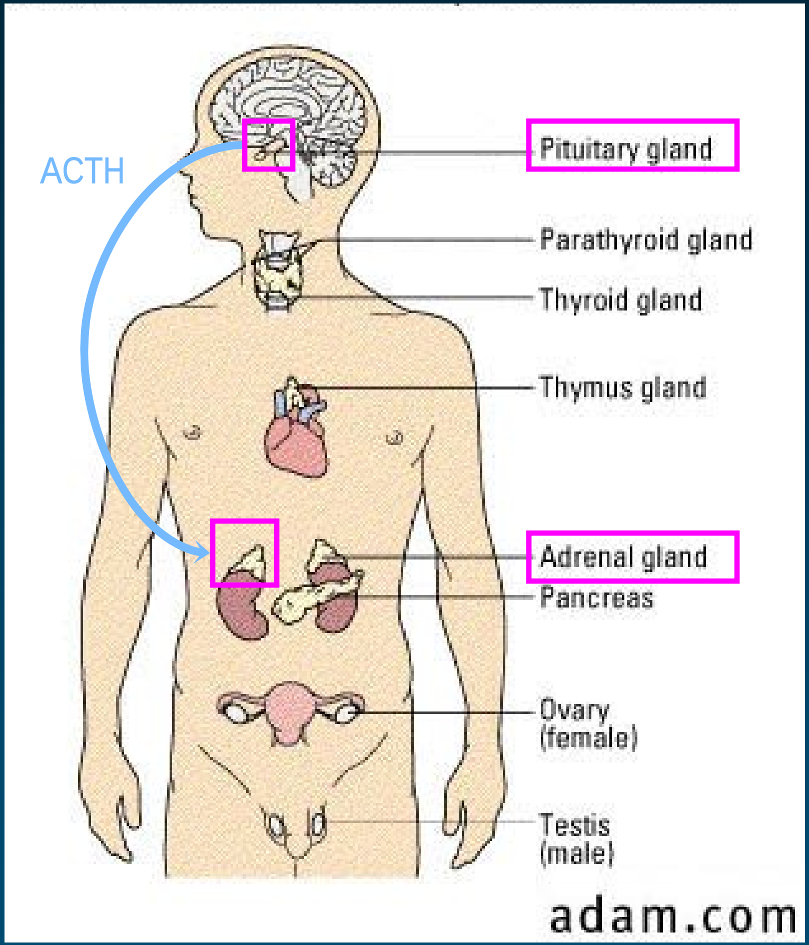
Addison’s disease

- 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



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

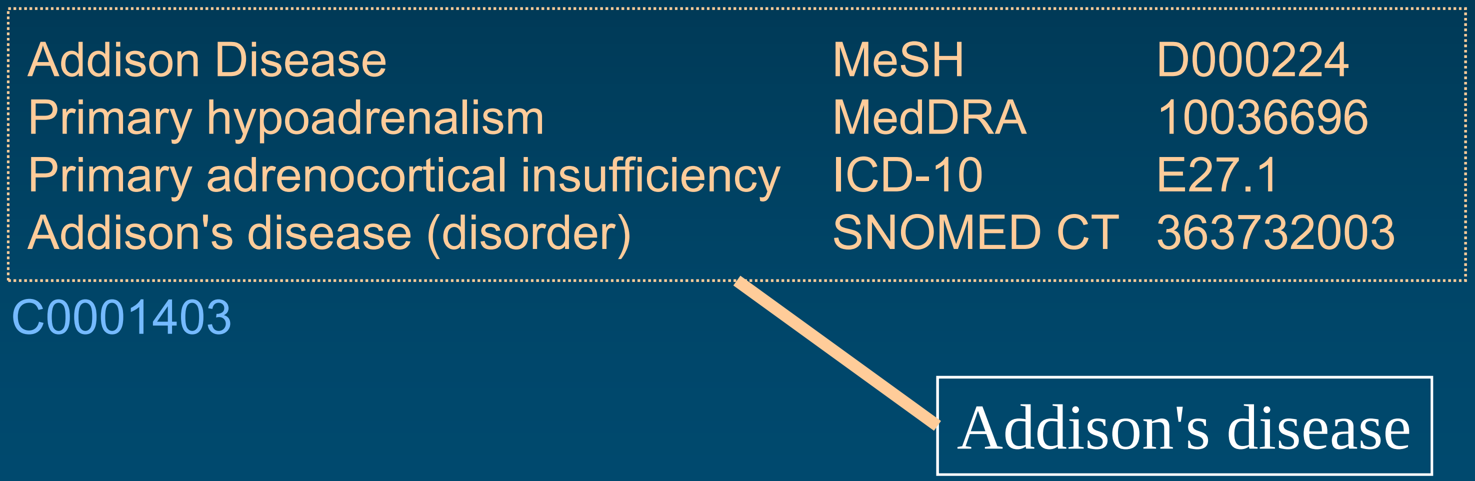
- Synonyms: different terms

• Addisonian syndrome	eponym
• Bronzed disease	symptoms
• Addison melanoderma	
• Asthenia pigmentosa	
• Primary adrenal deficiency	clinical variants
• Primary adrenal insufficiency	
• Primary adrenocortical insufficiency	
• Chronic adrenocortical insufficiency	

- Contexts: different hierarchies

Organize terms

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

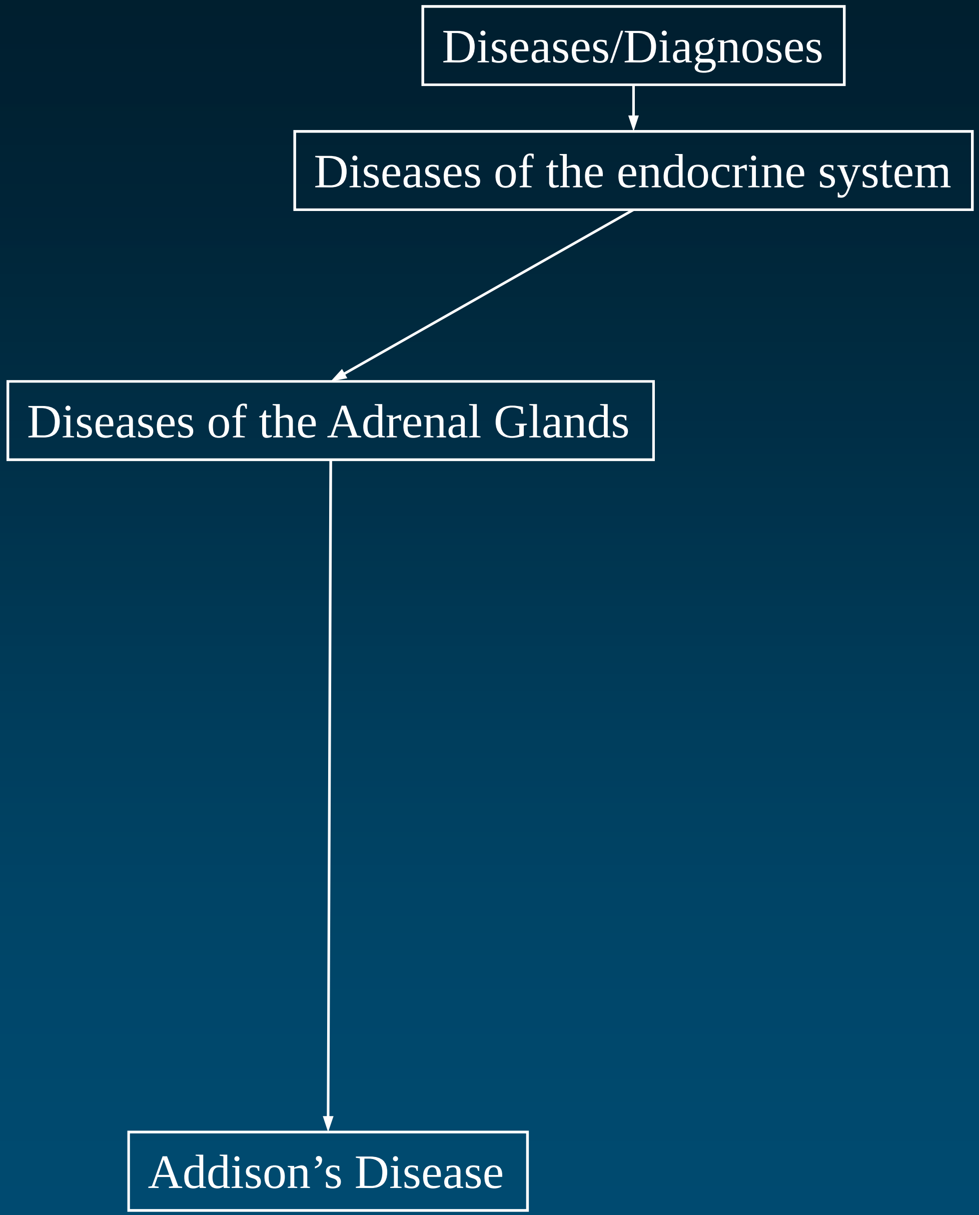


Addison Disease	MeSH	D000224
Primary hypoadrenalism	MedDRA	10036696
Primary adrenocortical insufficiency	ICD-10	E27.1
Addison's disease (disorder)	SNOMED CT	363732003

C0001403

Addison's disease

SNOMED International



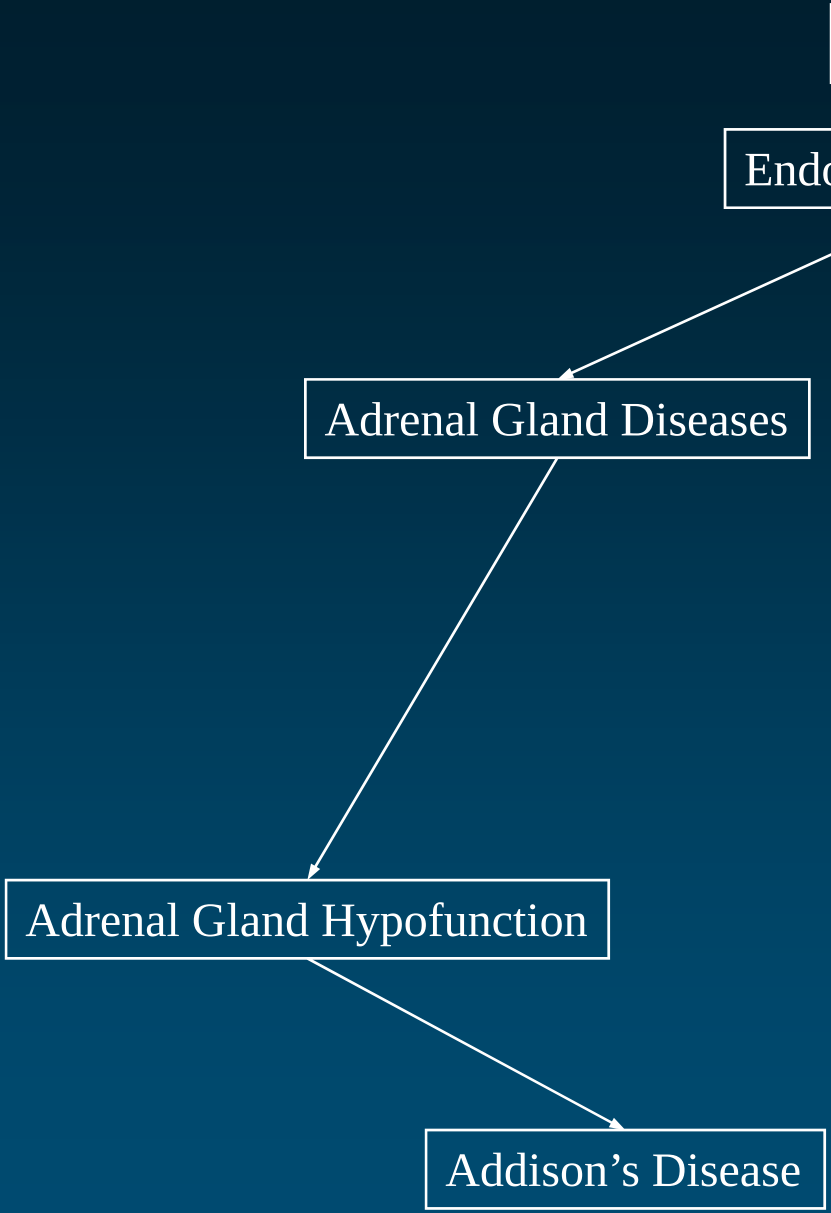
Diseases/Diagnoses

Diseases of the endocrine system

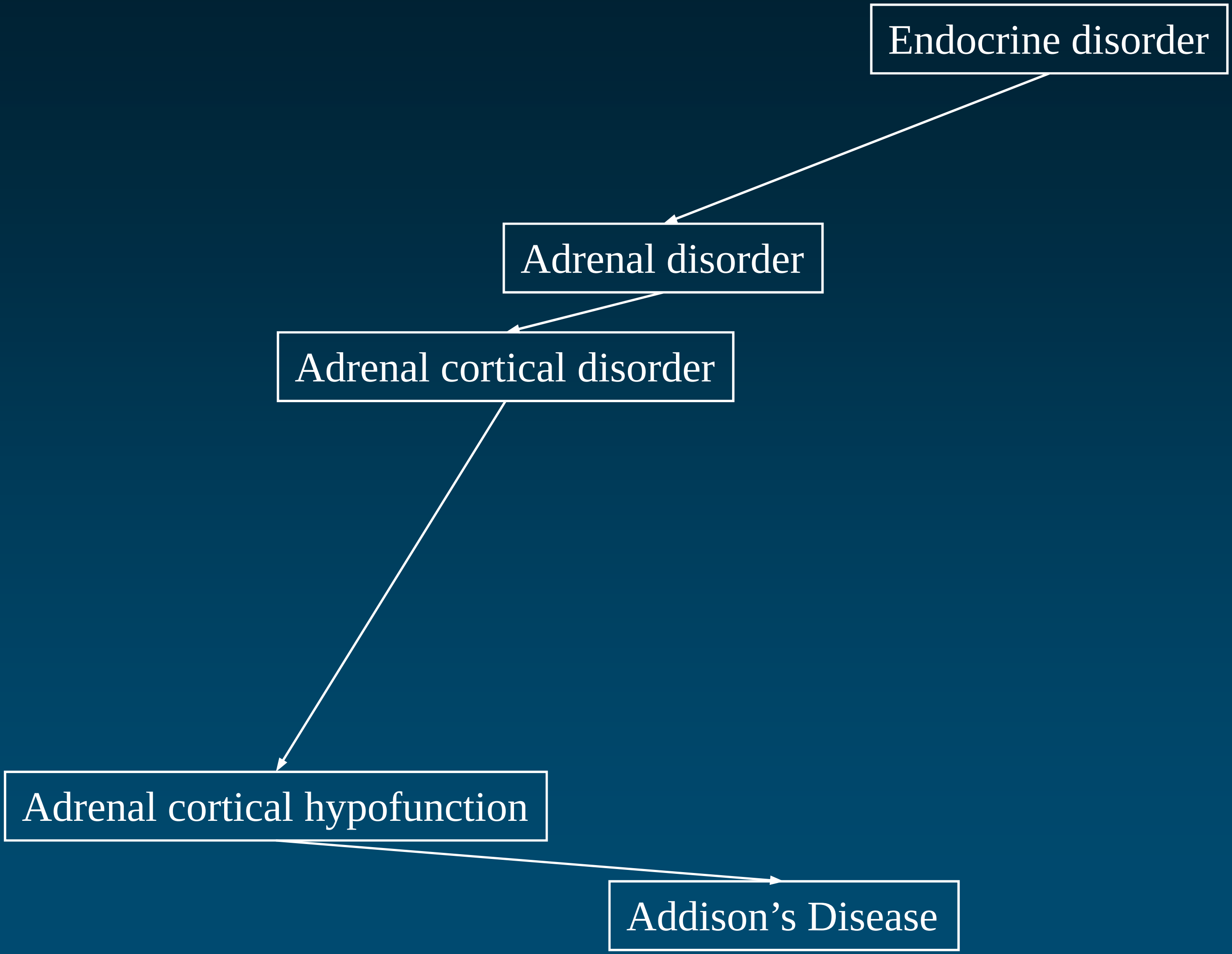
Diseases of the Adrenal Glands

Addison’s Disease

MeSH


Diseases
Endocrine Diseases
Adrenal Gland Diseases
Adrenal Gland Hypofunction
Addison’s Disease

AOD



Endocrine disorder

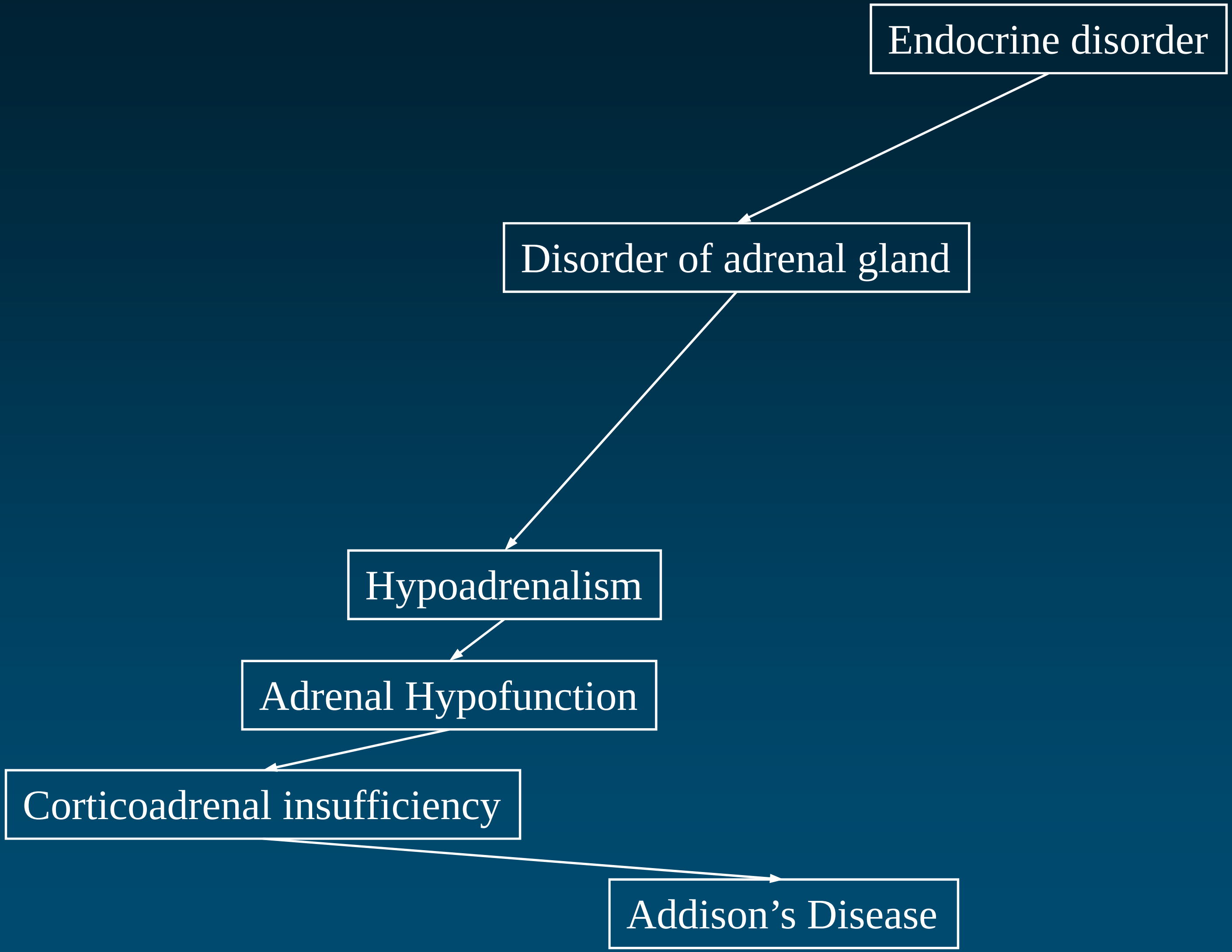
Adrenal disorder

Adrenal cortical disorder

Adrenal cortical hypofunction

Addison’s Disease

Read Codes



Endocrine disorder

Disorder of adrenal gland

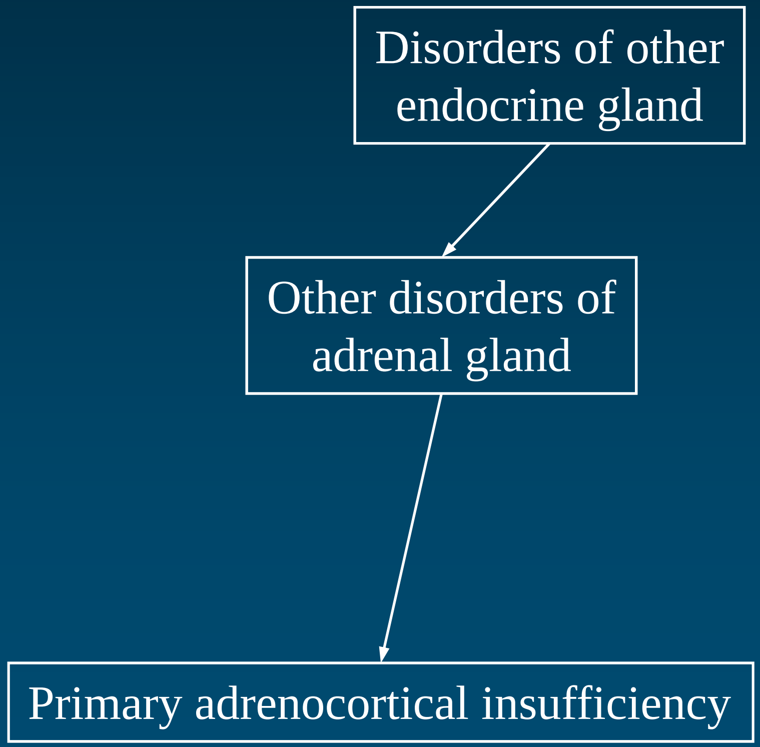
Hypoadrenalism

Adrenal Hypofunction

Corticoadrenal insufficiency

Addison’s Disease

ICD-10



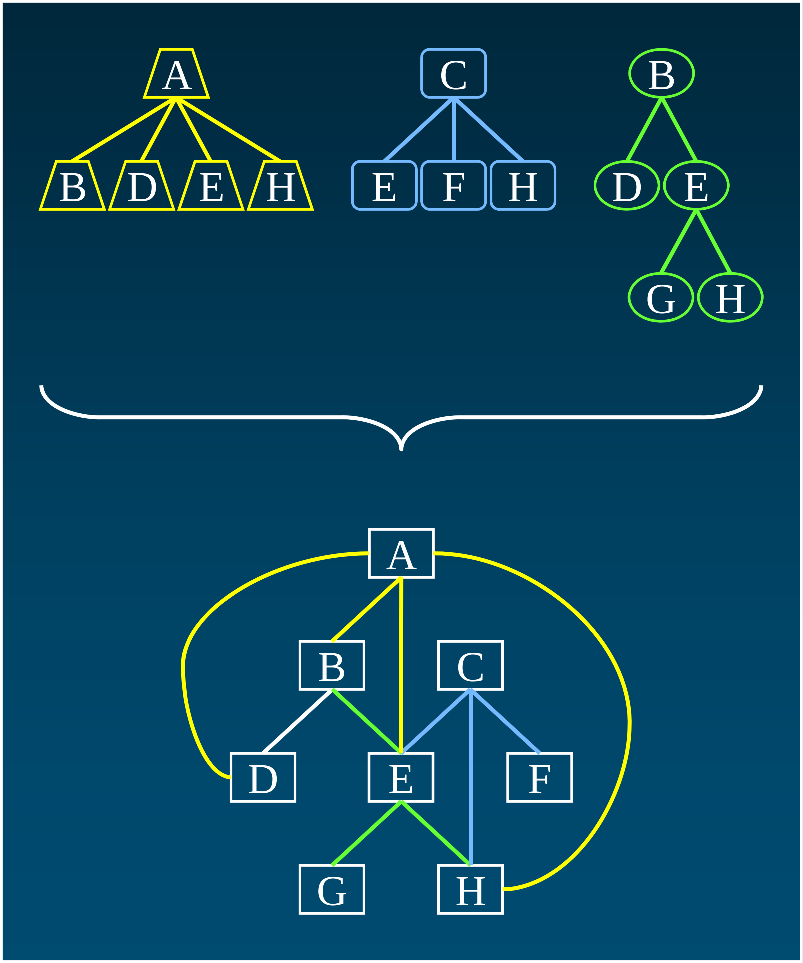
Disorders of other endocrine gland

Other disorders of adrenal gland

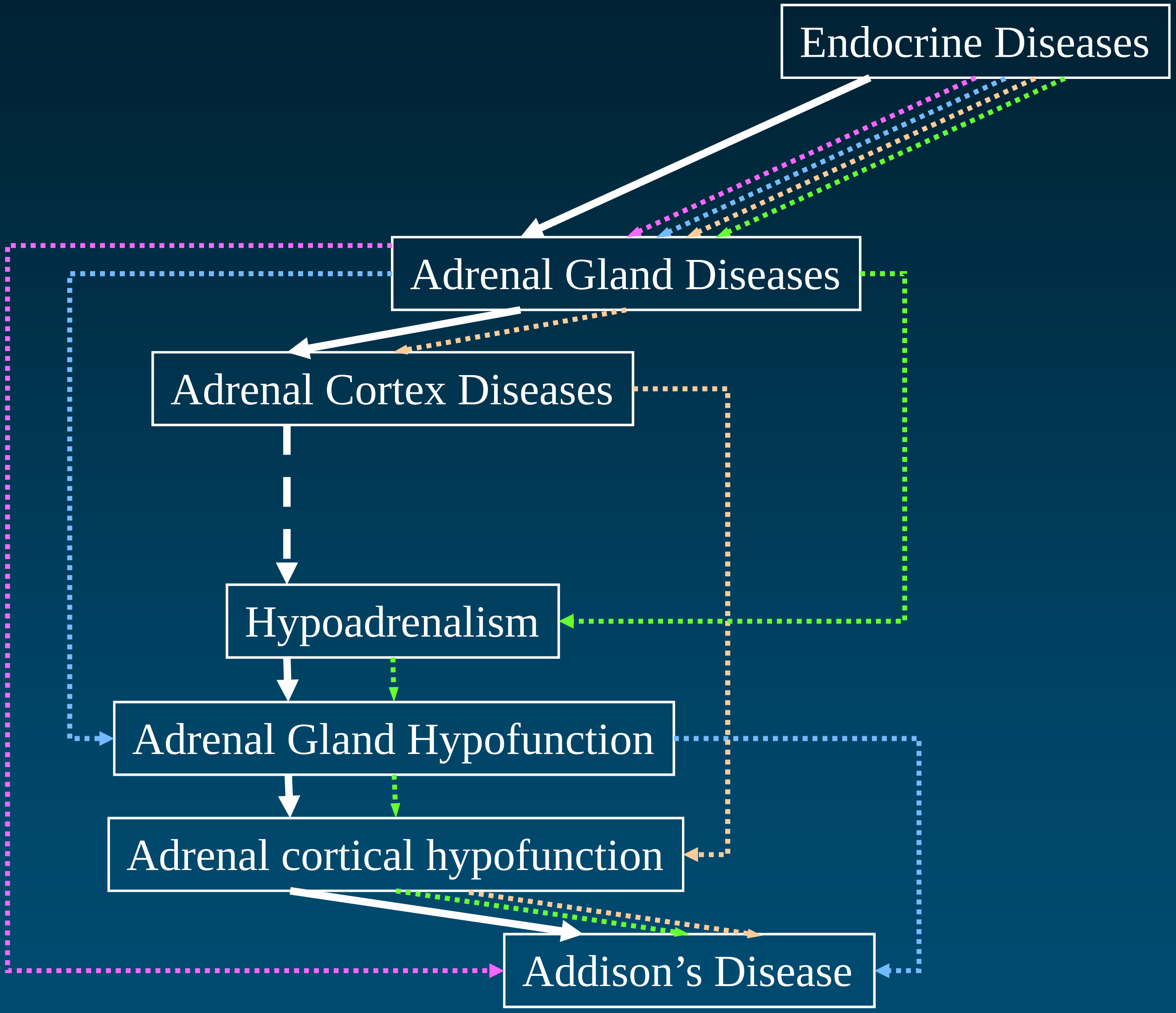
Primary adrenocortical insufficiency

Organize concepts

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



organize concepts



Endocrine Diseases

Adrenal Gland Diseases

Adrenal Cortex Diseases

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

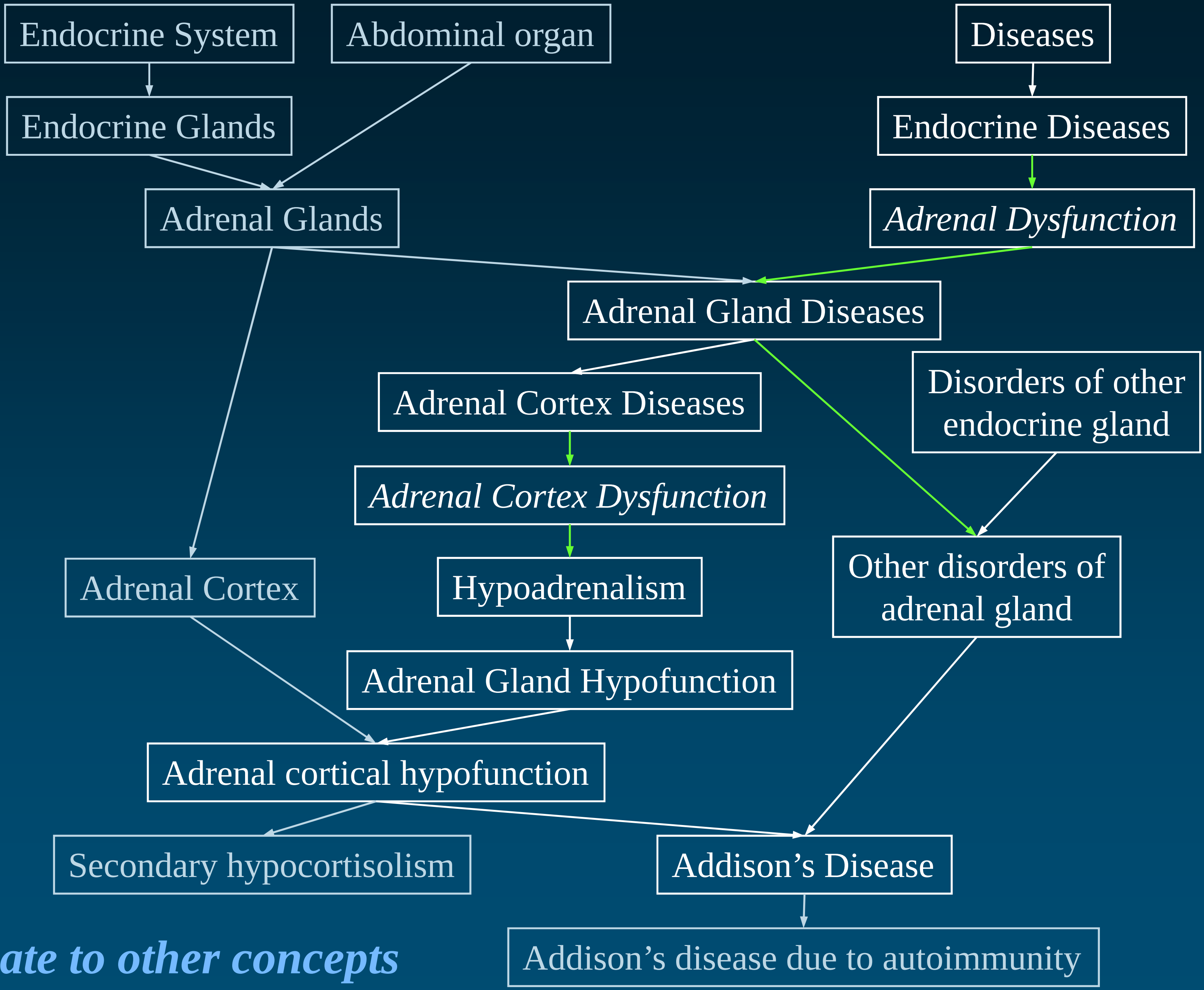
Addison’s Disease

SNOMED
MeSH
AOD
Read Codes

UMLS

Relate to other concepts

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



Endocrine System

Abdominal organ

Diseases

Endocrine Glands

Endocrine Diseases

Adrenal Glands

Adrenal Dysfunction

Adrenal Gland Diseases

Adrenal Cortex Diseases

Disorders of other endocrine gland

Adrenal Cortex Dysfunction

Adrenal Cortex

Hypoadrenalism

Other disorders of adrenal gland

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Secondary hypocortisolism

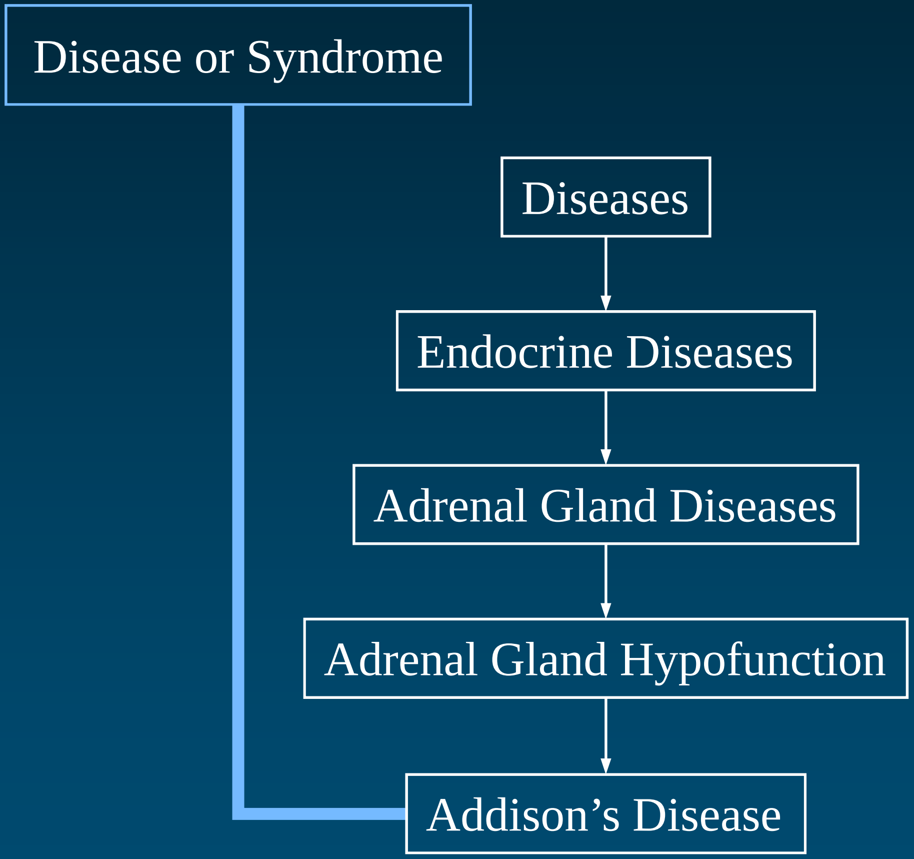
Addison's Disease

Addison's disease due to autoimmunity

relate to other concepts

Categorize concepts

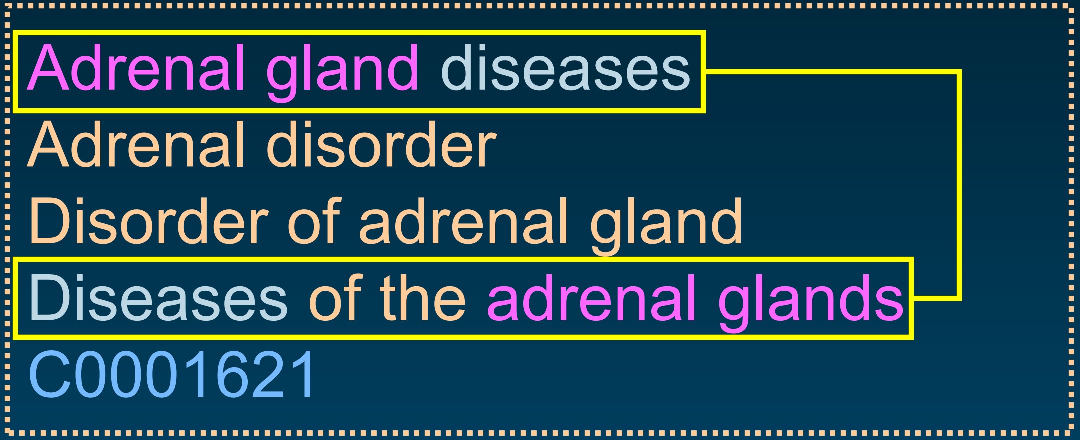
- High-level categories (semantic types)
- Assigned by the Metathesaurus editors
- Independently of the hierarchies in which these concepts are located



How do they do that?

- Lexical knowledge
- Semantic pre-processing
- UMLS editors

Lexical knowledge



Adrenal gland diseases

Adrenal disorder

Disorder of adrenal gland

Diseases of the adrenal glands

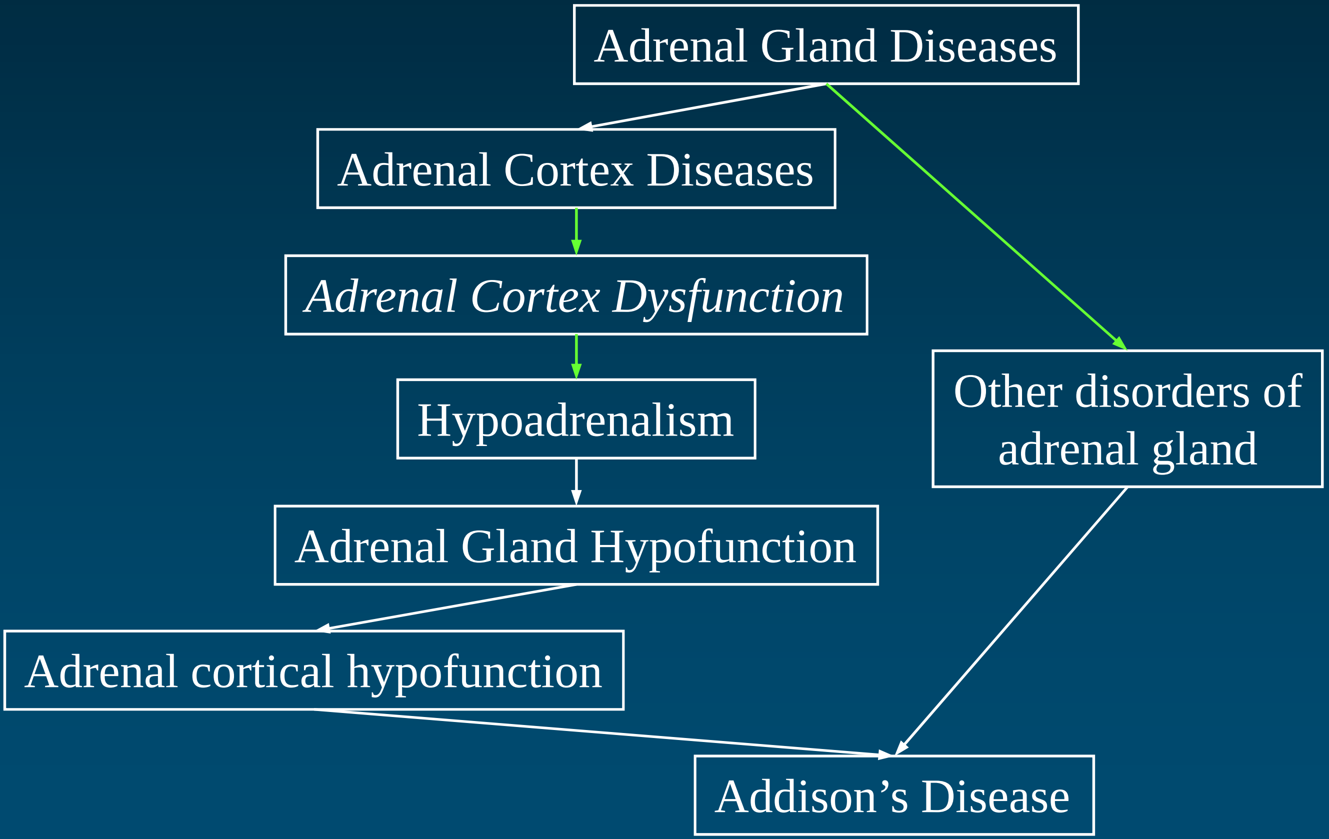
C0001621

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



Adrenal Gland Diseases

Adrenal Cortex Diseases

Adrenal Cortex Dysfunction

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Other disorders of adrenal gland

Addison’s Disease

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

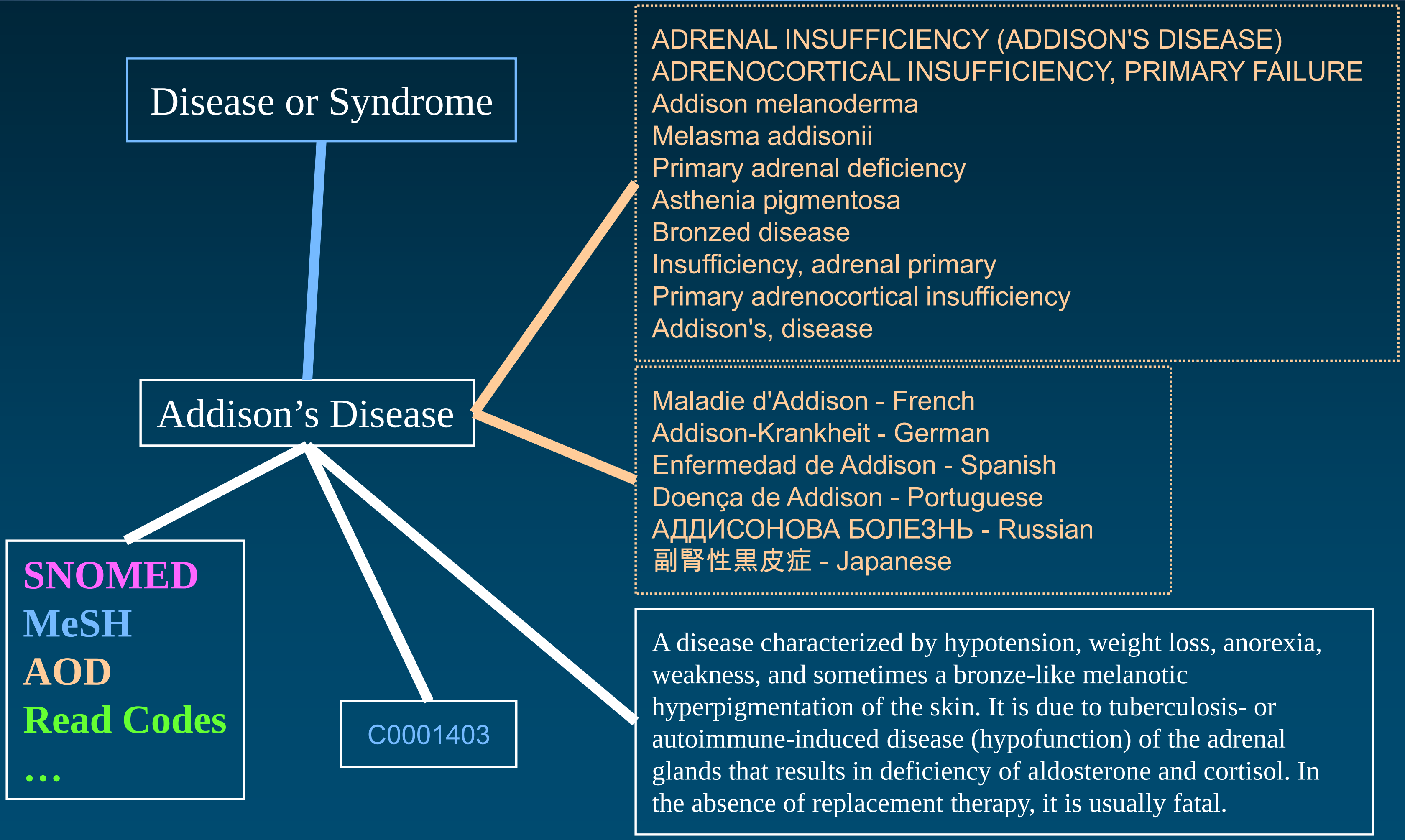
Ontological resources

UMLS Metathesaurus

Source Vocabularies (2006AC)

- 138 source vocabularies
  - 17 languages
- Broad coverage of biomedicine
  - 5.3M names
  - 1.3M concepts
  - 16M relations
- Common presentation

Addison’s Disease: Concept



Disease or Syndrome

Addison’s Disease

ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)
ADRENOCORTICAL INSUFFICIENCY, PRIMARY FAILURE
Addison melanoderma
Melasma addisonii
Primary adrenal deficiency
Asthenia pigmentosa
Bronzed disease
Insufficiency, adrenal primary
Primary adrenocortical insufficiency
Addison's, disease

Maladie d'Addison - French
Addison-Krankheit - German
Enfermedad de Addison - Spanish
Doença de Addison - Portuguese
АДДИСОНОВА БОЛЕЗНЬ - Russian
副腎性黒皮症 - Japanese

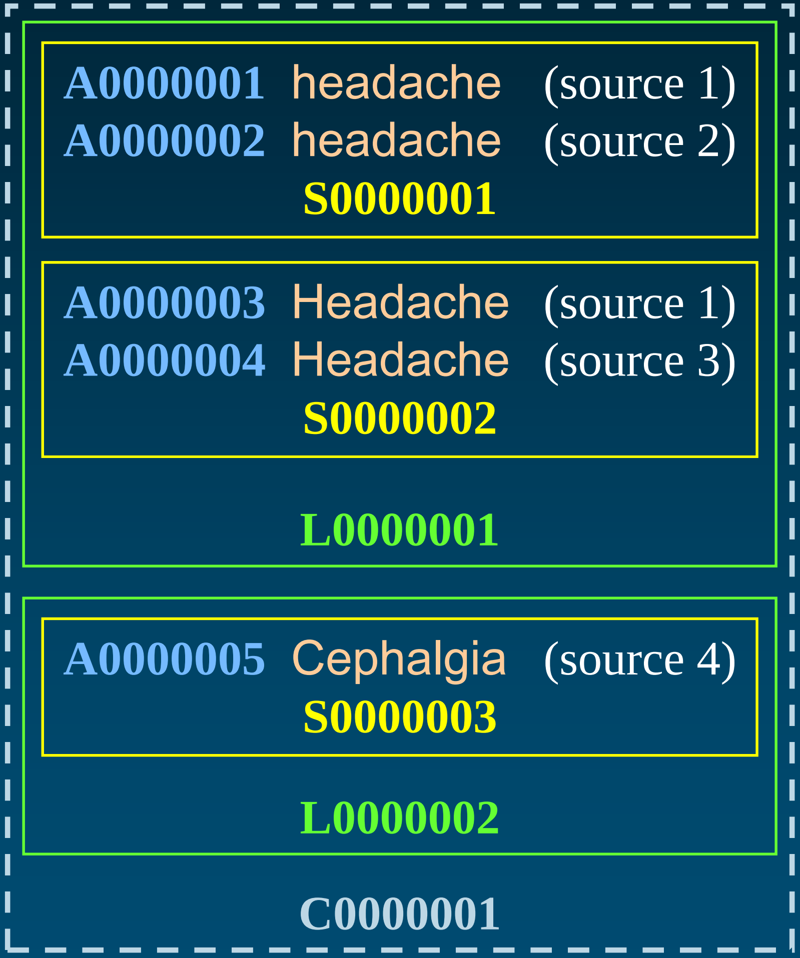
SNOMED
MeSH
AOD
Read Codes
…

C0001403

A disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin. It is due to tuberculosis- or autoimmune-induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. In the absence of replacement therapy, it is usually fatal.

Metathesaurus Concepts (2006AC)

- Concept (> 1.3M) CUI
  - Set of synonymous concept names
- Term (> 4.7M) LUI
  - Set of normalized names
- String (> 5.3M) SUI
  - Distinct concept name
- Atom (> 6.4M) AUI
  - Concept name in a given source



A0000001 headache (source 1)
A0000002 headache (source 2)
S0000001

A0000003 Headache (source 1)
A0000004 Headache (source 3)
S0000002

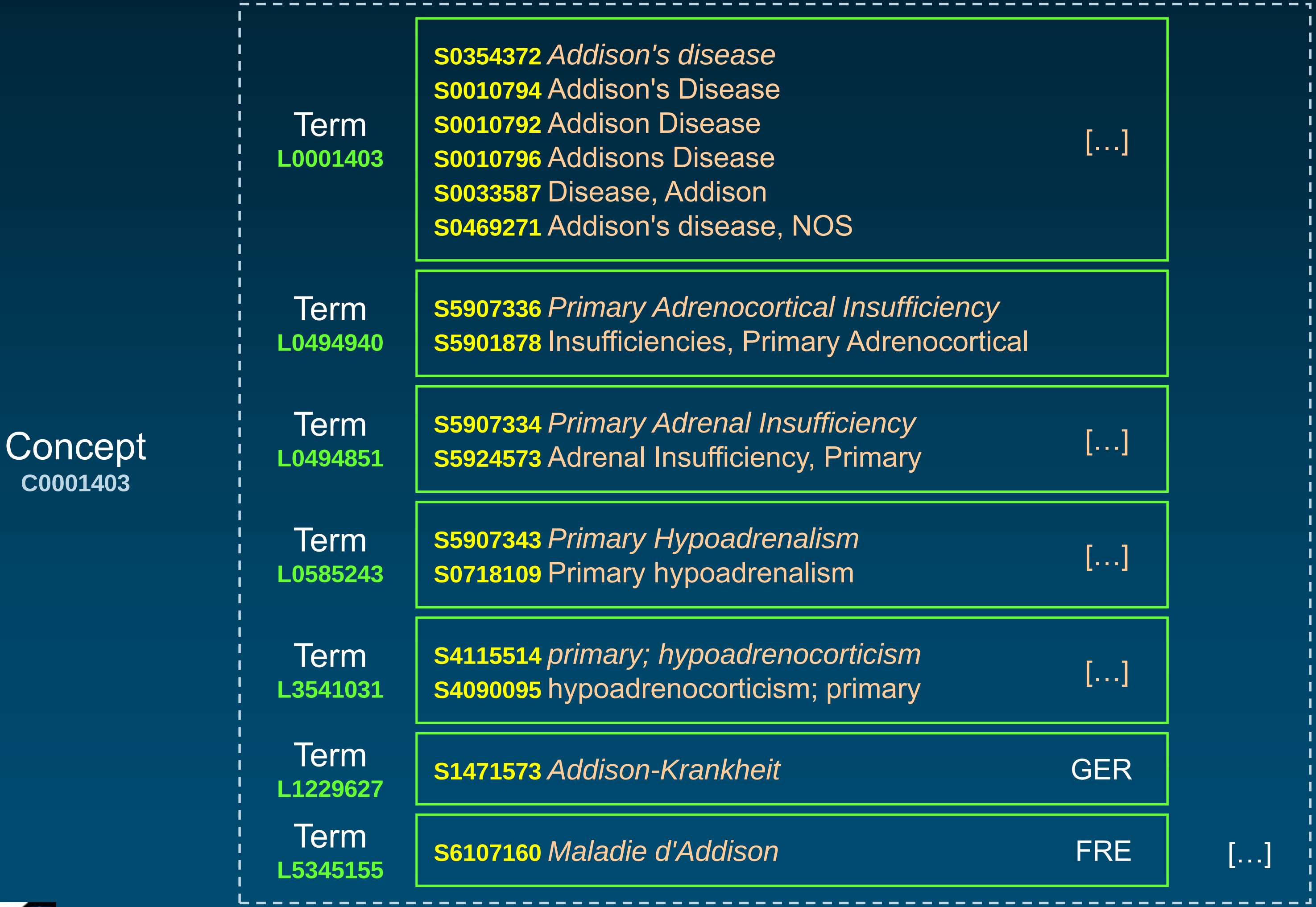
L0000001

A0000005 Cephalgia (source 4)
S0000003

L0000002

C0000001

Cluster of synonymous terms



Concept
C0001403

Term
L0001403

S0354372 *Addison's disease*
S0010794 Addison's Disease
S0010792 Addison Disease
S0010796 Addisons Disease
S0033587 Disease, Addison
S0469271 Addison's disease, NOS
[…]

Term
L0494940

S5907336 *Primary Adrenocortical Insufficiency*
S5901878 Insufficiencies, Primary Adrenocortical

Term
L0494851

S5907334 *Primary Adrenal Insufficiency*
S5924573 Adrenal Insufficiency, Primary
[…]

Term
L0585243

S5907343 *Primary Hypoadrenalism*
S0718109 Primary hypoadrenalism
[…]

Term
L3541031

S4115514 *primary; hypoadrenocorticism*
S4090095 hypoadrenocorticism; primary
[…]

Term
L1229627

S1471573 *Addison-Krankheit* GER

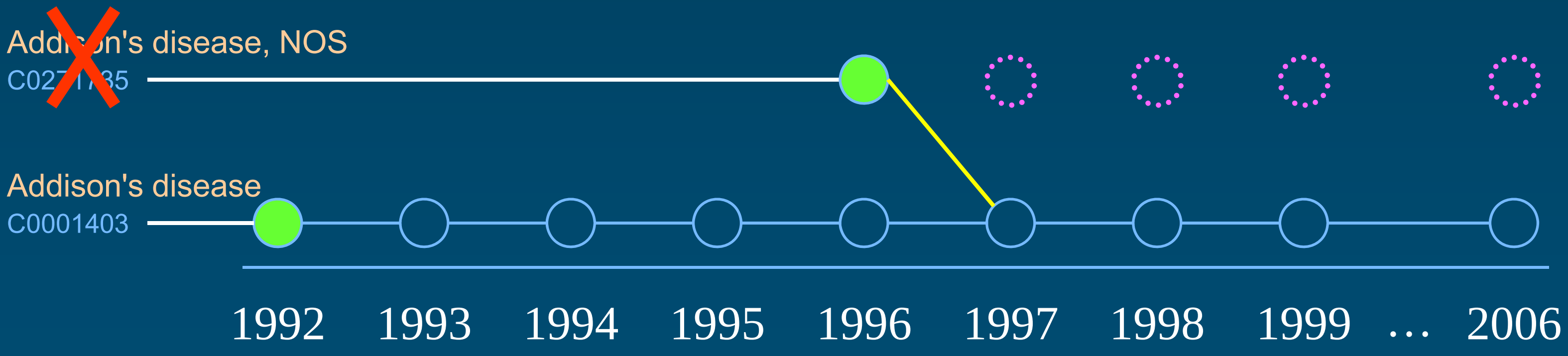
Term
L5345155

S6107160 *Maladie d'Addison* FRE

[…]

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: ~9 M pairs of concepts
- Statistical relations : ~7 M pairs of concepts (co-occurring concepts)
- Mapping relations: 100,000 pairs of concepts

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

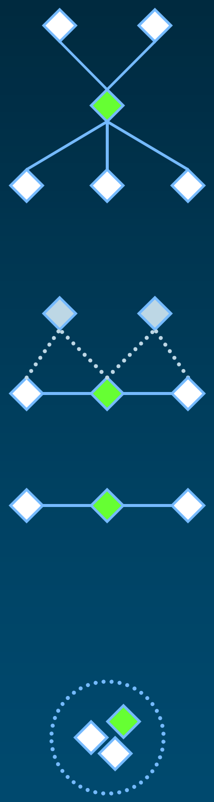
Symbolic relations

- 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

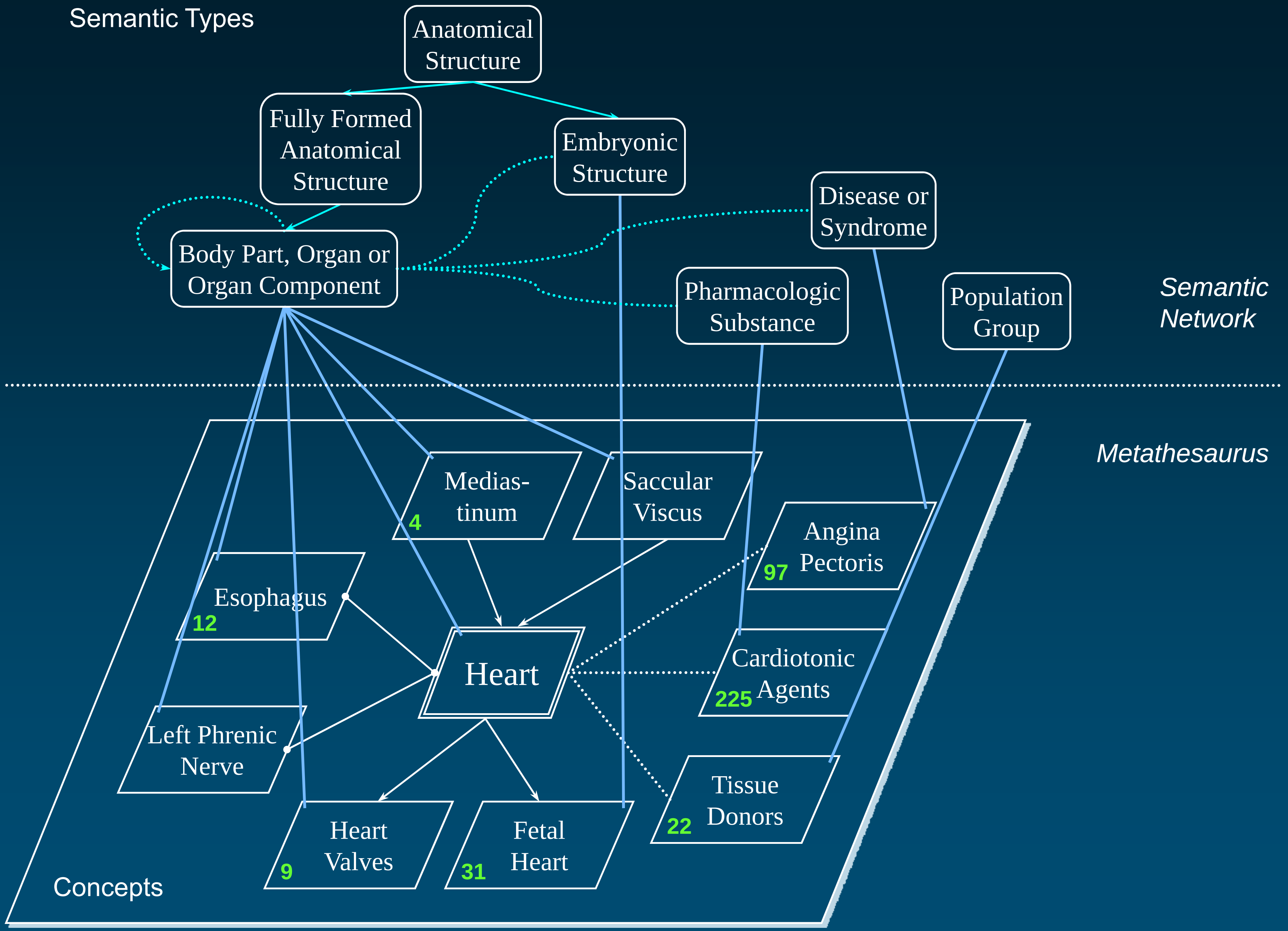
Symbolic relationships Type

◆ Hierarchical	
• Parent / Child	PAR/CHD
• Broader / Narrower than	RB/RN
◆ Derived from hierarchies	
• Siblings (children of parents)	SIB
◆ Associative	
• Other	RO
◆ Various flavors of near-synonymy	
• Similar	RL
• Source asserted synonymy	SY
• Possible synonymy	RQ



Symbolic relationships Attribute

- Hierarchical
  - isa (is-a-kind-of)
  - part-of
- Associative
  - location-of
  - caused-by
  - treats
  - …
- Cross-references (mapping)


Semantic Types
Anatomical Structure
Fully Formed Anatomical Structure
Embryonic Structure
Body Part, Organ or Organ Component
Disease or Syndrome
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

UMLS Semantic Network

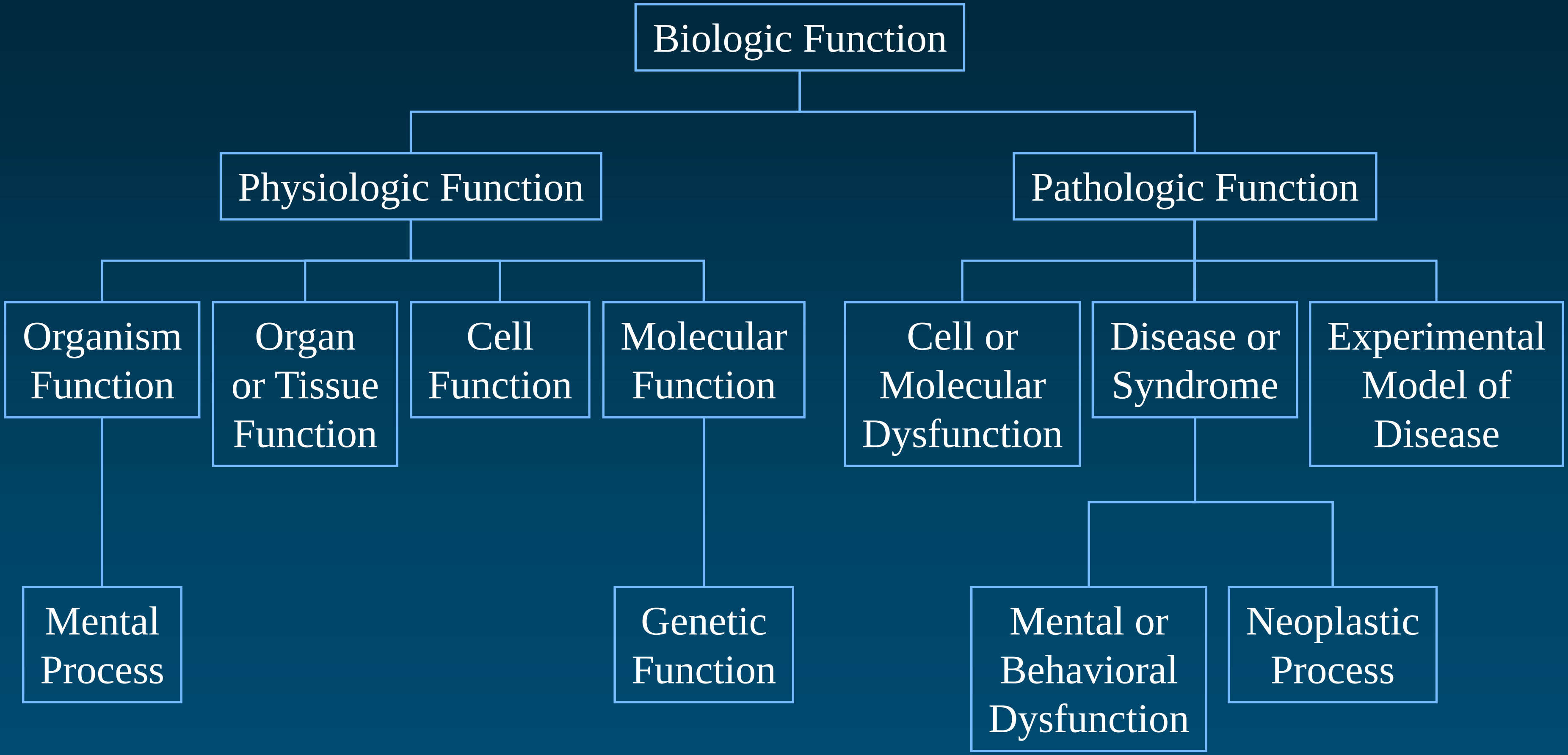
Semantic Network

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

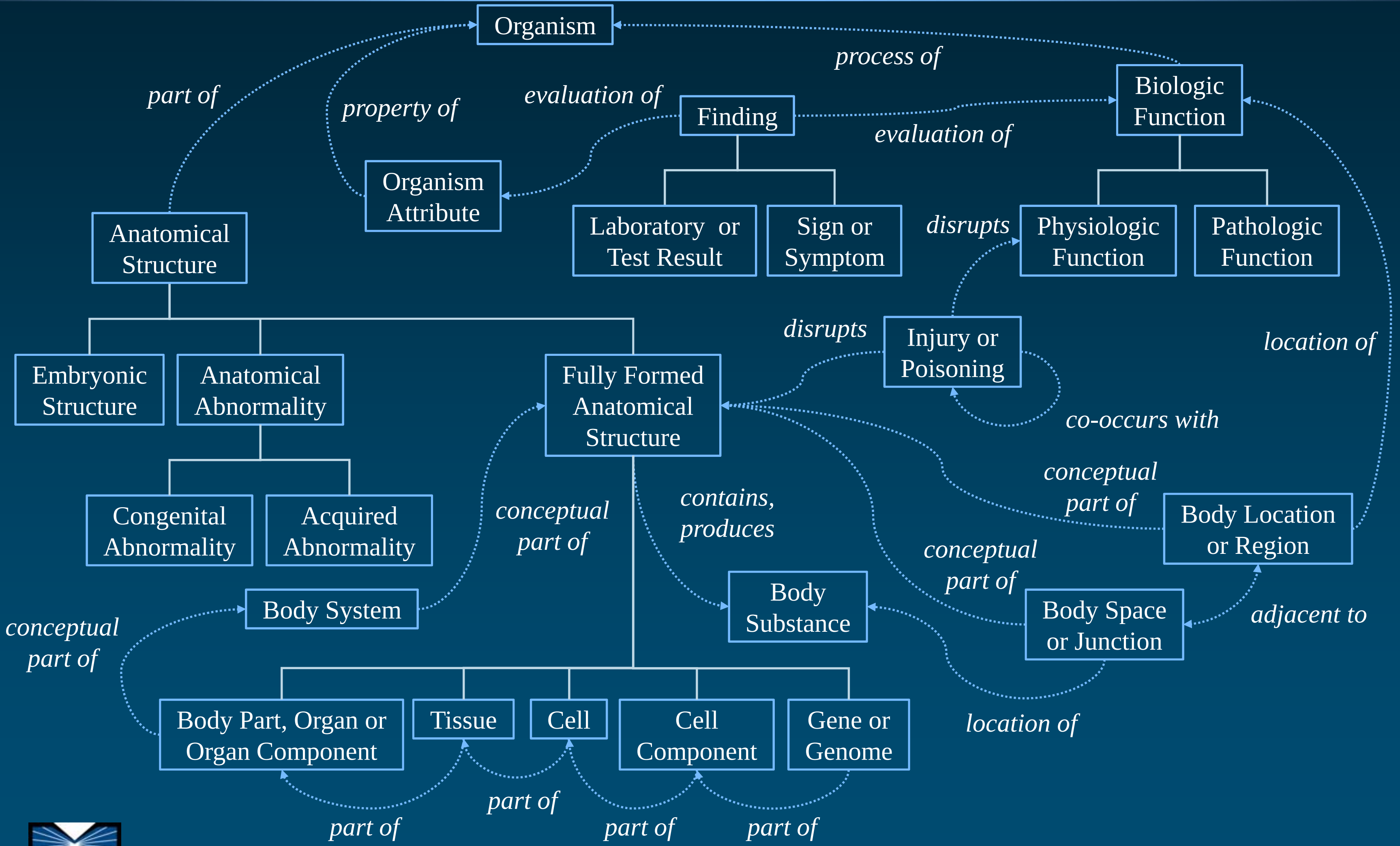
Semantic Network

- 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



Organism

part of

property of

evaluation of

process of

Finding

Biologic Function

evaluation of

Organism Attribute

Anatomical Structure

Laboratory or Test Result

Sign or Symptom

disrupts

Physiologic Function

Pathologic Function

disrupts

Injury or Poisoning

location of

Embryonic Structure

Anatomical Abnormality

Fully Formed Anatomical Structure

co-occurs with

conceptual part of

Congenital Abnormality

Acquired Abnormality

conceptual part of

contains, produces

Body Location or Region

conceptual part of

Body System

Body Substance

Body Space or Junction

adjacent to

conceptual part of

Body Part, Organ or Organ Component

Tissue

Cell

Cell Component

Gene or Genome

location of

part of

part of

part of

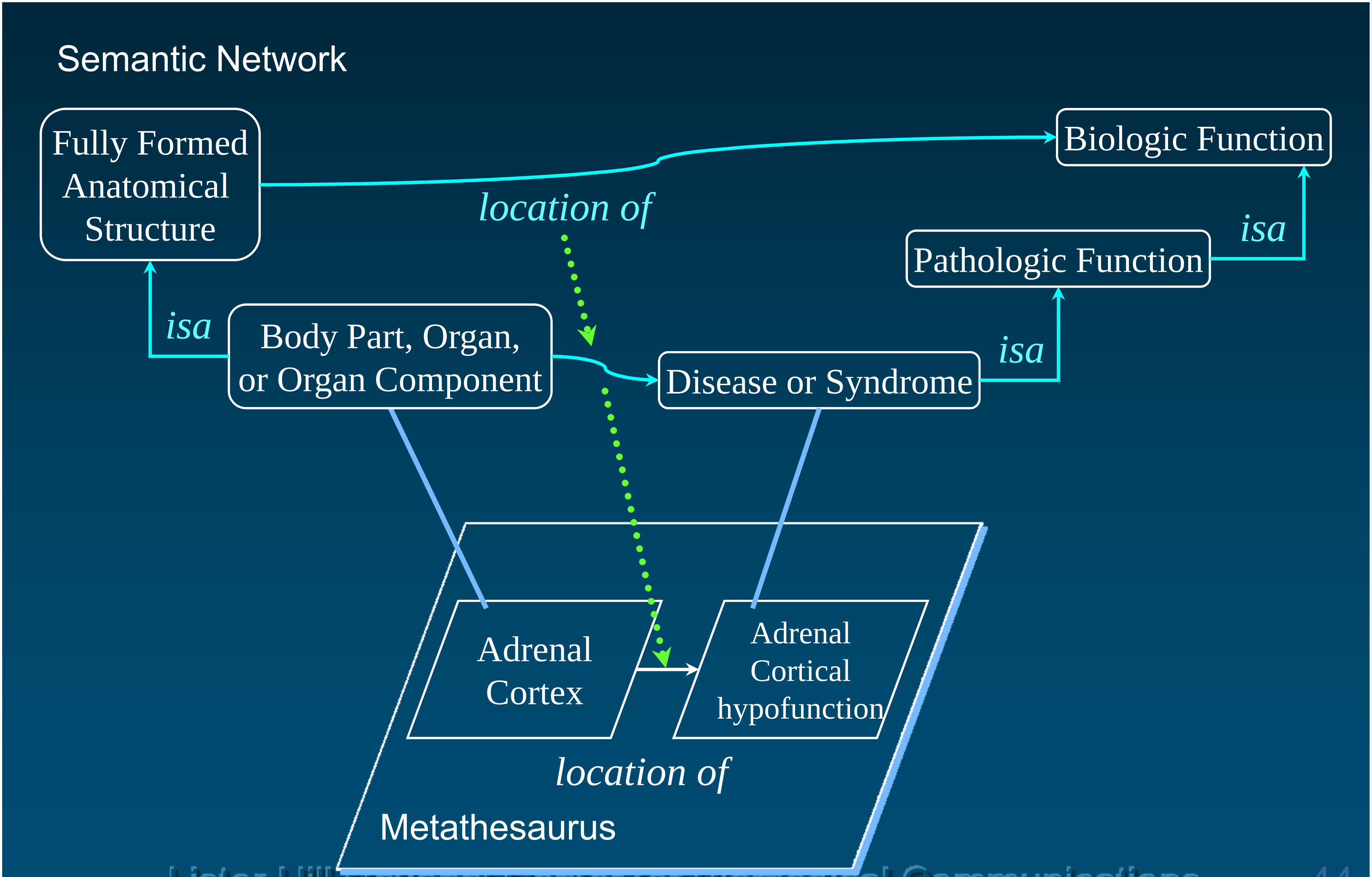
part of

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



Semantic Network

Fully Formed Anatomical Structure

location of

Biologic Function

isa

Pathologic Function

isa

Body Part, Organ, or Organ Component

Disease or Syndrome

isa

Adrenal Cortex

Adrenal Cortical hypofunction

location of

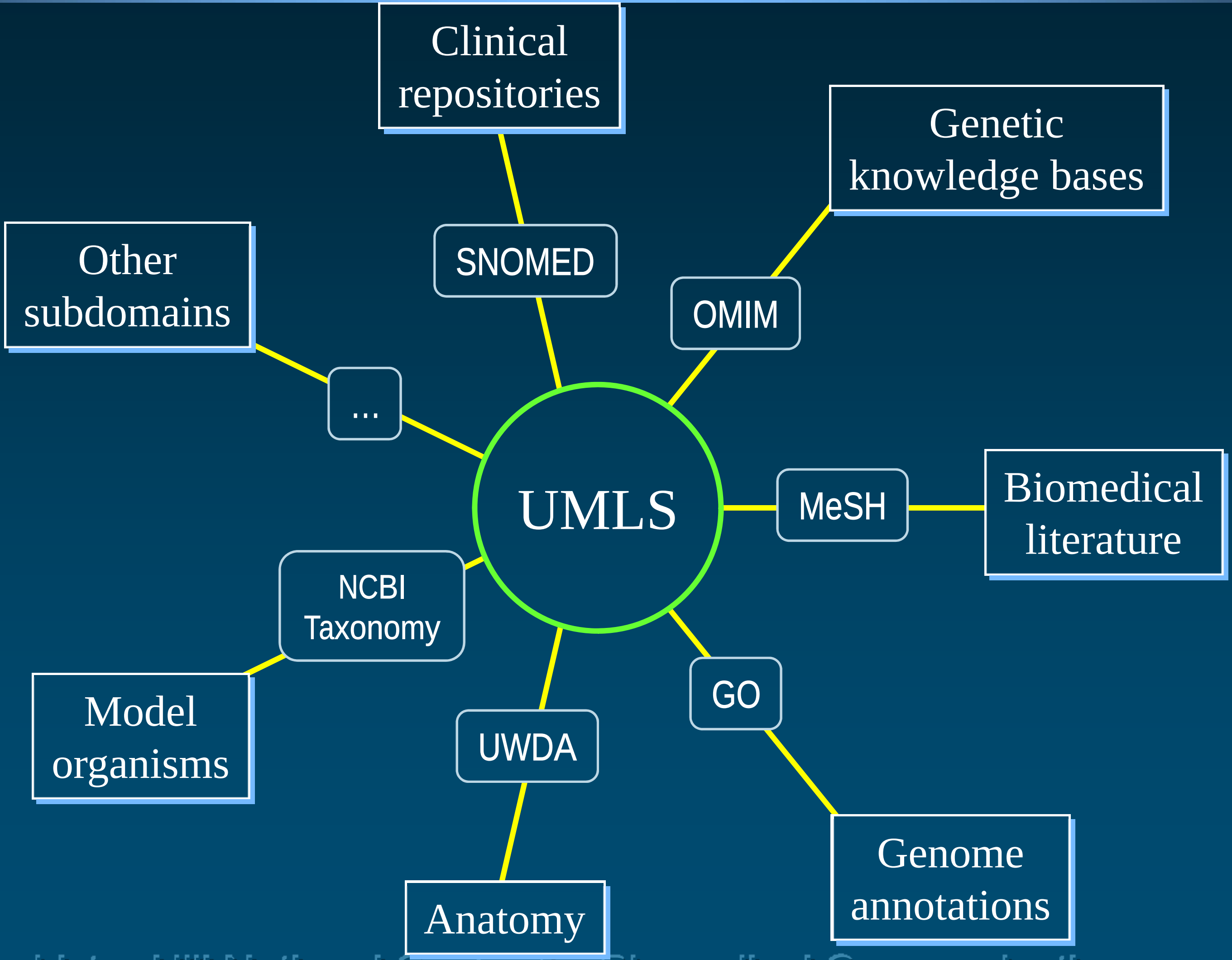
Metathesaurus

UMLS Summary

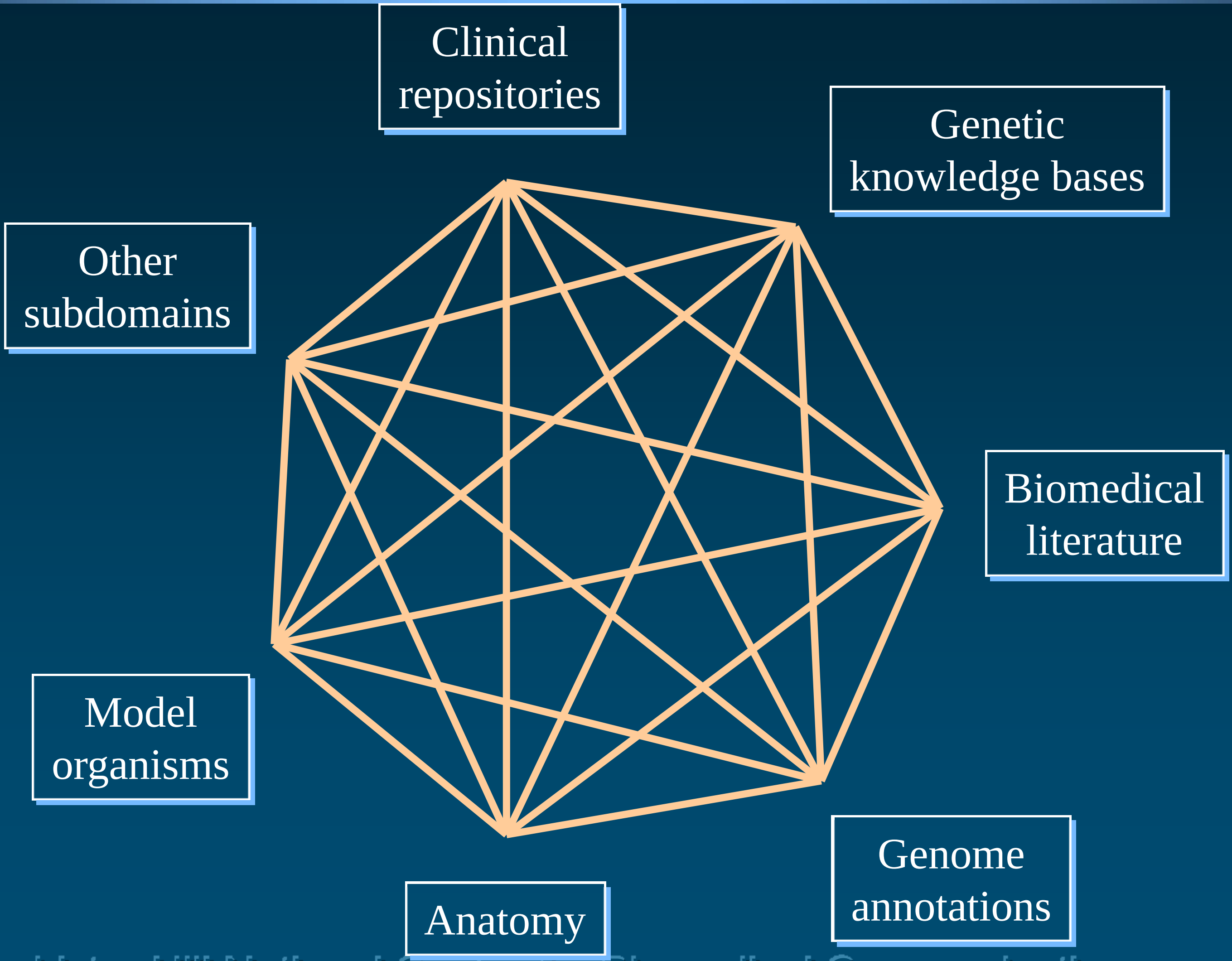
- Synonymous terms clustered into concepts
- Unique identifier

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

Integrating subdomains



Integrating subdomains



Clinical repositories

Genetic knowledge bases

Biomedical literature

Genome annotations

Anatomy

Model organisms

Other subdomains

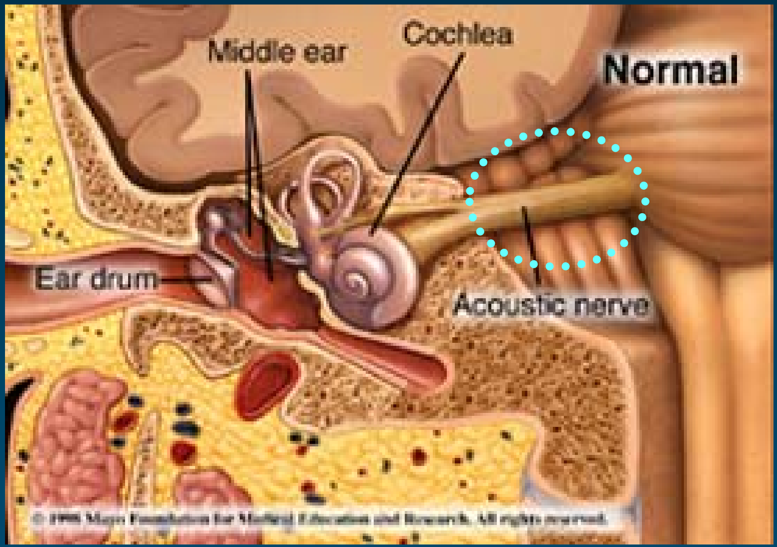
Information integration

Genomics as an example

NF2 Gene, protein, and disease

Neurofibromatosis 2 is an autosomal dominant disease characterized by tumors called schwannomas involving the acoustic nerve, as well as other features. The disorder is caused by mutations of the *NF2 gene* resulting in absence or inactivation of the protein product. The protein product of NF2 is commonly called *merlin* (but also neurofibromin 2 and schwannomin) and functions as a tumor suppressor.

Schwannoma (acoustic neuroma)


Middle ear
Cochlea
Normal
Ear drum
Acoustic nerve
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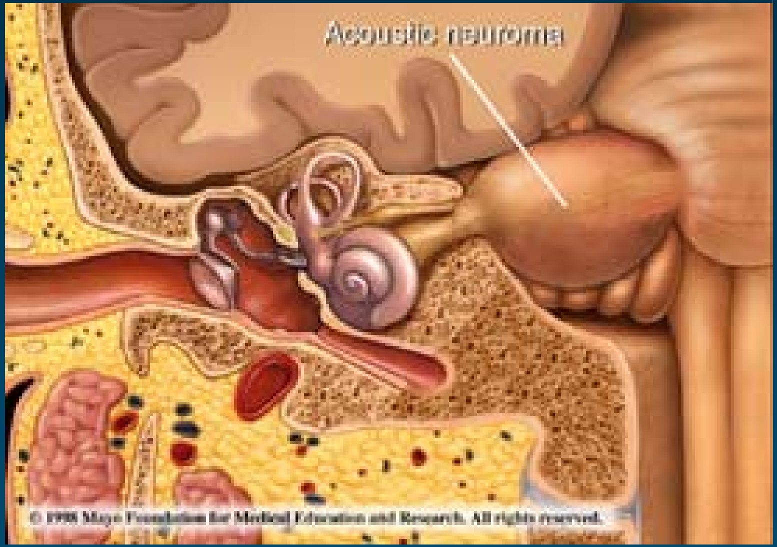
Acoustic neuroma
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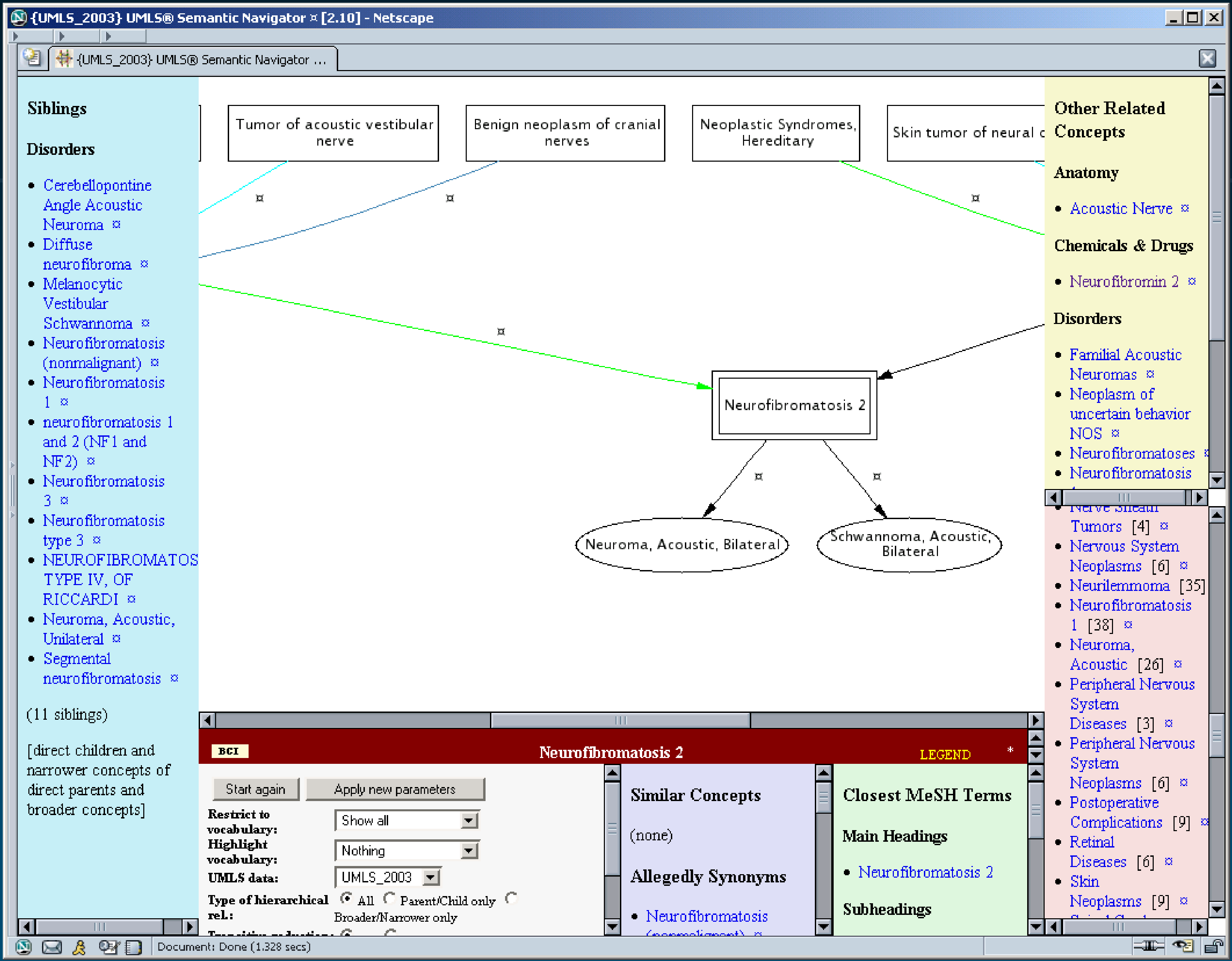
http://www.mayoclinic.com


{UMLS_2003} UMLS® Semantic Navigator ¤ [2.10] - Netscape
{UMLS_2003} UMLS® Semantic Navigator ...
Tumor of acoustic vestibular nerve
Benign neoplasm of cranial nerves
Neoplastic Syndromes, Hereditary
Skin tumor of neural c
Neurofibromatosis 2
Neuroma, Acoustic, Bilateral
Schwannoma, Acoustic, Bilateral
BCI
LEGEND
Start again
Apply new parameters
Document: Done (1.328 secs)

Siblings

Disorders

- Cerebellopontine Angle Acoustic Neuroma ¤
- Diffuse neurofibroma ¤
- Melanocytic Vestibular Schwannoma ¤
- Neurofibromatosis (nonmalignant) ¤
- Neurofibromatosis 1 ¤
- neurofibromatosis 1 and 2 (NF1 and NF2) ¤
- Neurofibromatosis 3 ¤
- Neurofibromatosis type 3 ¤
- NEUROFIBROMATOS TYPE IV, OF RICCARDI ¤
- Neuroma, Acoustic, Unilateral ¤
- Segmental neurofibromatosis ¤

(11 siblings)

[direct children and narrower concepts of direct parents and broader concepts]

Other Related Concepts

Anatomy

- Acoustic Nerve ¤

Chemicals & Drugs

- Neurofibromin 2 ¤

Disorders

- Familial Acoustic Neuromas ¤
- Neoplasm of uncertain behavior NOS ¤
- Neurofibromatoses ¤
- Neurofibromatosis

- Nerve Sheath Tumors [4] ¤
- Nervous System Neoplasms [6] ¤
- Neurilemmoma [35]
- Neurofibromatosis 1 [38] ¤
- Neuroma, Acoustic [26] ¤
- Peripheral Nervous System Diseases [3] ¤
- Peripheral Nervous System Neoplasms [6] ¤
- Postoperative Complications [9] ¤
- Retinal Diseases [6] ¤
- Skin Neoplasms [9] ¤

Neurofibromatosis 2

Restrict to vocabulary: Show all

Highlight vocabulary: Nothing

UMLS data: UMLS_2003

Type of hierarchical rel.: ● All ○ Parent/Child only ○ Broader/Narrower only

Similar Concepts

(none)

Allegedly Synonyms

- Neurofibromatosis (nonmalignant) ¤

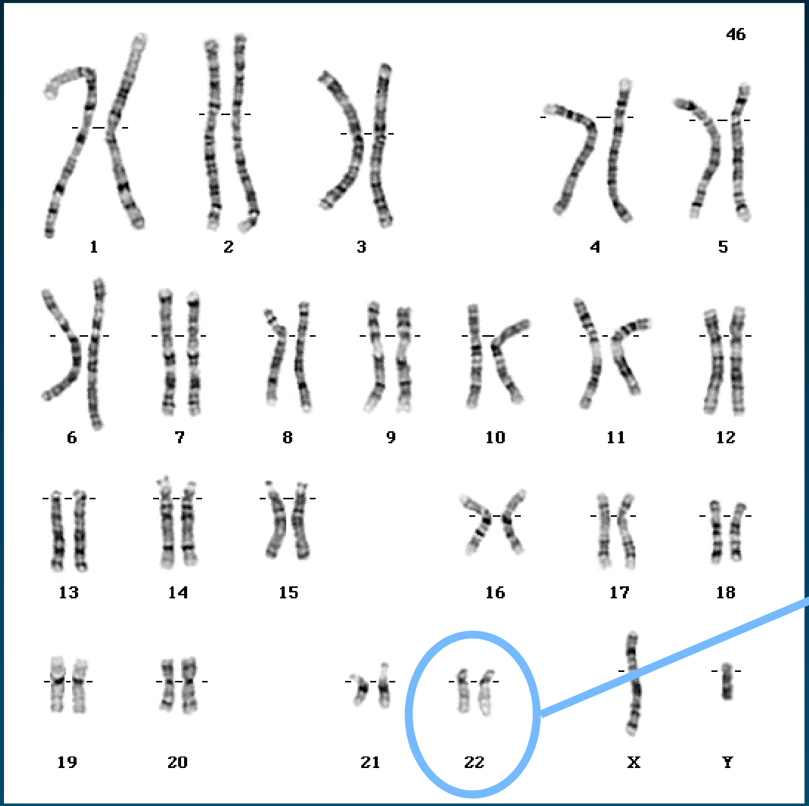
Closest MeSH Terms

Main Headings

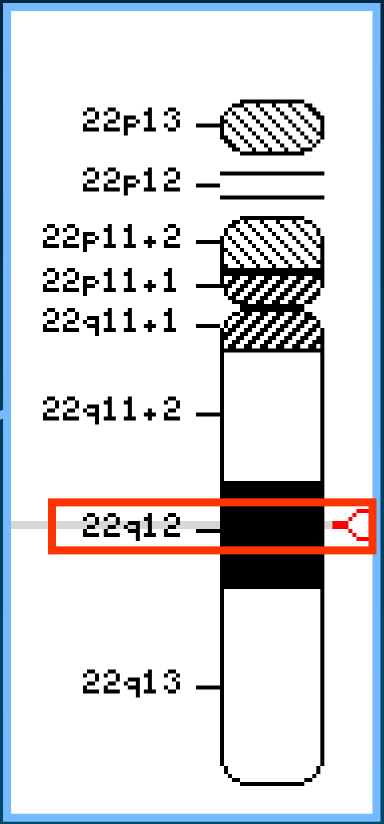
- Neurofibromatosis 2

Subheadings

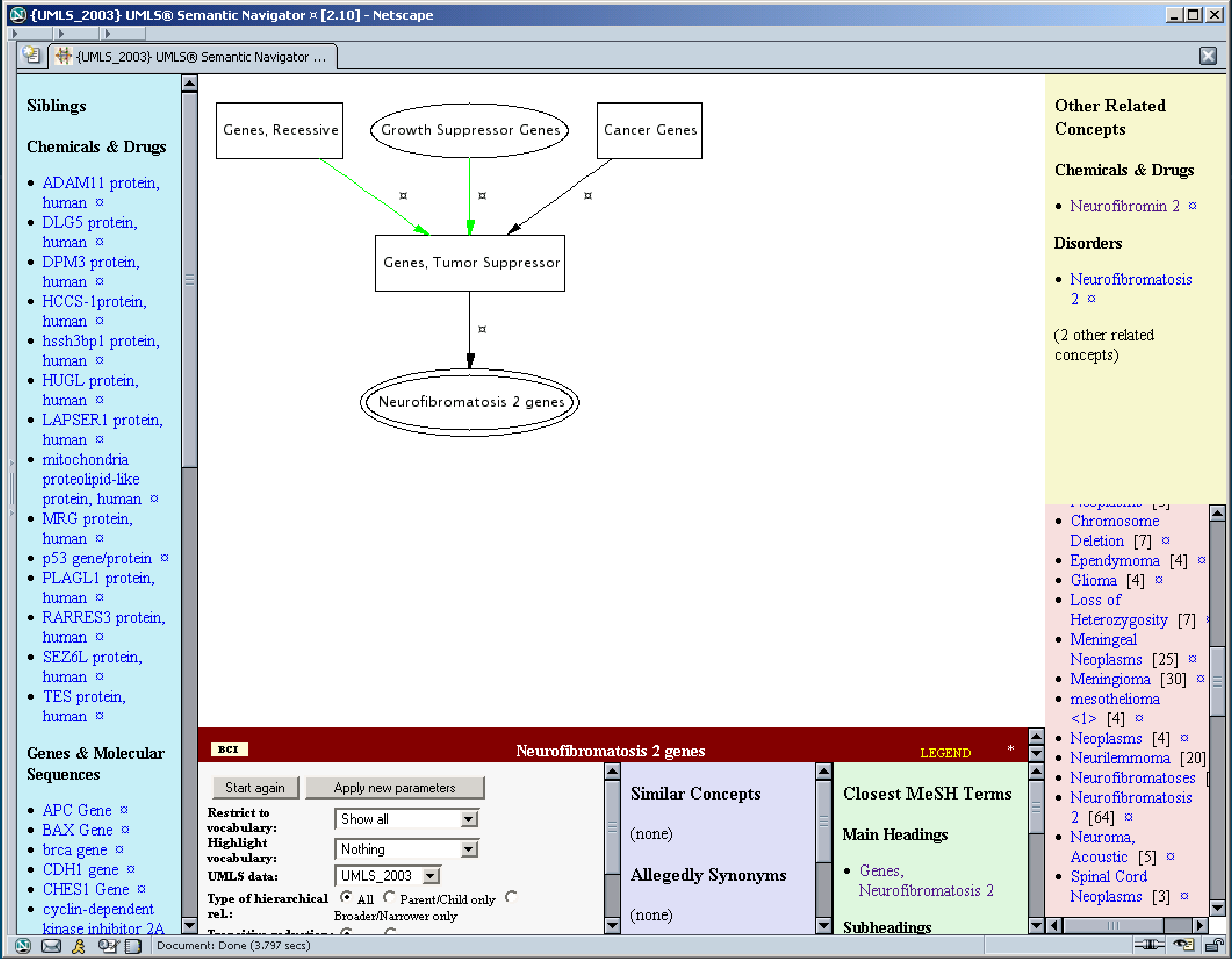
NF2 gene



http://staff.washington.edu/timk/cyto/human/



http://www.ncbi.nlm.nih.gov/mapview/



{UMLS_2003} UMLS® Semantic Navigator ¤ [2.10] - Netscape
{UMLS_2003} UMLS® Semantic Navigator ...
Genes, Recessive
Growth Suppressor Genes
Cancer Genes
¤
¤
¤
Genes, Tumor Suppressor
¤
Neurofibromatosis 2 genes
BCI
LEGEND
*
Start again
Apply new parameters

Siblings

Chemicals & Drugs

- ADAM11 protein, human ¤
- DLG5 protein, human ¤
- DPM3 protein, human ¤
- HCCS-1protein, human ¤
- hssh3bp1 protein, human ¤
- HUGL protein, human ¤
- LAPSER1 protein, human ¤
- mitochondria proteolipid-like protein, human ¤
- MRG protein, human ¤
- p53 gene/protein ¤
- PLAGL1 protein, human ¤
- RARRES3 protein, human ¤
- SEZ6L protein, human ¤
- TES protein, human ¤

Genes & Molecular Sequences

- APC Gene ¤
- BAX Gene ¤
- brca gene ¤
- CDH1 gene ¤
- CHES1 Gene ¤
- cyclin-dependent kinase inhibitor 2A

Other Related Concepts

Chemicals & Drugs

- Neurofibromin 2 ¤

Disorders

- Neurofibromatosis 2 ¤

(2 other related concepts)

- Chromosome Deletion [7] ¤
- Ependymoma [4] ¤
- Glioma [4] ¤
- Loss of Heterozygosity [7]
- Meningeal Neoplasms [25] ¤
- Meningioma [30] ¤
- mesothelioma <1> [4] ¤
- Neoplasms [4] ¤
- Neurilemmoma [20]
- Neurofibromatoses
- Neurofibromatosis 2 [64] ¤
- Neuroma, Acoustic [5] ¤
- Spinal Cord Neoplasms [3] ¤

Neurofibromatosis 2 genes

Restrict to vocabulary: Show all

Highlight vocabulary: Nothing

UMLS data: UMLS_2003

Type of hierarchical rel.: ● All ○ Parent/Child only ○ Broader/Narrower only

Similar Concepts

(none)

Allegedly Synonyms

(none)

Closest MeSH Terms

Main Headings

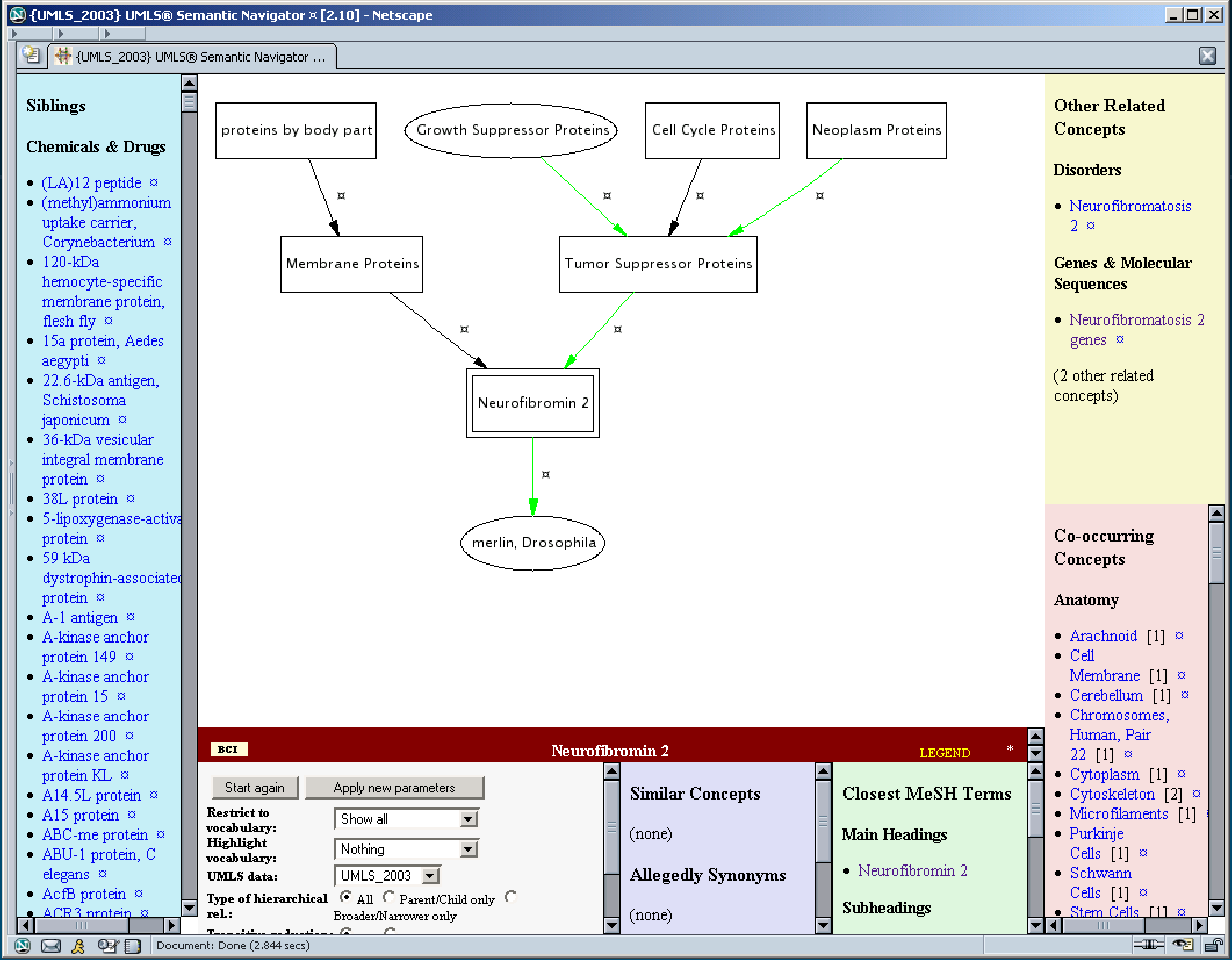
- Genes, Neurofibromatosis 2

Subheadings

Document: Done (3.797 secs)

Merlin

- Synonyms
  - Neurofibromin 2
  - Schwannomin
  - Schwannomerlin
  - Neurofibromatosis-2
- 10 isoforms
- Annotations
  - Negative regulation of cell proliferation
  - Cytoskeleton
  - Plasma membrane



{UMLS_2003} UMLS® Semantic Navigator ¤ [2.10] - Netscape
{UMLS_2003} UMLS® Semantic Navigator ...
proteins by body part
Growth Suppressor Proteins
Cell Cycle Proteins
Neoplasm Proteins
Membrane Proteins
Tumor Suppressor Proteins
Neurofibromin 2
merlin, Drosophila
BCI
LEGEND
Start again
Apply new parameters
Document: Done (2.844 secs)

Siblings

Chemicals & Drugs

- (LA)12 peptide ¤
- (methyl)ammonium uptake carrier, Corynebacterium ¤
- 120-kDa hemocyte-specific membrane protein, flesh fly ¤
- 15a protein, Aedes aegypti ¤
- 22.6-kDa antigen, Schistosoma japonicum ¤
- 36-kDa vesicular integral membrane protein ¤
- 38L protein ¤
- 5-lipoxygenase-activa protein ¤
- 59 kDa dystrophin-associated protein ¤
- A-1 antigen ¤
- A-kinase anchor protein 149 ¤
- A-kinase anchor protein 15 ¤
- A-kinase anchor protein 200 ¤
- A-kinase anchor protein KL ¤
- A14.5L protein ¤
- A15 protein ¤
- ABC-me protein ¤
- ABU-1 protein, C elegans ¤
- AcfB protein ¤
- ACR3 protein ¤

Neurofibromin 2

Restrict to vocabulary: Show all

Highlight vocabulary: Nothing

UMLS data: UMLS_2003

Type of hierarchical rel.: ● All ○ Parent/Child only ○ Broader/Narrower only

Similar Concepts

(none)

Allegedly Synonyms

(none)

Closest MeSH Terms

Main Headings

- Neurofibromin 2

Subheadings

Other Related Concepts

Disorders

- Neurofibromatosis 2 ¤

Genes & Molecular Sequences

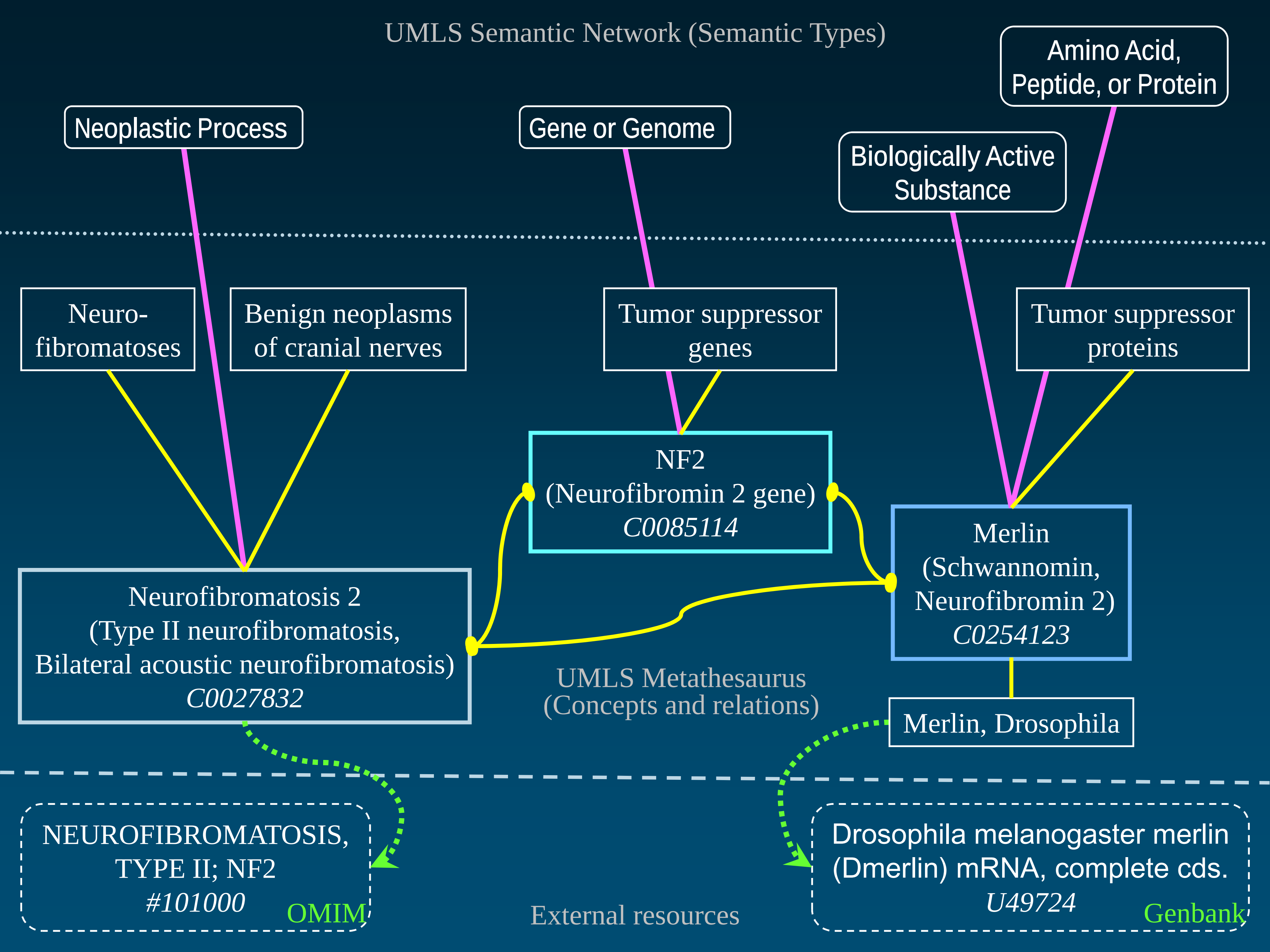
- Neurofibromatosis 2 genes ¤

(2 other related concepts)

Co-occurring Concepts

Anatomy

- Arachnoid [1] ¤
- Cell Membrane [1] ¤
- Cerebellum [1] ¤
- Chromosomes, Human, Pair 22 [1] ¤
- Cytoplasm [1] ¤
- Cytoskeleton [2] ¤
- Microfilaments [1]
- Purkinje Cells [1] ¤
- Schwann Cells [1] ¤
- Stem Cells [1] ¤



UMLS Semantic Network (Semantic Types)

Neoplastic Process

Gene or Genome

Biologically Active Substance

Amino Acid, Peptide, or Protein

Neuro-fibromatoses

Benign neoplasms of cranial nerves

Tumor suppressor genes

Tumor suppressor proteins

NF2
(Neurofibromin 2 gene)
C0085114

Merlin
(Schwannomin, Neurofibromin 2)
C0254123

Neurofibromatosis 2
(Type II neurofibromatosis, Bilateral acoustic neurofibromatosis)
C0027832

UMLS Metathesaurus
(Concepts and relations)

Merlin, Drosophila

NEUROFIBROMATOSIS, TYPE II; NF2
#101000
OMIM

External resources

Drosophila melanogaster merlin (Dmerlin) mRNA, complete cds.
U49724
Genbank

Limitations

- Genes not systematically represented
  - Most gene products and diseases are
- Gene/Gene product-Disease relations
  - Not systematically represented
  - Not explicitly represented (e.g., co-occurrence)
- Cross-references not systematically represented
- Naming conventions (genes)

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.
  - Nelson, S. J., Powell, T. & Humphreys, B. L. (2002). The Unified Medical Language System (UMLS) Project. In: Kent, Allen; Hall, Carolyn M., editors. *Encyclopedia of Library and Information Science*. New York: Marcel Dekker. p.369-378.

References

- UMLS as a research project
  - Lindberg, D. A., Humphreys, B. L., & McCray, A. T. (1993). The Unified Medical Language System. *Methods Inf Med*, *32*(4), 281-91.
  - 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.

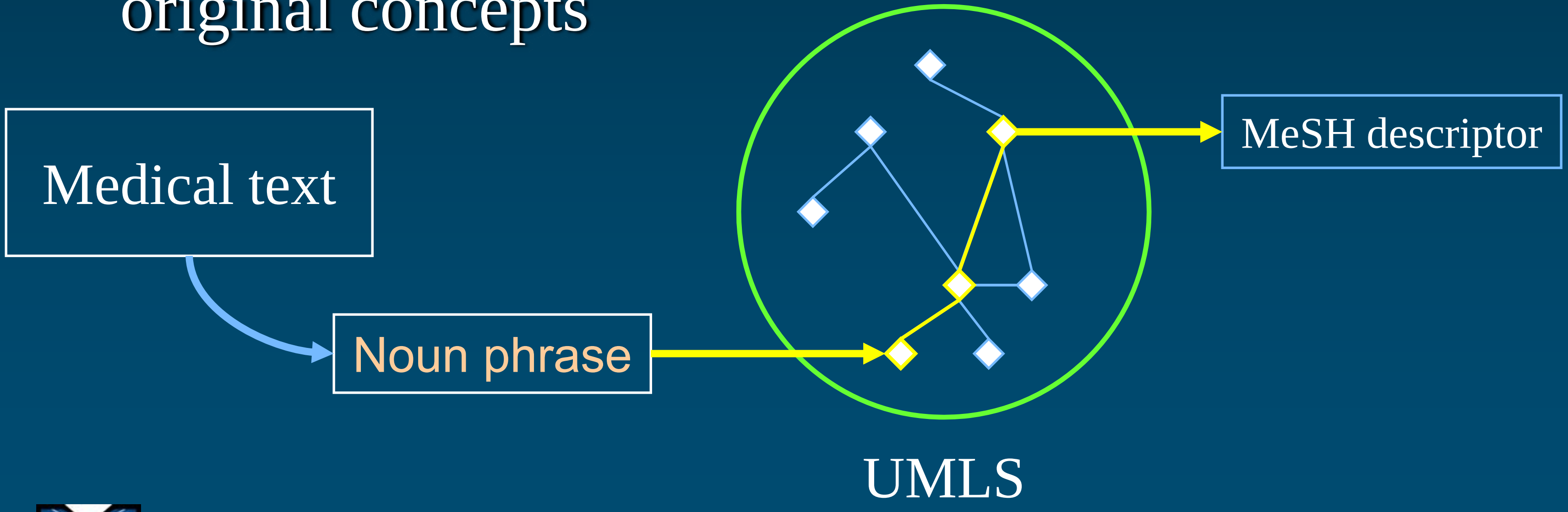
References

- Technical papers
  - McCray, A. T., & Nelson, S. J. (1995). The representation of meaning in the UMLS. *Methods Inf Med*, *34*(1-2), 193-201.
  - Bodenreider O. & McCray A. T. (2003). Exploring semantic groups through visual approaches. *Journal of Biomedical Informatics*, 36(6), 414-432.

UMLS in Use
Mapping across Vocabularies

The problem

- For noun phrases extracted from medical texts, map to UMLS concepts
- Then, select from the MeSH vocabulary the concepts that are the most closely related to the original concepts



Map noun phrases to UMLS

- Normalization
  - normalize noun phrases
  - use the normalized string index
- MetaMap
  - approximate matching
  - more aggressive approach
    - use derivational variants
    - allow partial matches

Restrict to MeSH

- Based on the principle of semantic locality
- Use different components of the UMLS
- 4 techniques of increasing aggressiveness

Use Synonymy	MRCON + MRSO
Use Associated expressions (ATXs)	MRATX
Explore the Ancestors	MRREL + SN
Explore the Other related concepts	MRREL + SN

Restrict to MeSH: Synonymy

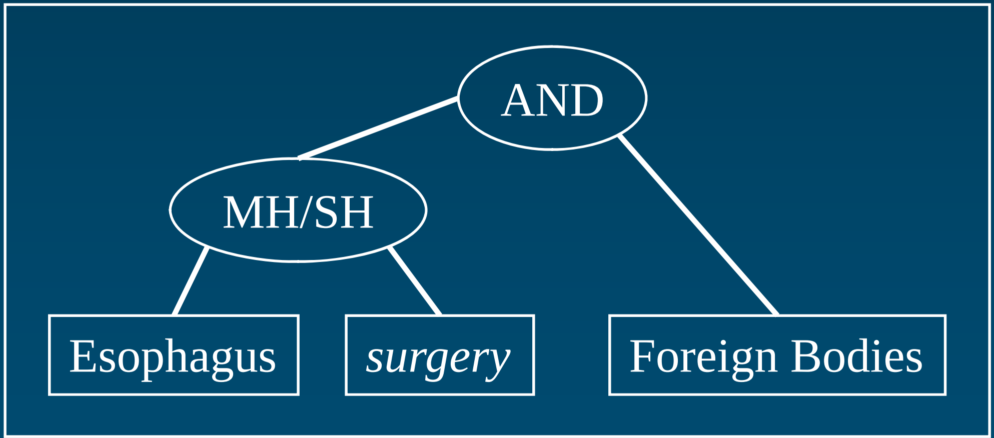
- Term mapped to Source concept
- For this concept, is there a synonym term that comes from MeSH? (MRSO)

Restrict to MeSH: Assoc. expressions

- If not,
- Is there an associated expression (ATX) that describes this concept using a combination of MeSH descriptors? (MRATX)

Endoscopic removal of intraluminal foreign body from oesophagus without incision

⇔



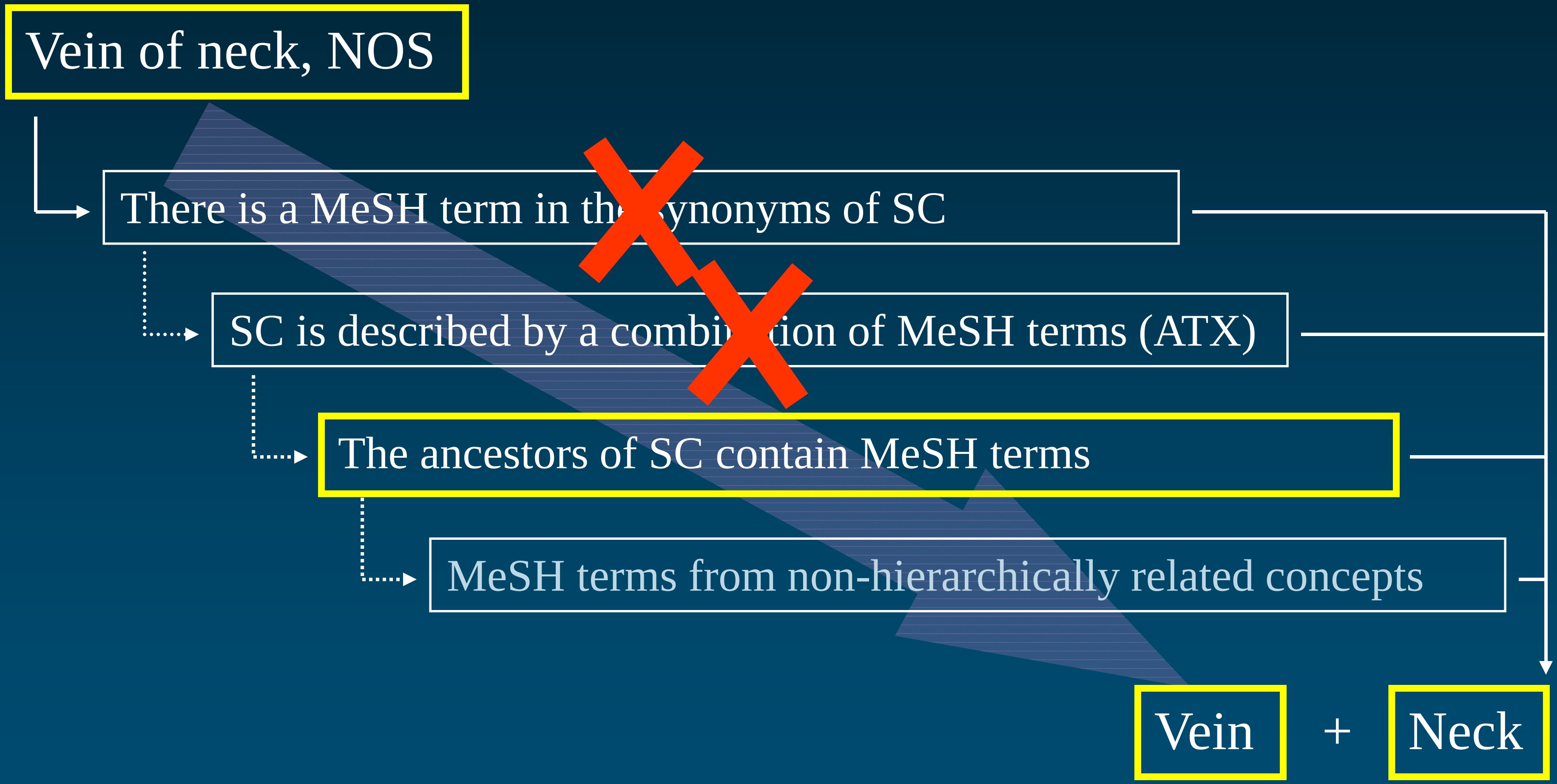
Restrict to MeSH: Ancestors

- If not, let us build the graph of the ancestors of this concept
  - using parents and broader concepts (MRREL)
  - all the way to the top
  - excluding ancestors whose semantic types are not compatible with those of the source concept (MRSTY)
- From the graph, select the concepts that come from MeSH (MRCONSO)
- Remove those that are ancestors of another concept coming from MeSH

Restrict to MeSH: Other related concepts

- If not, explore the other related concepts (MRREL) whose semantic types are compatible with those of the source concept (MRSTY)
- From those, select the concepts that come from MeSH (MRCONSO)

Restrict to MeSH: Example



Vein of neck, NOS

There is a MeSH term in the synonyms of SC

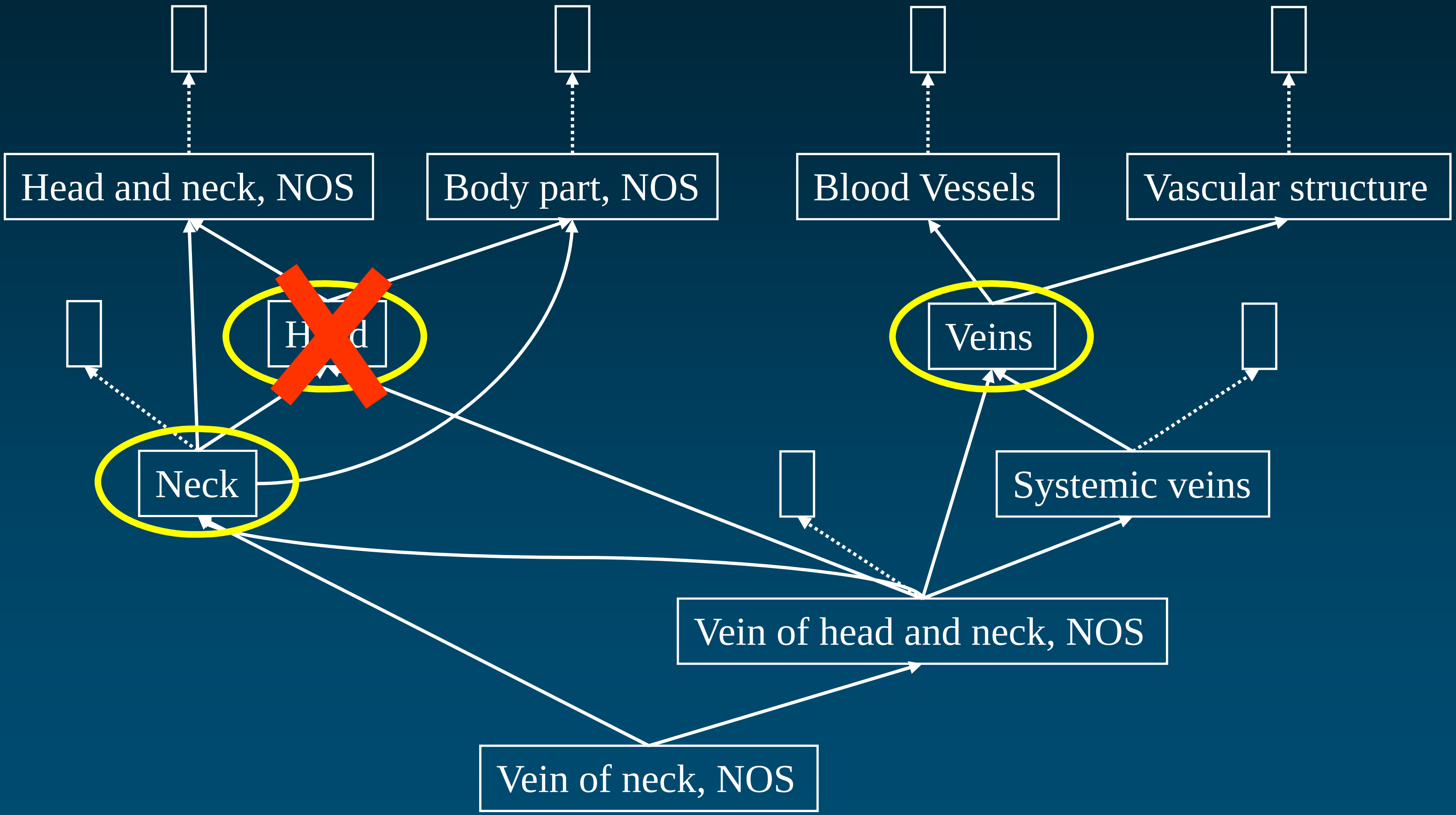
SC is described by a combination of MeSH terms (ATX)

The ancestors of SC contain MeSH terms

MeSH terms from non-hierarchically related concepts

Vein + Neck

Restrict to MeSH: Example



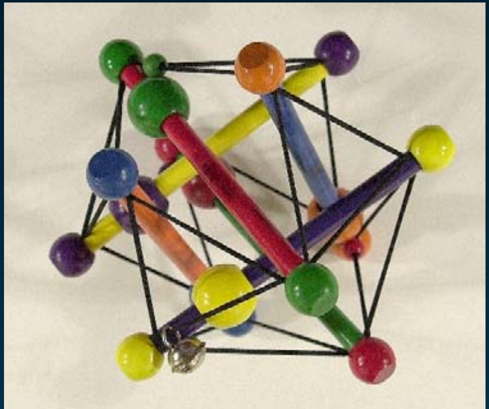
Overall results

◆ Synonymy:	24%
◆ Built-in mapping:	1%
◆ Ancestors	
• From concept:	49%
• From children:	2%
• From siblings:	1%
◆ Other:	11%
◆ No mapping	12%

References

- Bodenreider O, Nelson SJ, Hole WT, Chang HF. *Beyond synonymy: exploiting the UMLS semantics in mapping vocabularies*. Proceedings of AMIA Annual Symposium 1998:815-9. http://mor.nlm.nih.gov/pubs/pdf/1998-amia-ob.pdf

- Fung KW, Bodenreider O. *Utilizing the UMLS for semantic mapping between terminologies*. Proceedings of AMIA Annual Symposium 2005:266-270. http://mor.nlm.nih.gov/pubs/pdf/2005-amia-kwf.pdf



Medical Ontology Research

Contact: olivier@nlm.nih.gov
Web: mor.nlm.nih.gov

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Olivier Bodenreider

Lister Hill National Center
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