



Johns Hopkins University
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Introduction to the Unified Medical Language System



Olivier Bodenreider

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

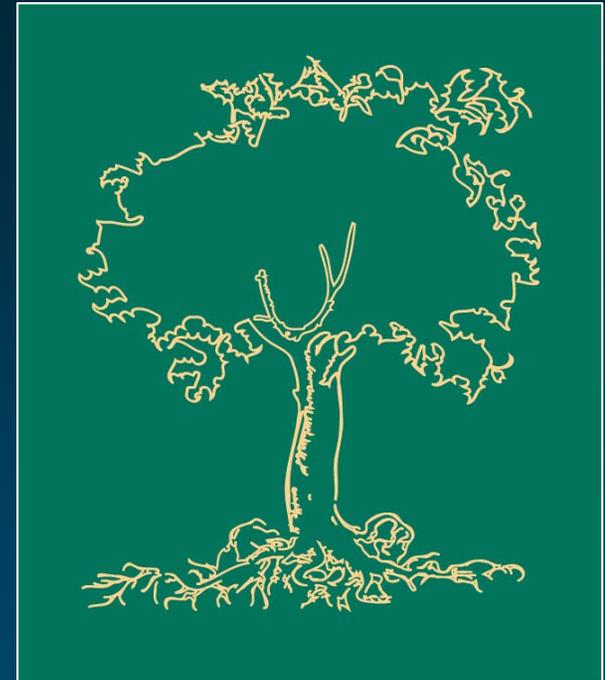
Outline

- ◆ Introduction
- ◆ Overview through an example
- ◆ The three UMLS Knowledge Sources
 - UMLS Metathesaurus
 - UMLS Semantic Network
 - SPECIALIST Lexicon and lexical tools
- ◆ UMLS in action: *MetaMap*

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”

«[...] the UMLS project is an effort to overcome two significant barriers to effective retrieval of machine-readable information.

- The first is **the variety of ways the same concepts are expressed** in different machine-readable sources and by different people.
- The second is the **distribution** of useful information among many disparate databases and systems.»

The UMLS in practice

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

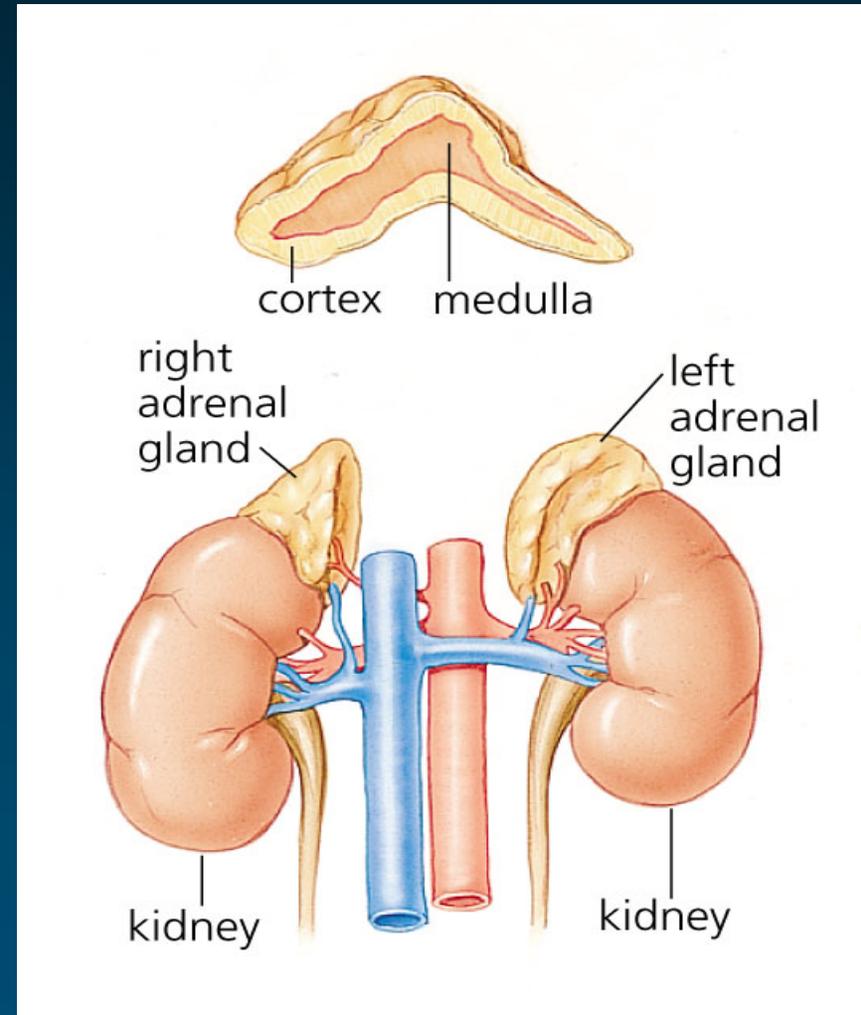


The UMLS is *not* an end-user application

Overview through an example

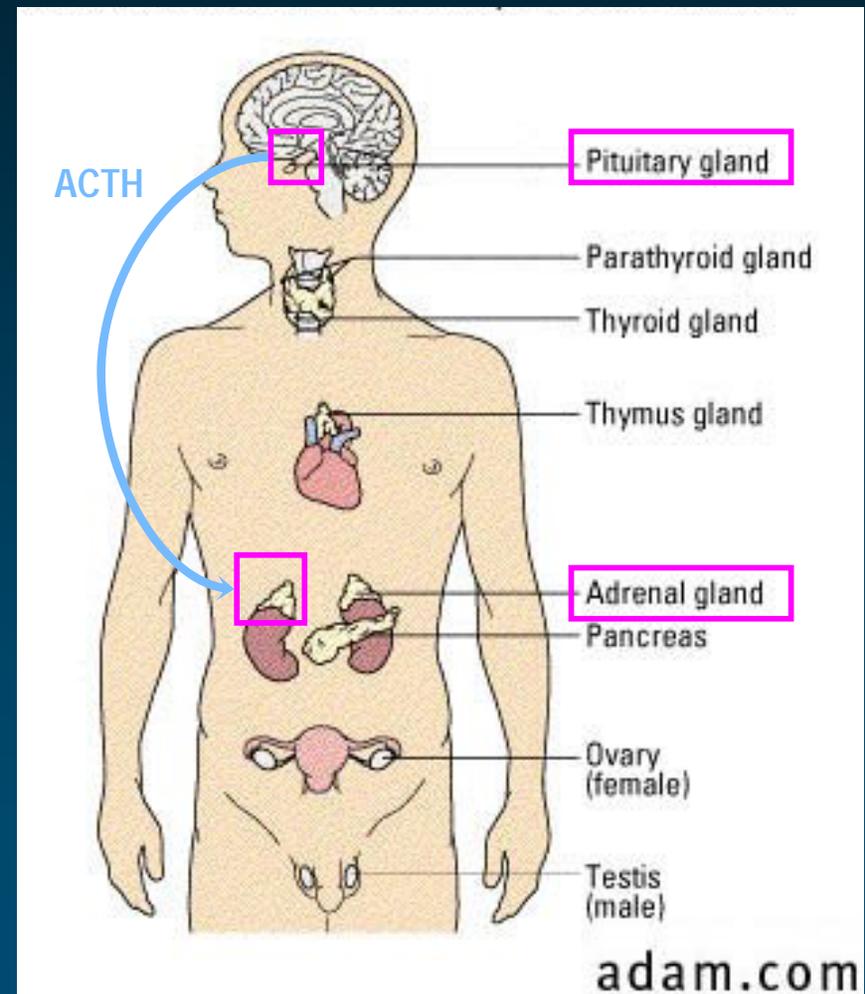
Addison's disease

- ◆ 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



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
 - Bronzed disease
 - Addison melanoderma
 - Asthenia pigmentosa
 - Primary adrenal deficiency
 - Primary adrenal insufficiency
 - Primary adrenocortical insufficiency
 - Chronic adrenocortical insufficiency
- } eponym
- } symptoms
- } clinical variants

◆ Contexts: different hierarchies



Organize terms

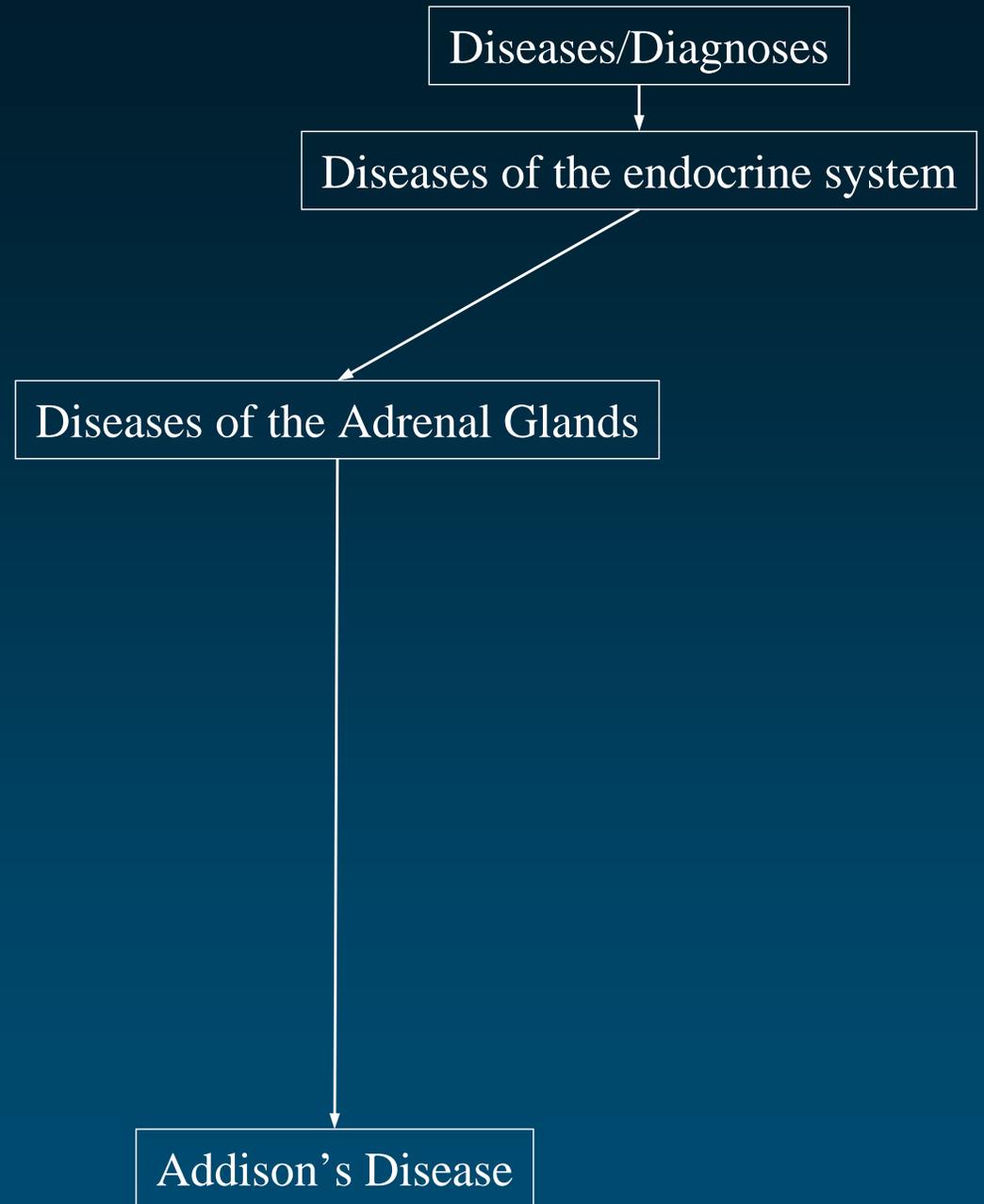
- ◆ Synonymous terms clustered into a concept
- ◆ Preferred term
- ◆ Unique identifier (CUI)

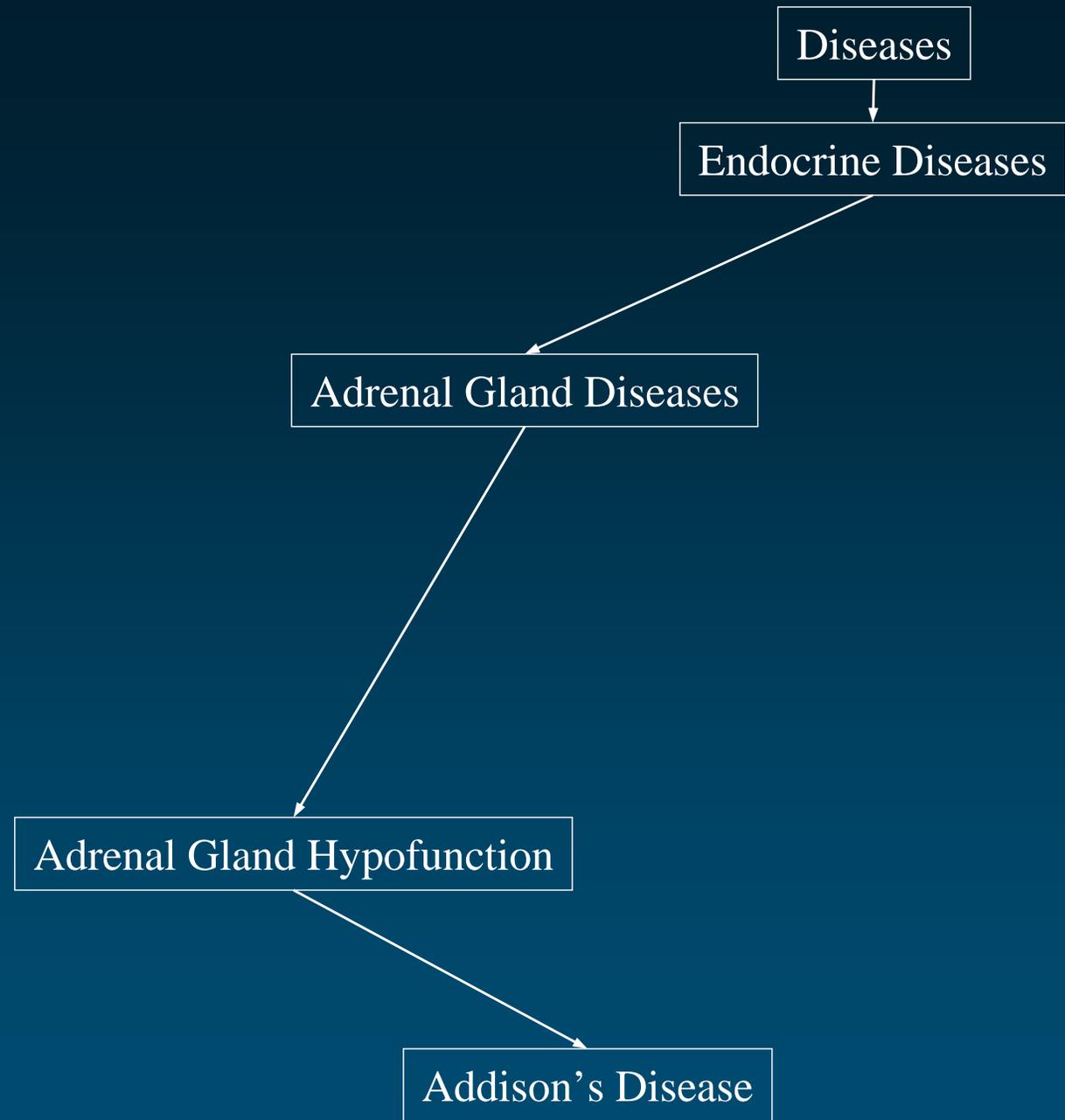
Addison Disease	MeSH	D000224
Primary hypoadrenalism	MedDRA	10036696
Primary adrenocortical insufficiency	ICD-10	E27.1
Addison's disease (disorder)	SNOMED CT	363732003

C0001403

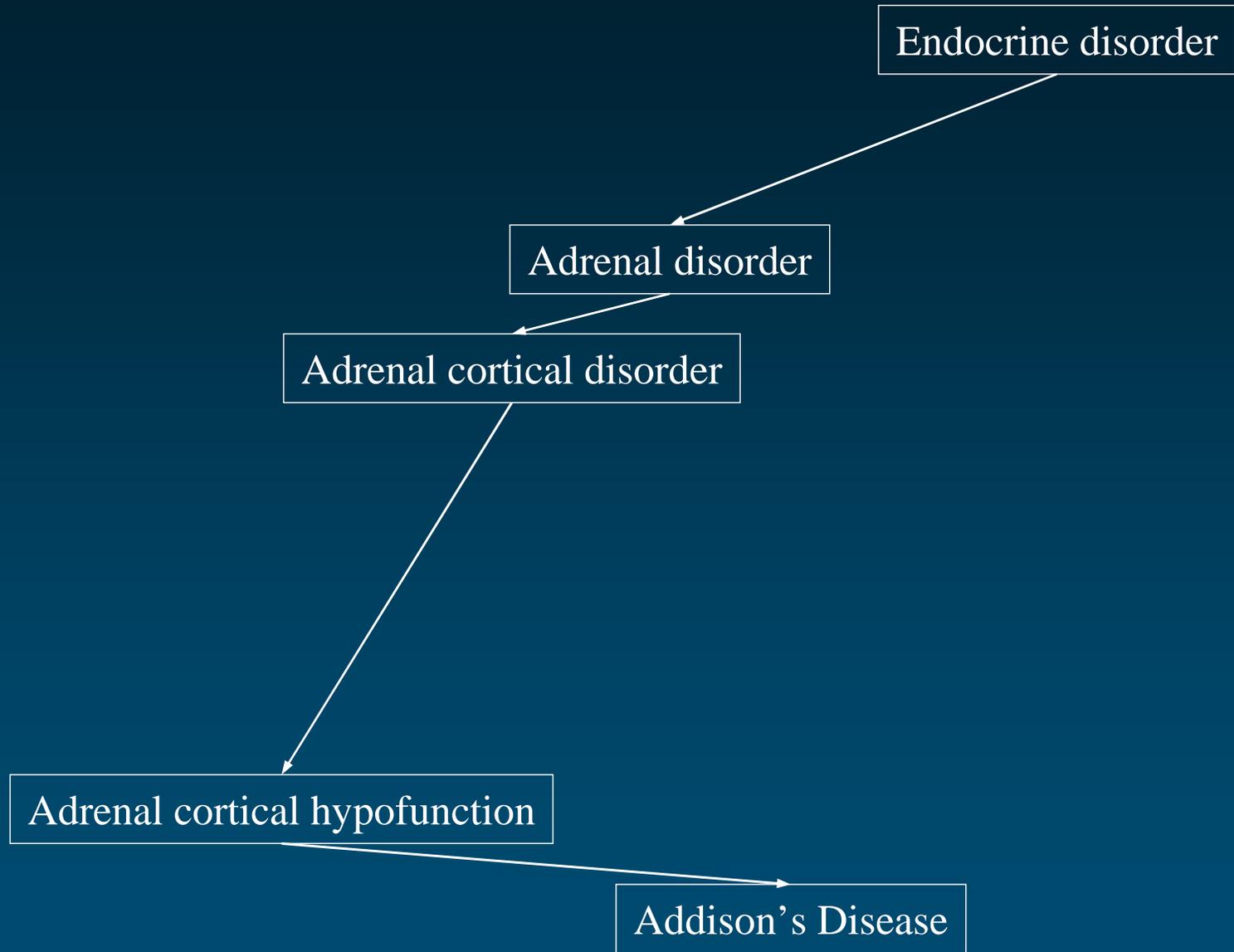
Addison's disease

SNOMED International

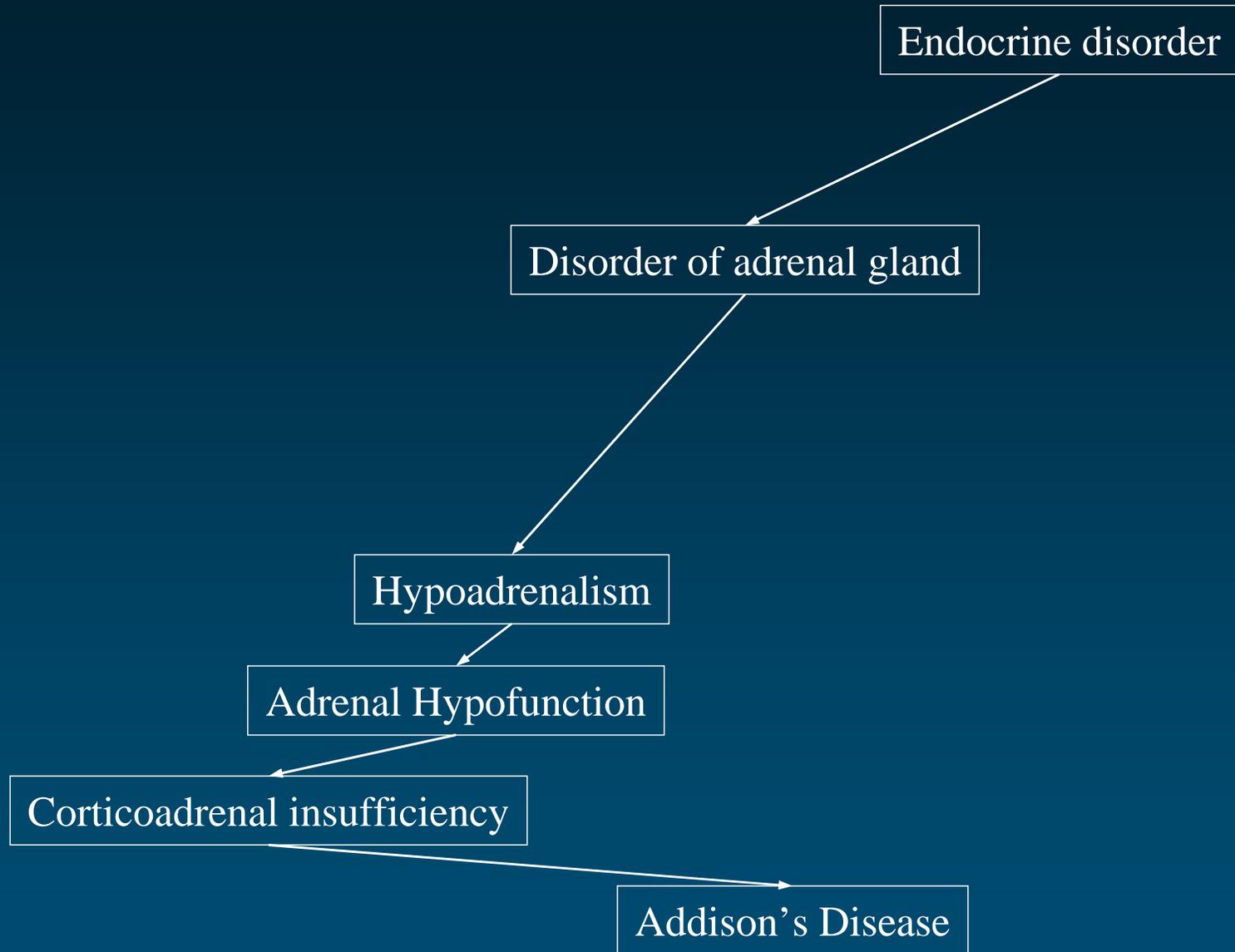




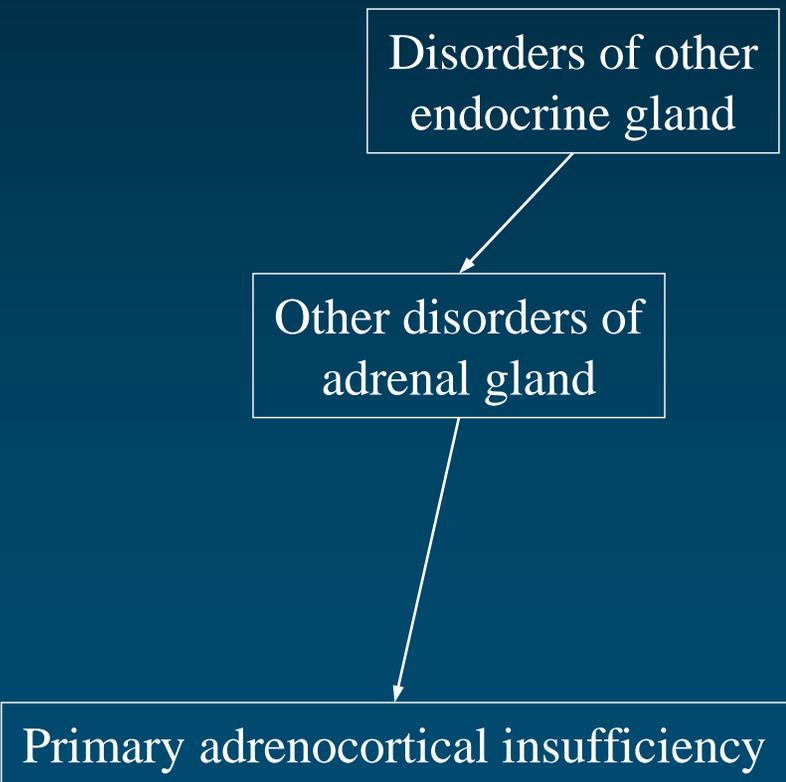
AOD



Read Codes

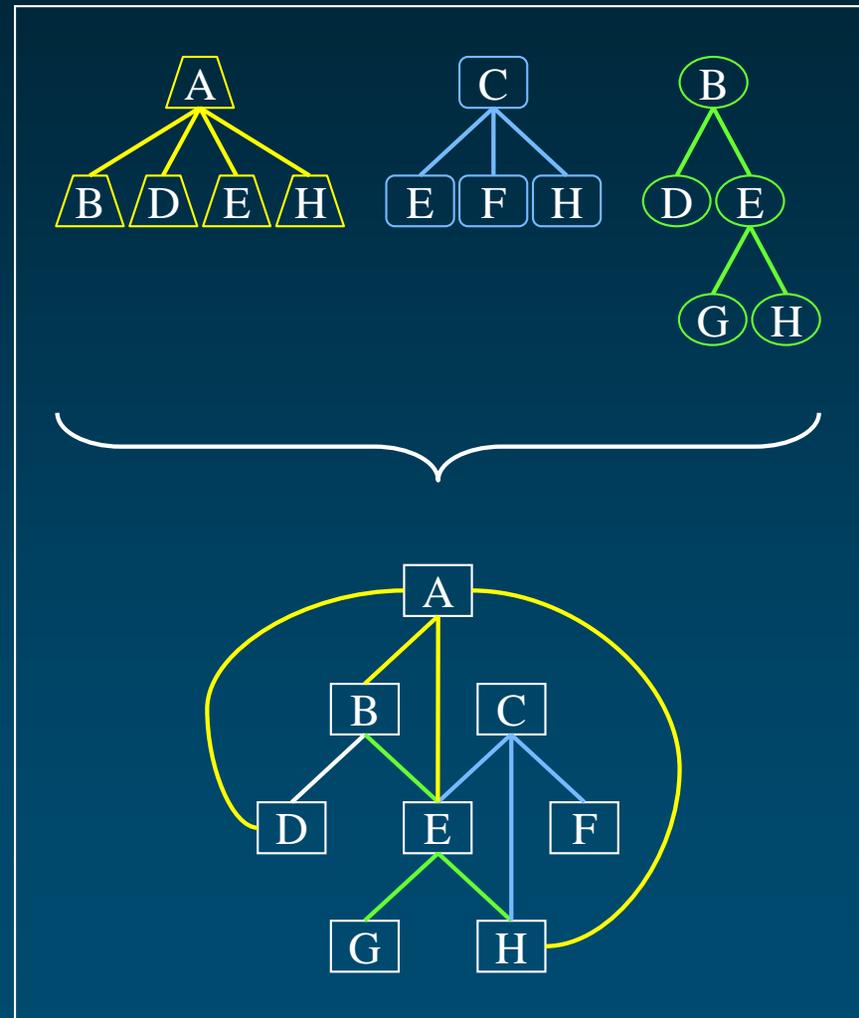


ICD-10



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

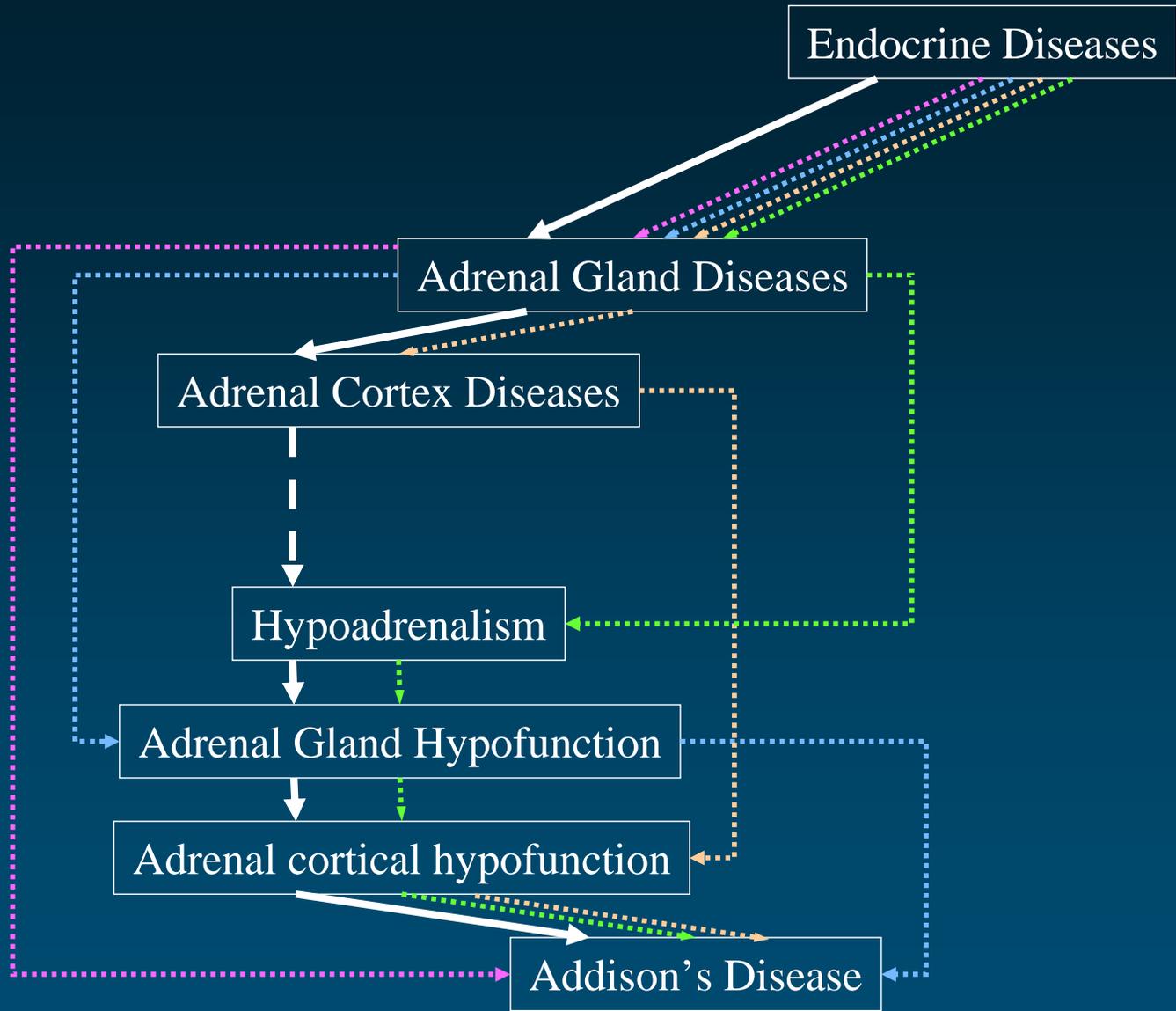
SNOMED

MeSH

AOD

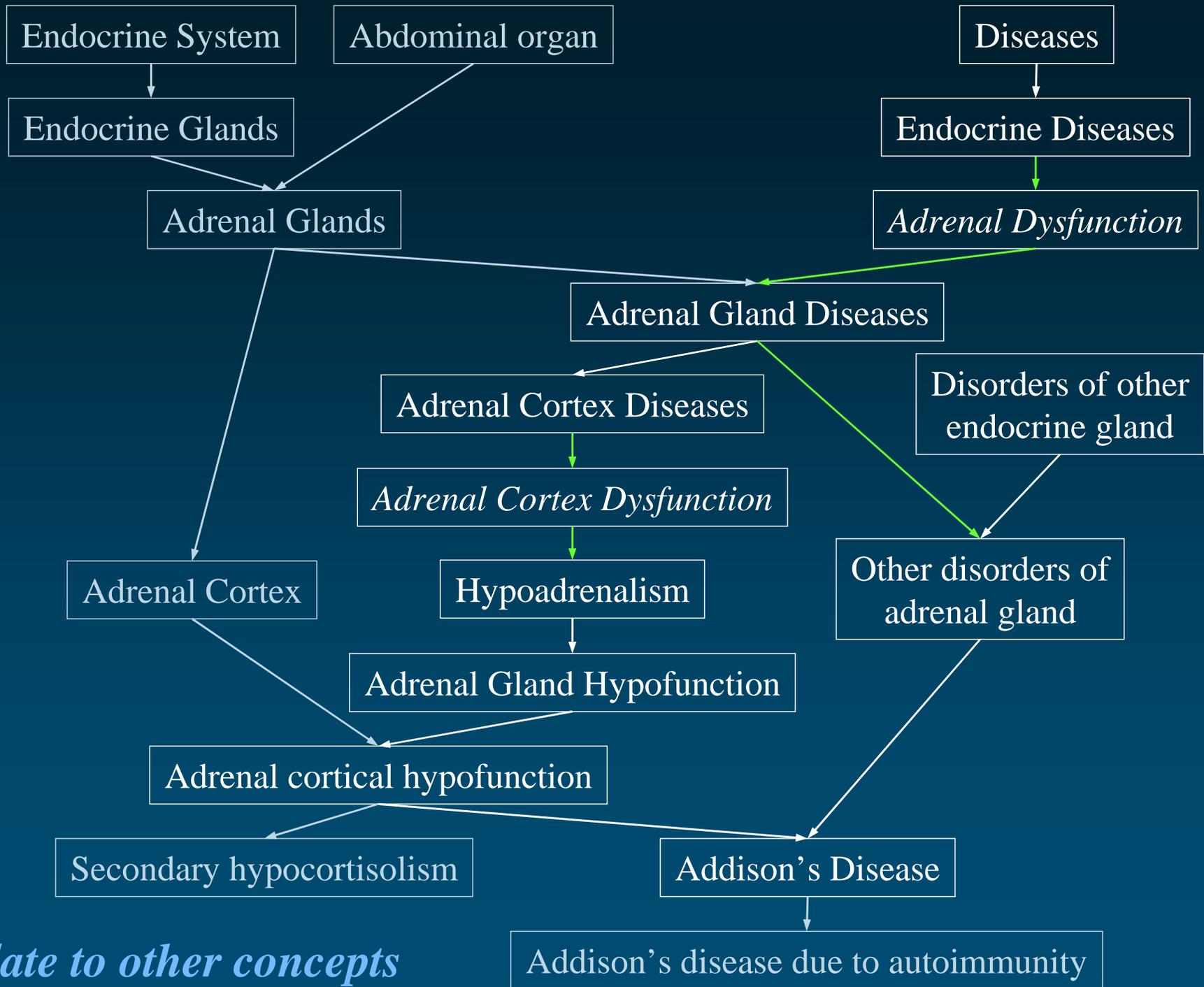
Read Codes

UMLS



Relate to other concepts

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

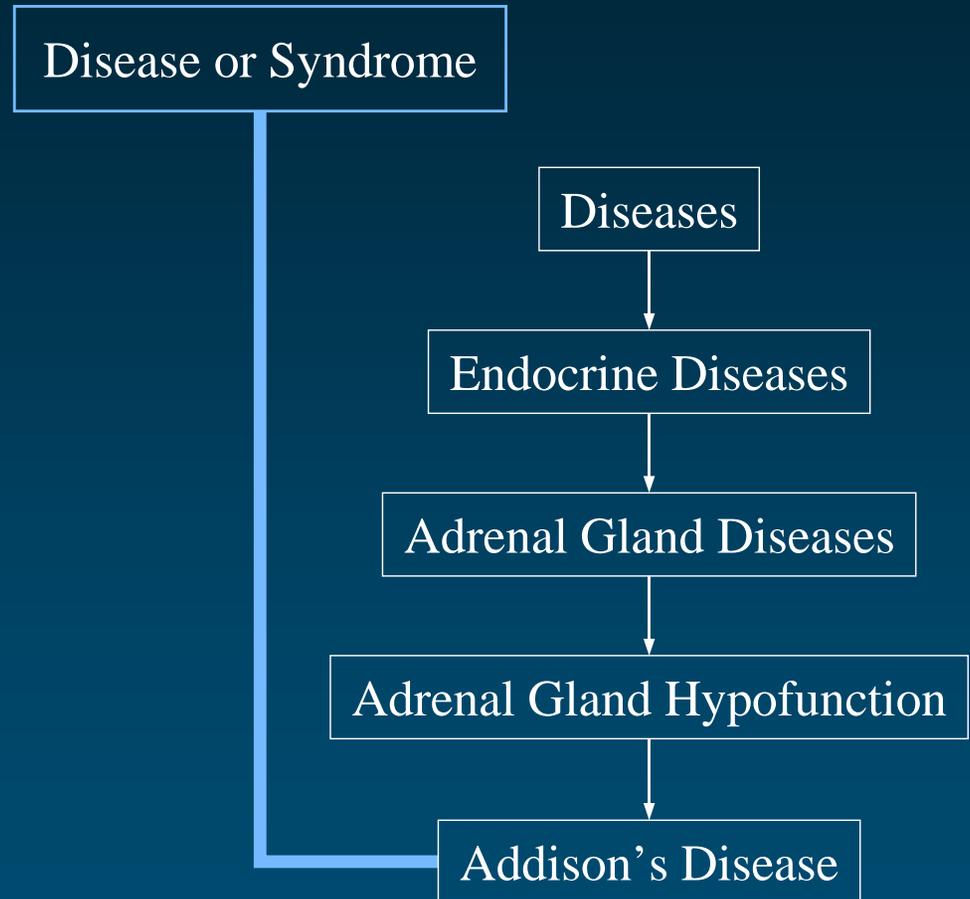


relate to other concepts

Addison's disease due to autoimmunity

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

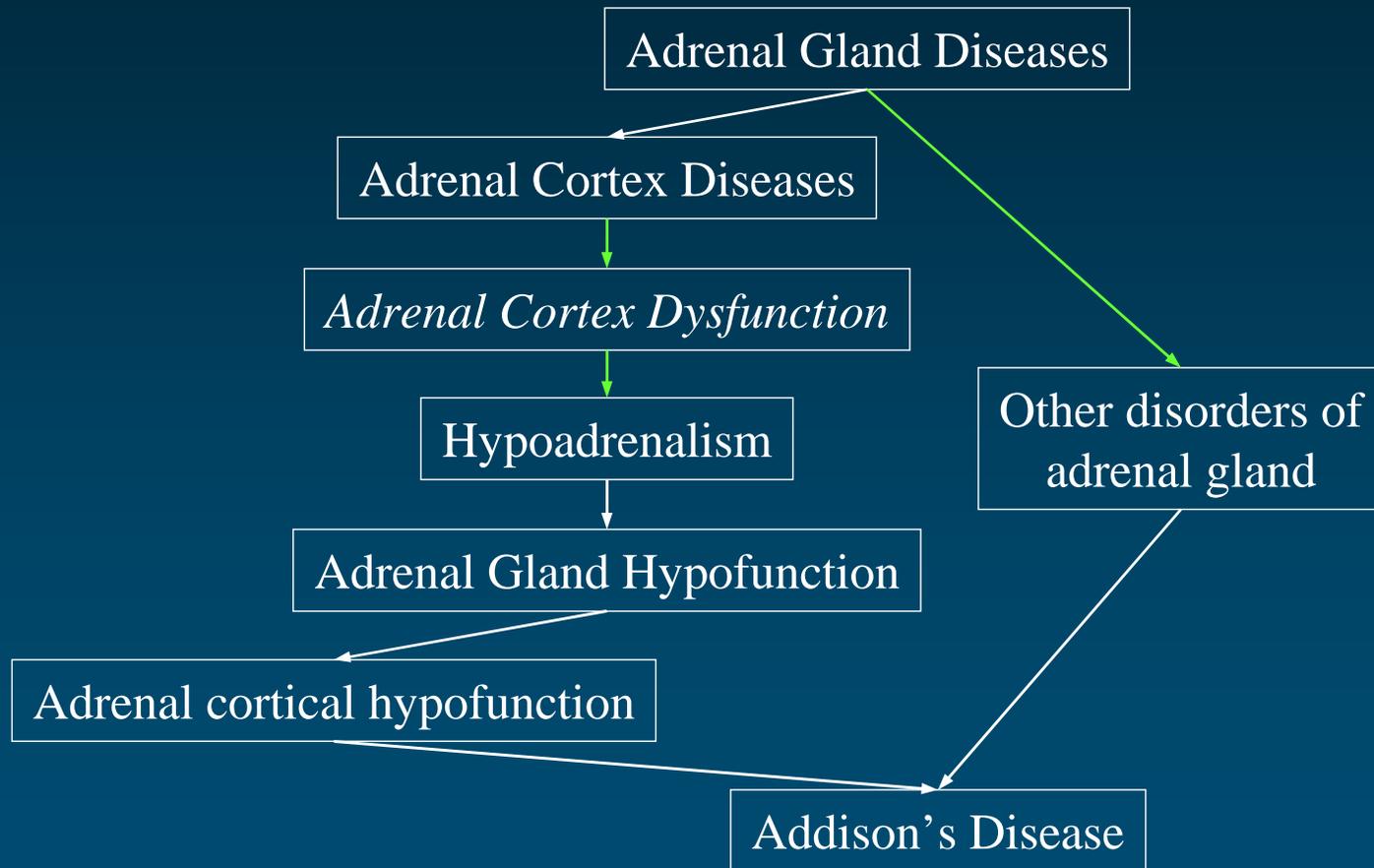
Diseases of the adrenal glands

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



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 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

(2005AB)

- ◆ 133 source vocabularies contributing concept names
- ◆ ~80 families of vocabularies
 - multiple translations (e.g., MeSH, ICPC, ICD-10)
 - variants (American-English equivalents, Australian extension/adaptation)
 - subsequent editions usually considered distinct families (ICD: 9-10; DSM: IIR-IV)
- ◆ Broad coverage of biomedicine
- ◆ Common presentation



Biomedical terminologies

◆ General vocabularies

- anatomy (UWDA, Neuronames)
- drugs (RxNorm, First DataBank, Micromedex)
- medical devices (UMD, SPN)

◆ Several perspectives

- clinical terms (SNOMED CT)
- information sciences (MeSH, CRISP)
- administrative terminologies (ICD-9-CM, CPT-4)
- data exchange terminologies (HL7, LOINC)

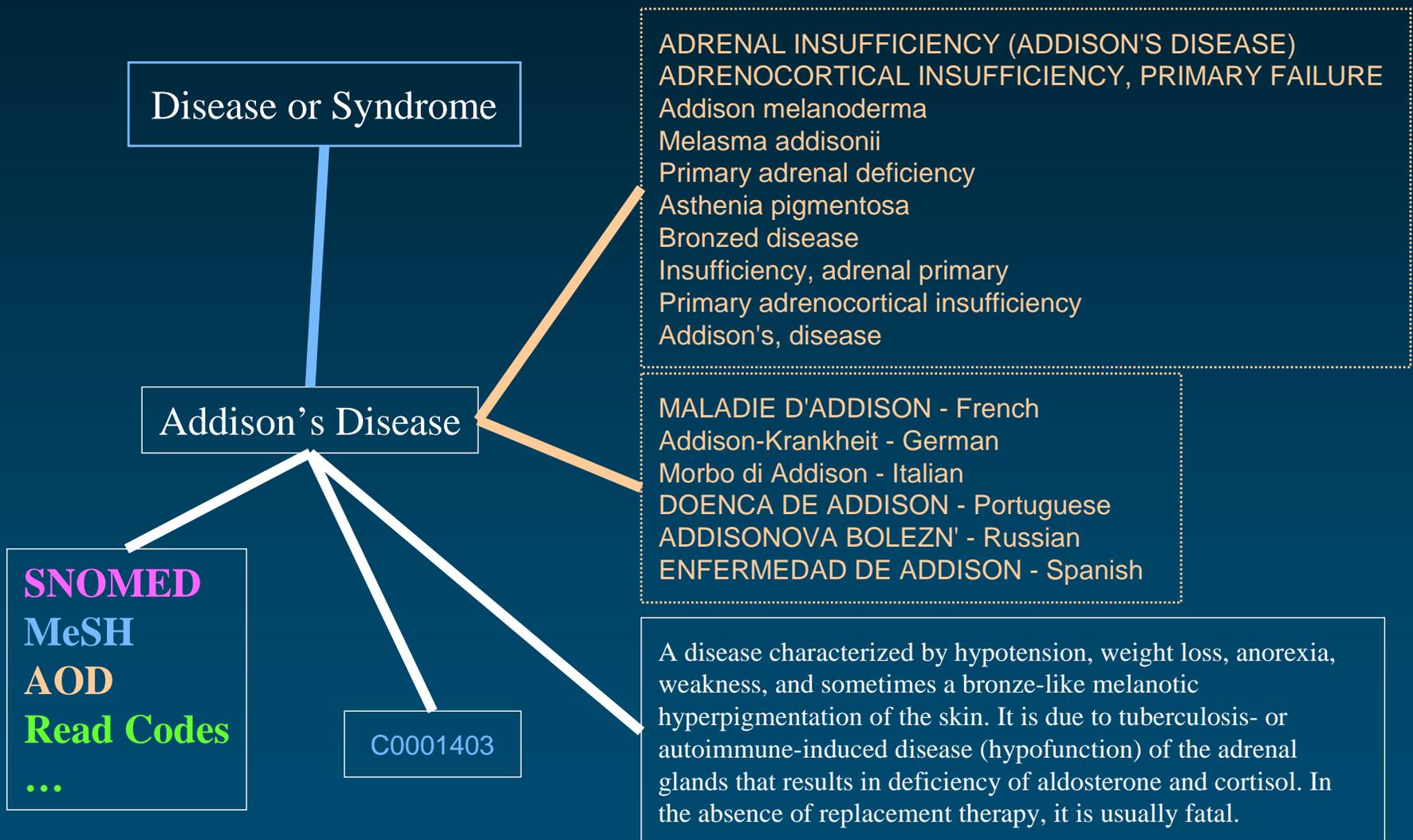
Biomedical terminologies (cont'd)

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



The UMLS serves as a vehicle for the regulatory standards (HIPAA, CHI)

Addison's Disease: Concept



Metathesaurus Concepts

(2005AB)

- ◆ Concept (~ 1.2 M) CUI
 - Set of synonymous concept names
- ◆ Term (~ 4.2 M) LUI
 - Set of normalized names
- ◆ String (~ 4.8 M) SUI
 - Distinct concept name
- ◆ Atom (~ 5.6 M) AUI
 - Concept name in a given source

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

A0000003 Headache (source 1)
A0000004 Headache (source 2)
S0000002

L0000001

A0000005 Cephalgia (source 1)
S0000003

L0000002

C0000001



Cluster of synonymous terms

Concept
C0001403

Term L0001403	<p>S0354372 <i>Addison's disease</i></p> <p>S0010792 Addison Disease</p> <p>S0010794 Addison's Disease</p> <p>S0010796 Addisons Disease</p> <p>S0033587 Disease, Addison</p> <p>S0352253 ADDISON'S DISEASE</p>	[...]
Term L2799243	S3341310 <i>Addison's disease (disorder)</i>	
Term L0494940	<p>S5907336 <i>Primary Adrenocortical Insufficiency</i></p> <p>S5901878 Insufficiencies, Primary Adrenocortical</p>	[...]
Term L0494851	<p>S5907334 <i>Primary Adrenal Insufficiency</i></p> <p>S4094828 adrenal; insufficiency, primary</p>	[...]
Term L0585243	<p>S5907343 <i>Primary Hypoadrenalism</i></p> <p>S5901432 Hypoadrenalism, Primary</p>	[...]
Term L1229627	S1471573 <i>Addison-Krankheit</i>	GER
Term L5345155	S6107160 <i>Maladie d'Addison</i>	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 Relationships

- ◆ 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: 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

Symbolic relationships Type

◆ Hierarchical

- Parent / Child
- Broader / Narrower than

PAR/CHD

RB/RN



◆ Derived from hierarchies

- Siblings (children of parents)

SIB



◆ Associative

- Other

RO



◆ Various flavors of near-synonymy

- Similar
- Source asserted synonymy
- Possible synonymy

RL

SY

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

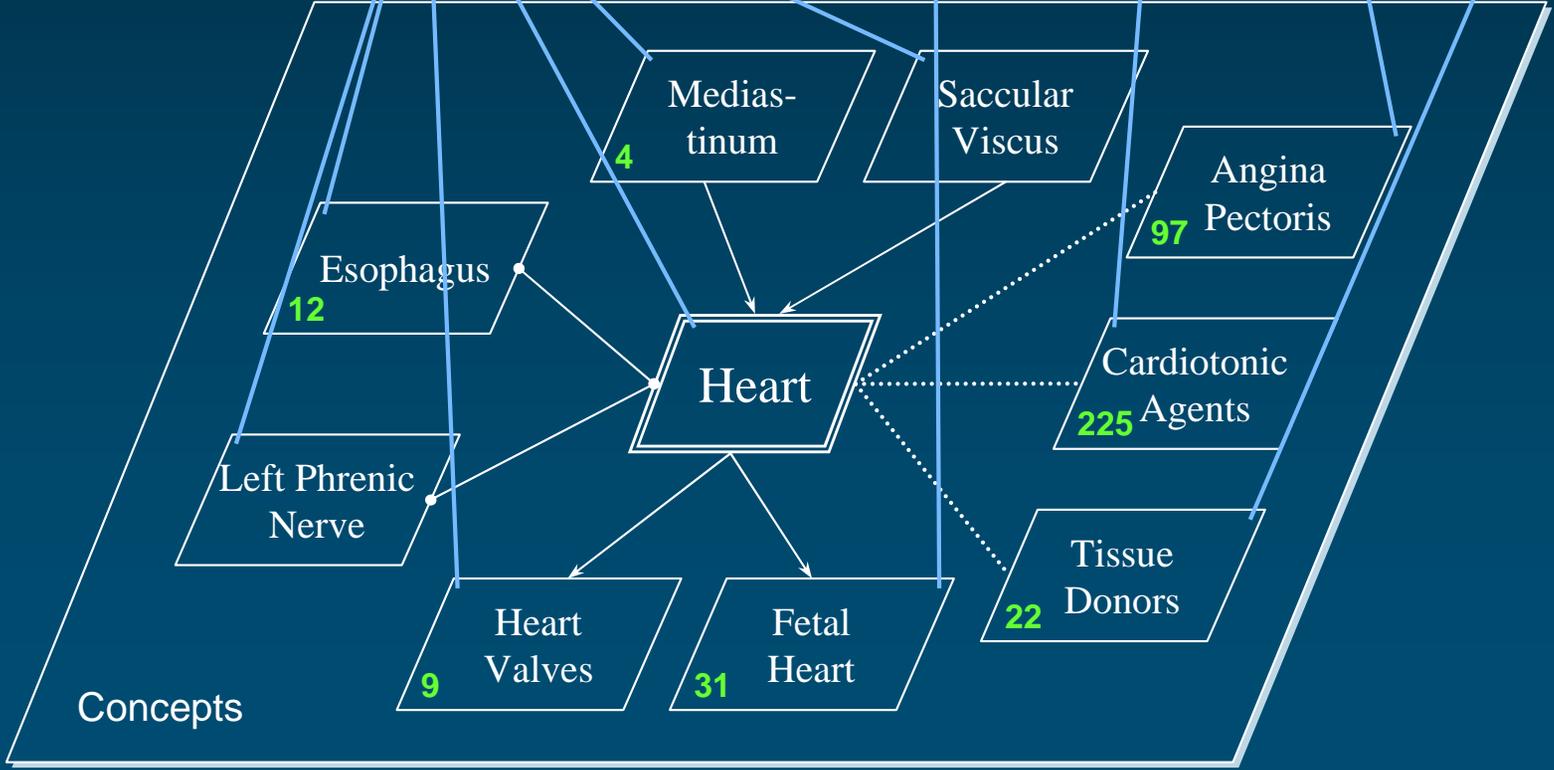
Disease or Syndrome

Pharmacologic Substance

Population Group

Semantic Network

Metathesaurus



Concepts

Esophagus
12

Mediastinum
4

Saccular Viscus

Heart

Left Phrenic Nerve

Heart Valves
9

Fetal Heart
31

Angina Pectoris
97

Cardiotonic Agents
225

Tissue Donors
22

UMLS Semantic Network

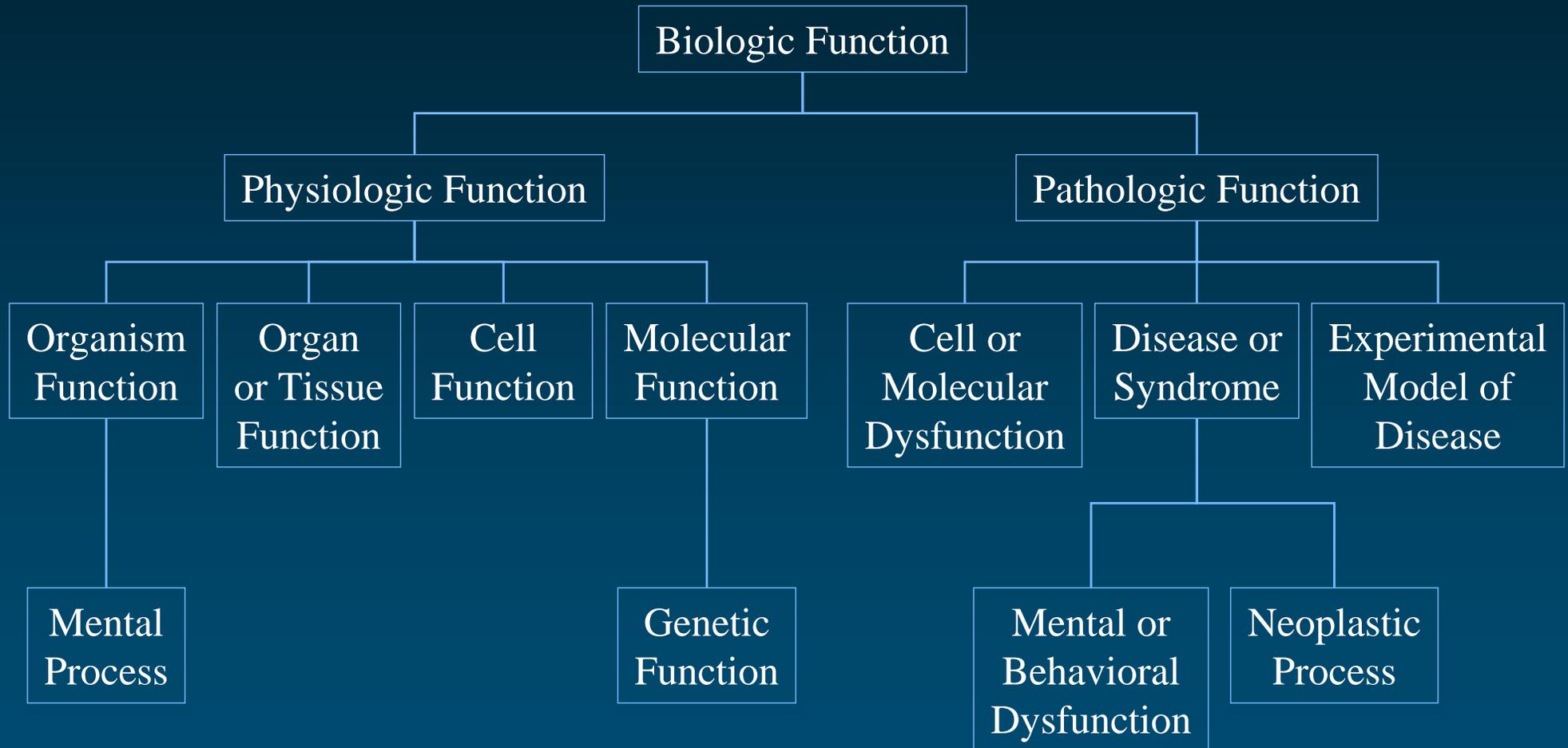
Semantic Network

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

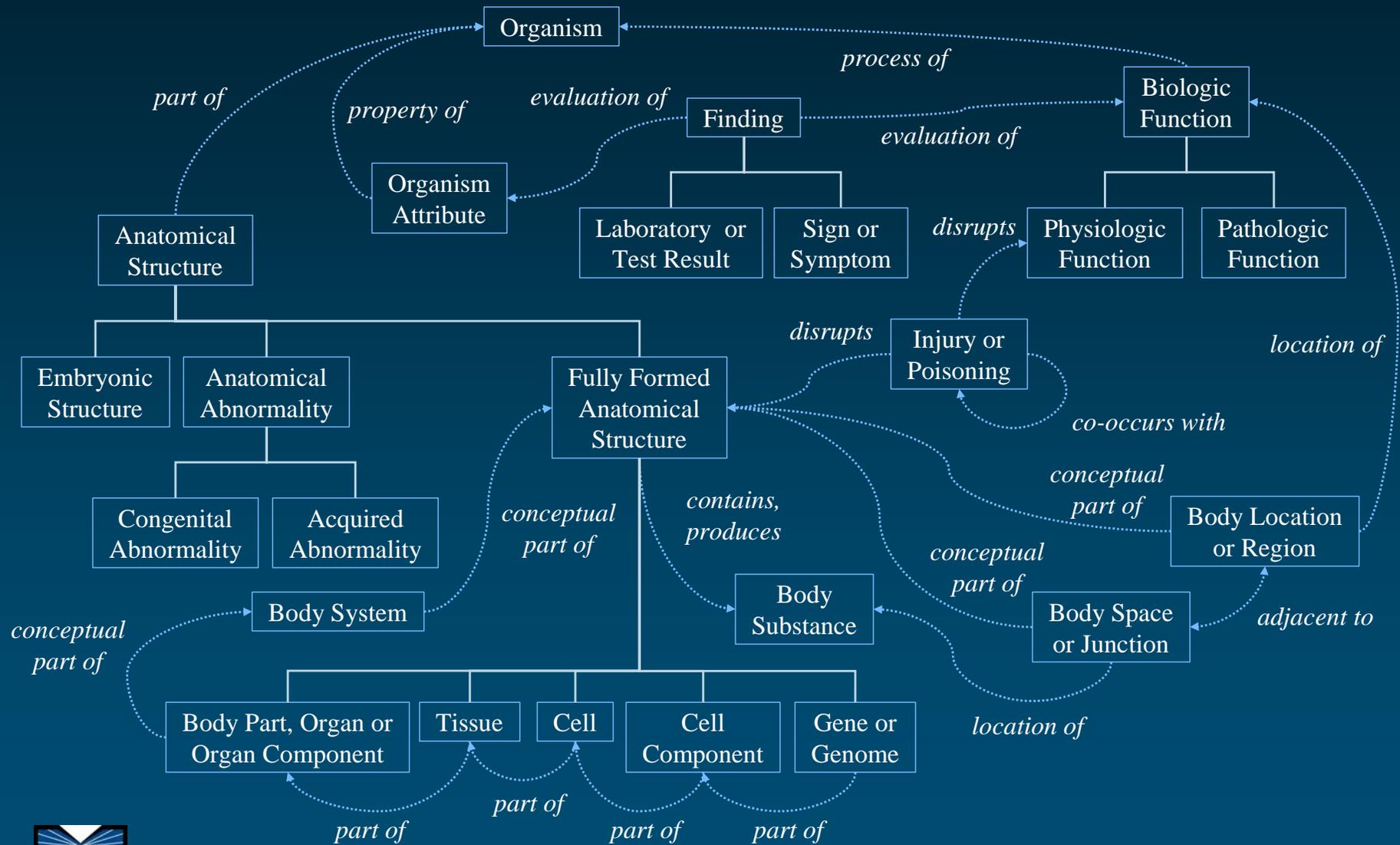
Semantic Network

- ◆ 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)



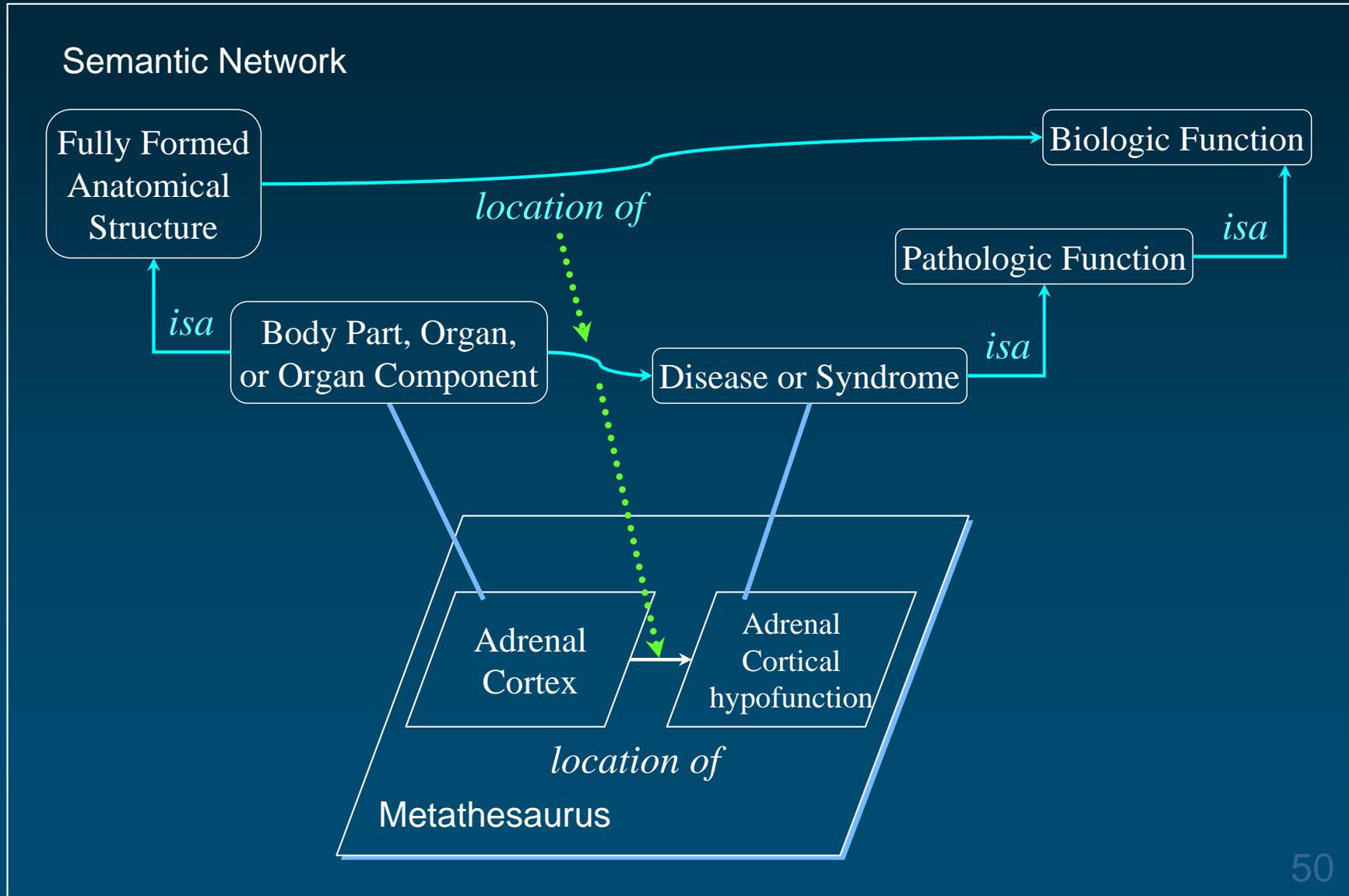
Associative (non-isa) relationships



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



SPECIALIST Lexicon and lexical tools

SPECIALIST Lexicon

- ◆ Content
 - English lexicon
 - Many words from the biomedical domain
- ◆ 200,000+ lexical items
- ◆ Word properties
 - morphology
 - orthography
 - syntax
- ◆ Used by the lexical tools

Morphology

◆ Inflection

- noun nucleus, nuclei
- verb cauterize, cauterizes, cauterized, cauterizing
- adjective red, redder, reddest

◆ Derivation

- verb ↔ noun cauterize -- cauterization
- adjective ↔ noun red -- redness

Orthography

◆ Spelling variants

- oe/e

oesophagus - esophagus

- ae/e

anaemia - anemia

- ise/ize

cauterise - cauterize

- genitive mark

Addison's disease

Addison disease

Addisons disease

Syntax

◆ Complementation

● verbs

- intransitive I'll treat.
- transitive He treated the patient.
- ditransitive He treated the patient with a drug.

● nouns

- prepositional phrase

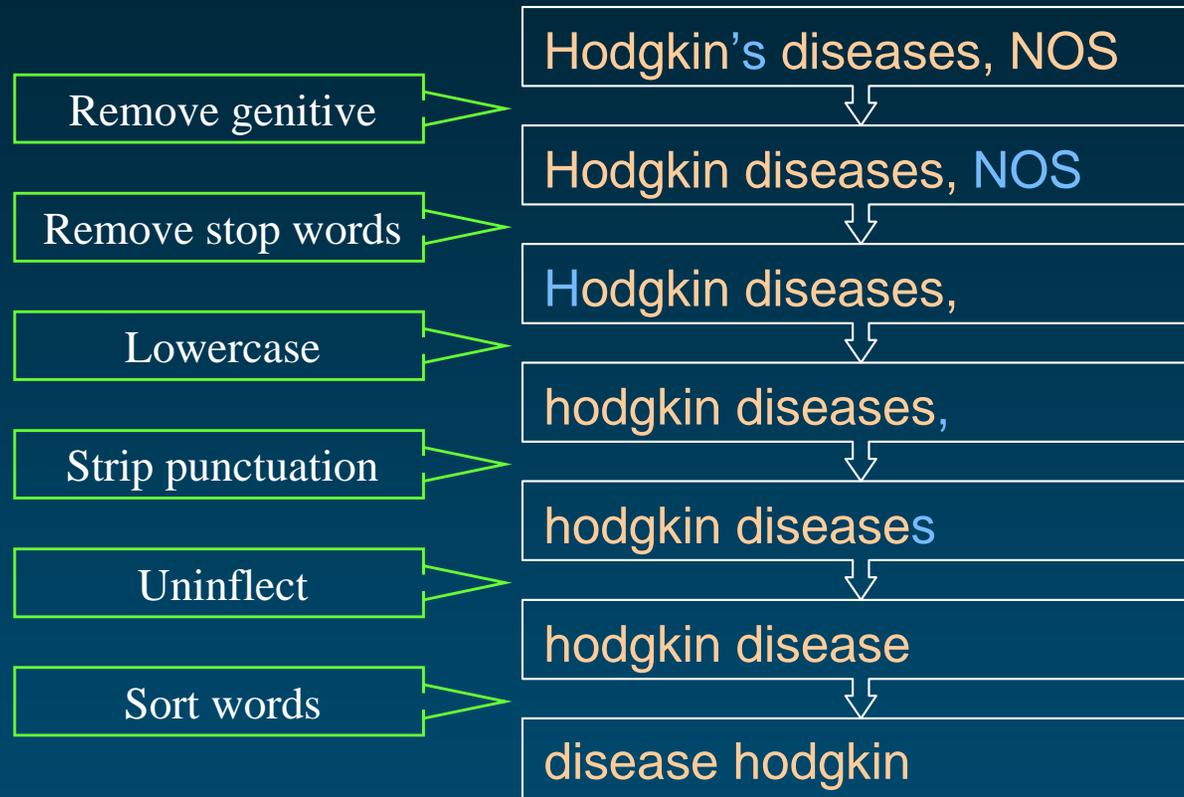
Valve of coronary sinus

◆ Position for adjectives

Lexical tools

- ◆ To manage lexical variation in biomedical terminologies
- ◆ Major tools
 - Normalization
 - Indexes
 - Lexical Variant Generation program (lvg)
- ◆ Based on the SPECIALIST Lexicon
- ◆ Used by noun phrase extractors, search engines

Normalization



Normalization: Example

Hodgkin Disease
HODGKINS DISEASE
Hodgkin's Disease
Disease, Hodgkin's
Hodgkin's, disease
HODGKIN'S DISEASE
Hodgkin's disease
Hodgkins Disease
Hodgkin's disease NOS
Hodgkin's disease, NOS
Disease, Hodgkins
Diseases, Hodgkins
Hodgkins Diseases
Hodgkins disease
hodgkin's disease
Disease, Hodgkin

normalize

disease hodgkin

Normalization Applications

- ◆ Model for lexical resemblance
- ◆ Help find lexical variants for a term
 - Terms that normalize the same usually share the same LUI
- ◆ Help find candidates to synonymy among terms
- ◆ Help map input terms to UMLS concepts

Indexes

- ◆ Word index
 - word to Metathesaurus strings
 - one word index per language
- ◆ Normalized word index
 - normalized word to Metathesaurus strings
 - English only
- ◆ Normalized string index
 - normalized term to Metathesaurus strings
 - English only

Lexical Variant Generation program

- ◆ Tool for specialists (linguists)
- ◆ Performs atomic lexical transformations
 - generating inflectional variants
 - lowercase
 - ...
- ◆ Performs sequences of atomic transformations
 - a specialized sequence of transformations provides the normalized form of a term (the *norm* program)

UMLS in action
MetaMap

MetaMap Motivation

[Aronson, *AMIA*, 2001]

- ◆ Term extraction
 - Identifying UMLS concepts from text
- ◆ Usage
 - Information indexing and retrieval
 - Knowledge extraction / discovery
 - Semantic interpretation
- ◆ Characteristics
 - Linguistic approach
 - Based on UMLS knowledge sources

MetaMap Methods

◆ Parsing

- Shallow syntactic analysis
- SPECIALIST lexicon
- Xerox part-of-speech tagger

◆ Variant generation

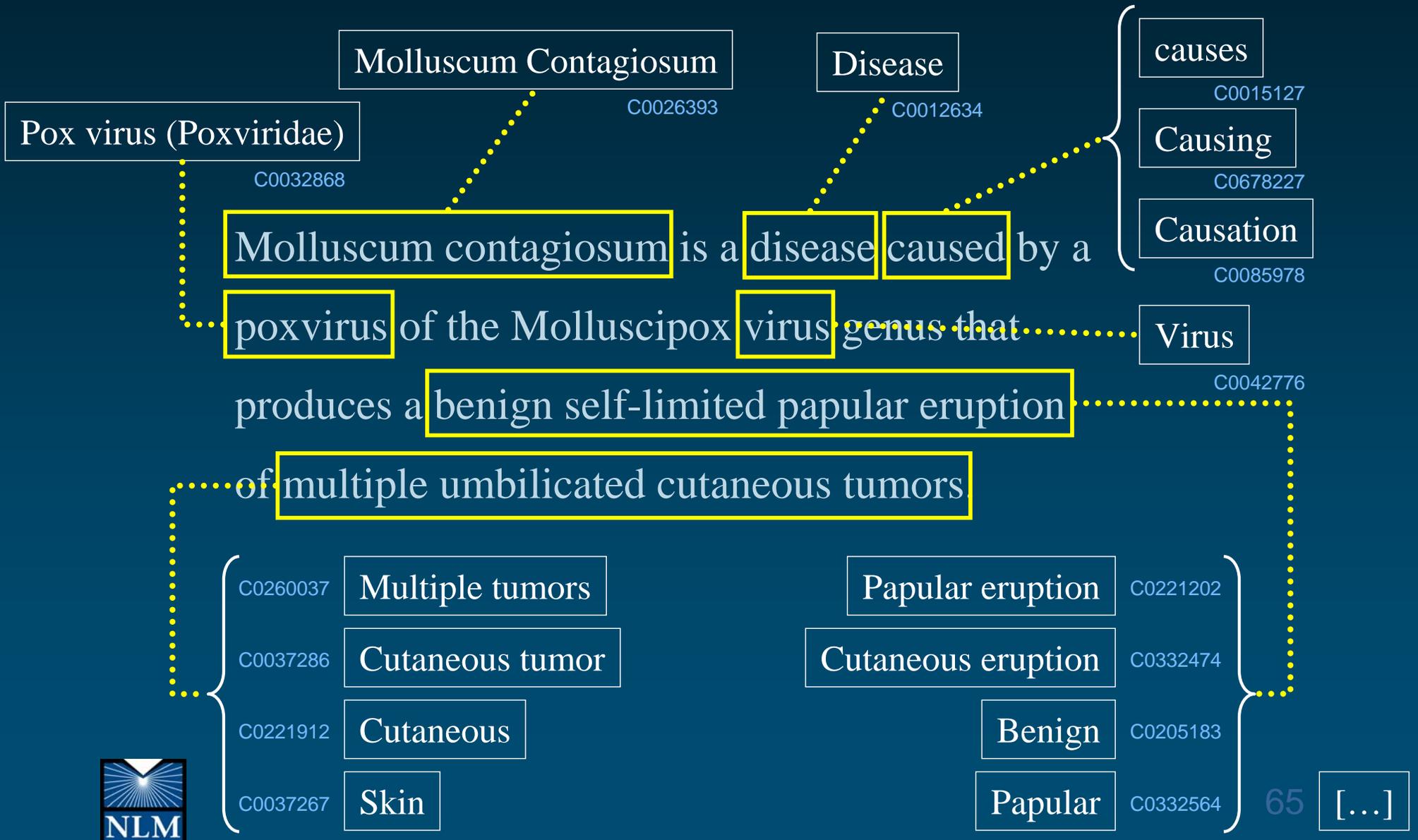
◆ Candidate retrieval

- Retrieve candidate terms containing at least one variant

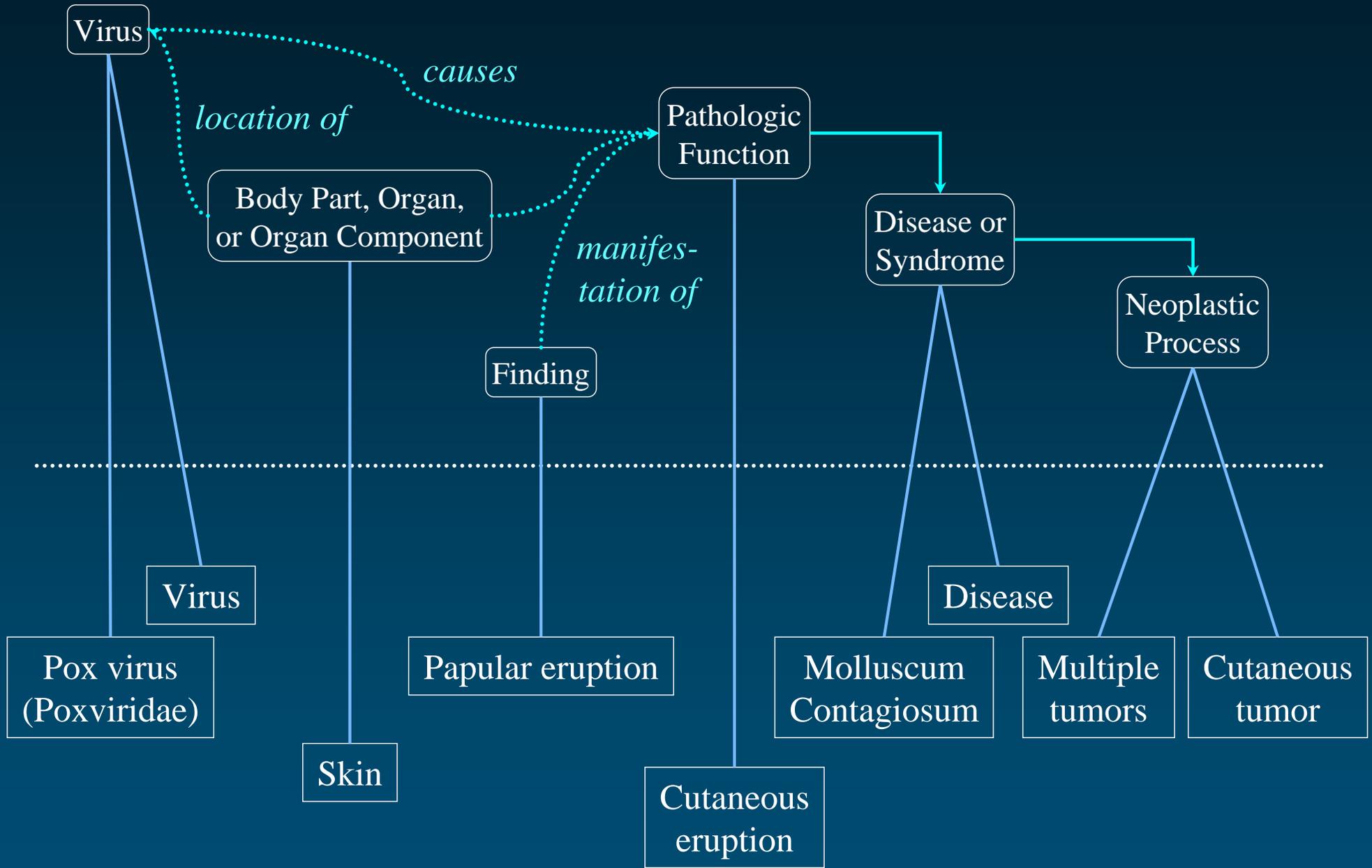
◆ Candidate evaluation

- Rank candidate terms with respect to closeness to input text (centrality, variation, coverage, and cohesiveness)

MetaMap Example



Semantic Network



Using MetaMap MMTx

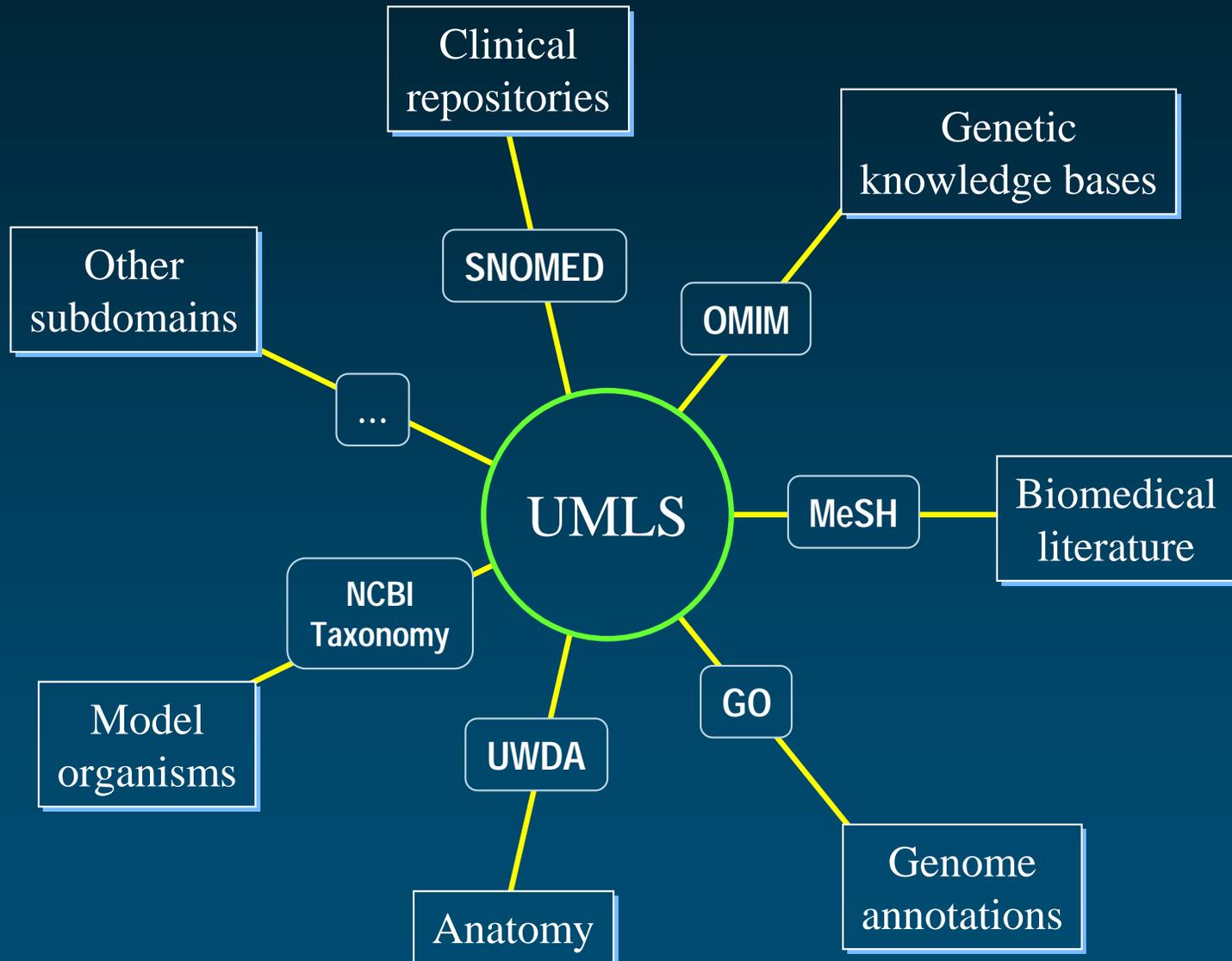
- ◆ Requires UMLS license
- ◆ Local implementation (Java-based)
- ◆ Provides
 - Stand-alone application
 - API for integrating in other applications

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

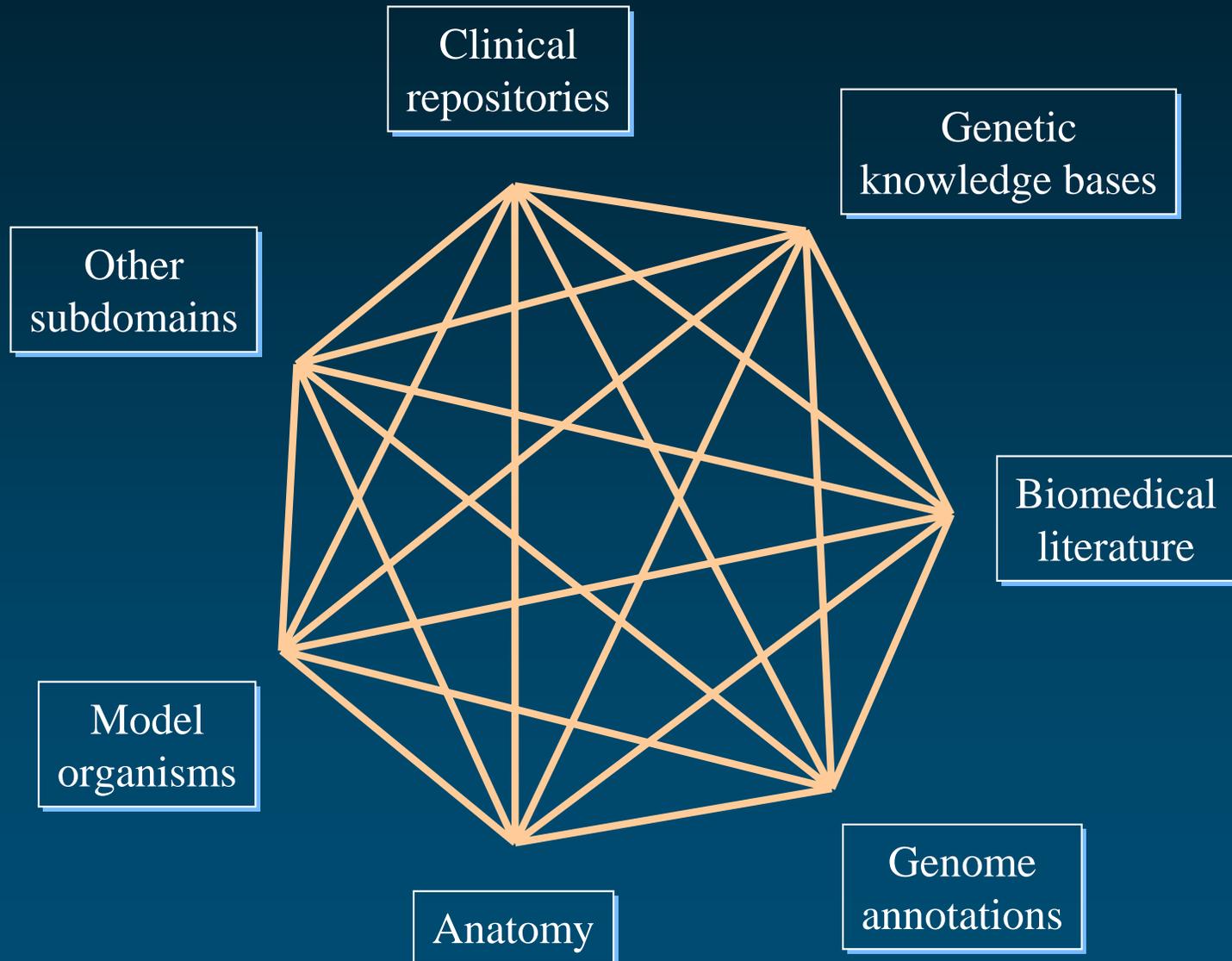


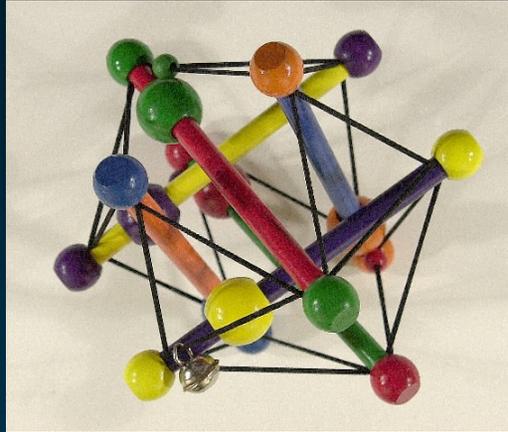
Conclusions

Integrating subdomains



Integrating subdomains





Medical Ontology Research

Contact: olivier@nlm.nih.gov

Web: mor.nlm.nih.gov



Olivier Bodenreider

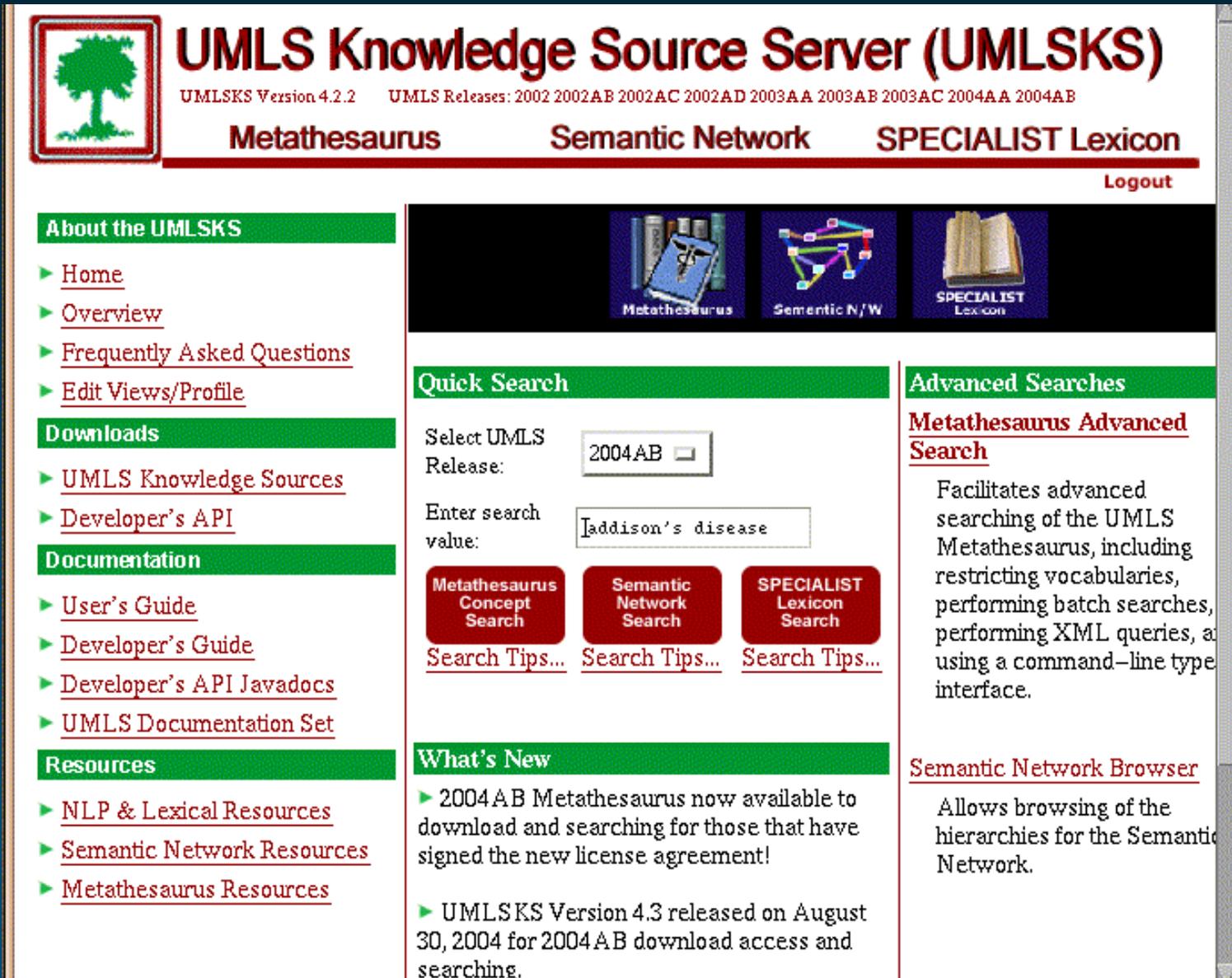
Lister Hill National Center
for Biomedical Communications
Bethesda, Maryland - USA

Appendix

Knowledge Source Server *Web Interface*

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

UMLS Knowledge Source Server Home Page



The screenshot shows the UMLS Knowledge Source Server (UMLSKS) home page. At the top left is a logo of a tree. The main title is "UMLS Knowledge Source Server (UMLSKS)" in large red font. Below it, the version "UMLSKS Version 4.2.2" and release dates "UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB" are listed. Three navigation tabs are visible: "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". A "Logout" link is in the top right. The left sidebar contains several menu sections: "About the UMLSKS" with links to Home, Overview, Frequently Asked Questions, and Edit Views/Profile; "Downloads" with links to UMLS Knowledge Sources and Developer's API; "Documentation" with links to User's Guide, Developer's Guide, Developer's API Javadocs, and UMLS Documentation Set; and "Resources" with links to NLP & Lexical Resources, Semantic Network Resources, and Metathesaurus Resources. The main content area features three icons: "Metathesaurus" (books), "Semantic N/W" (network diagram), and "SPECIALIST Lexicon" (open book). Below these are three search sections: "Quick Search" with a dropdown for "2004AB" and a search input field containing "Addison's disease"; "Advanced Searches" with a link to "Metathesaurus Advanced Search" and a description of advanced search capabilities; and "What's New" with two bullet points: "2004AB Metathesaurus now available to download and searching for those that have signed the new license agreement!" and "UMLSKS Version 4.3 released on August 30, 2004 for 2004AB download access and searching." There are also buttons for "Metathesaurus Concept Search", "Semantic Network Search", and "SPECIALIST Lexicon Search", each with a "Search Tips..." link. A "Semantic Network Browser" section is partially visible at the bottom right, describing browsing hierarchies.

UMLS Knowledge Source Server (UMLSKS)
UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

Metathesaurus Semantic Network SPECIALIST Lexicon

[Logout](#)

About the UMLSKS

- [Home](#)
- [Overview](#)
- [Frequently Asked Questions](#)
- [Edit Views/Profile](#)

Downloads

- [UMLS Knowledge Sources](#)
- [Developer's API](#)

Documentation

- [User's Guide](#)
- [Developer's Guide](#)
- [Developer's API Javadocs](#)
- [UMLS Documentation Set](#)

Resources

- [NLP & Lexical Resources](#)
- [Semantic Network Resources](#)
- [Metathesaurus Resources](#)

Quick Search

Select UMLS Release:

Enter search value:

Metathesaurus Concept Search **Semantic Network Search** **SPECIALIST Lexicon Search**

[Search Tips...](#) [Search Tips...](#) [Search Tips...](#)

Advanced Searches

[Metathesaurus Advanced Search](#)

Facilitates advanced searching of the UMLS Metathesaurus, including restricting vocabularies, performing batch searches, performing XML queries, and using a command-line type interface.

[Semantic Network Browser](#)

Allows browsing of the hierarchies for the Semantic Network.

What's New

- ▶ 2004AB Metathesaurus now available to download and searching for those that have signed the new license agreement!
- ▶ UMLSKS Version 4.3 released on August 30, 2004 for 2004AB download access and searching.

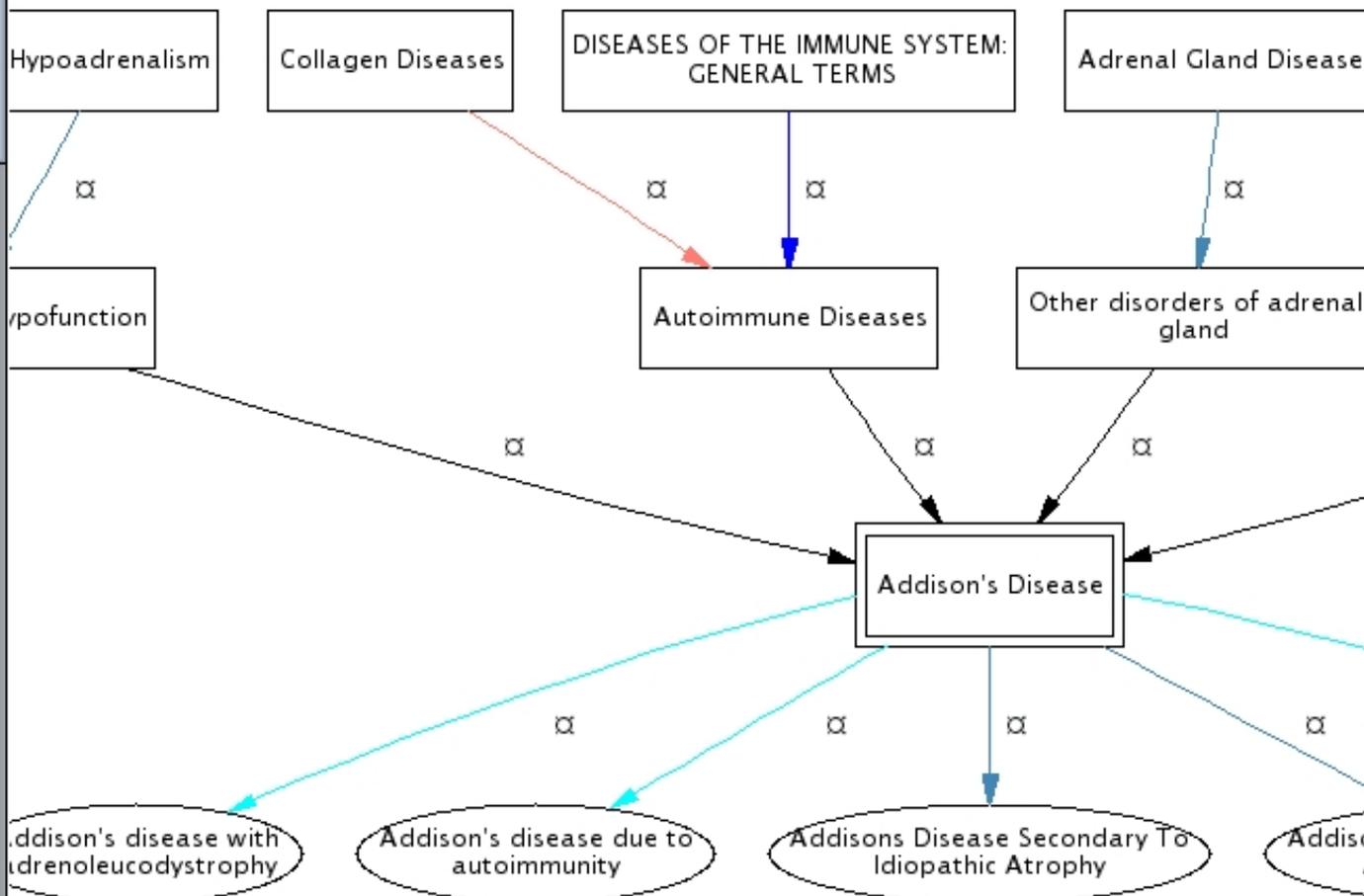
Siblings

Concepts & Ideas

- Clinical Syndromes ☒

Disorders

- Acquired Immunodeficiency Syndrome ☒
- Acute adrenal insufficiency ☒
- Addisonian crisis ☒
- Adrenal atrophy ☒
- Adrenal calcification ☒
- Adrenal hemorrhage ☒
- Adrenal infarction ☒
- Adrenal insufficiency due to adrenal metastasis ☒
- Adrenogenital Syndrome ☒
- Allergic arthritis ☒
- Angelman Syndrome ☒
- Asperger syndrome <1> ☒
- Autoerythrocyte sensitivity



Other Related Concepts

Disorders

- Addisonian crisis ☒
- Autoimmune Syndrome Type II, Polyglandular ☒
- Tuberculosis ☒
- Tuberculosis of adrenal glands ☒
- Tuberculous Addison's disease ☒

(5 other related)

Co-occurring Concepts

Anatomy

- Adrenal Cortex [14] ☒
- Adrenal Glands [17] ☒
- Liver [2] ☒
- Tears body substance [2] ☒
- X Chromosome [3]

Chemicals & Drugs

BCI

Addison's Disease

LEGEND *

Start again

Apply new parameters

Restrict to vocabulary: Show all
Highlight vocabulary: Nothing
UMLS data: UMLS_2002
Type of hierarchical: All Parent/Child only

Similar Concepts

- Adrenal cortical hypofunction ☒

(1 concept)

Closest MeSH Terms

Main Headings

- Addison's Disease

Knowledge Source Server
Application Programming Interface

UMLS KS API basics

- ◆ Remote server at NLM
- ◆ Local application connected through

Java RMI

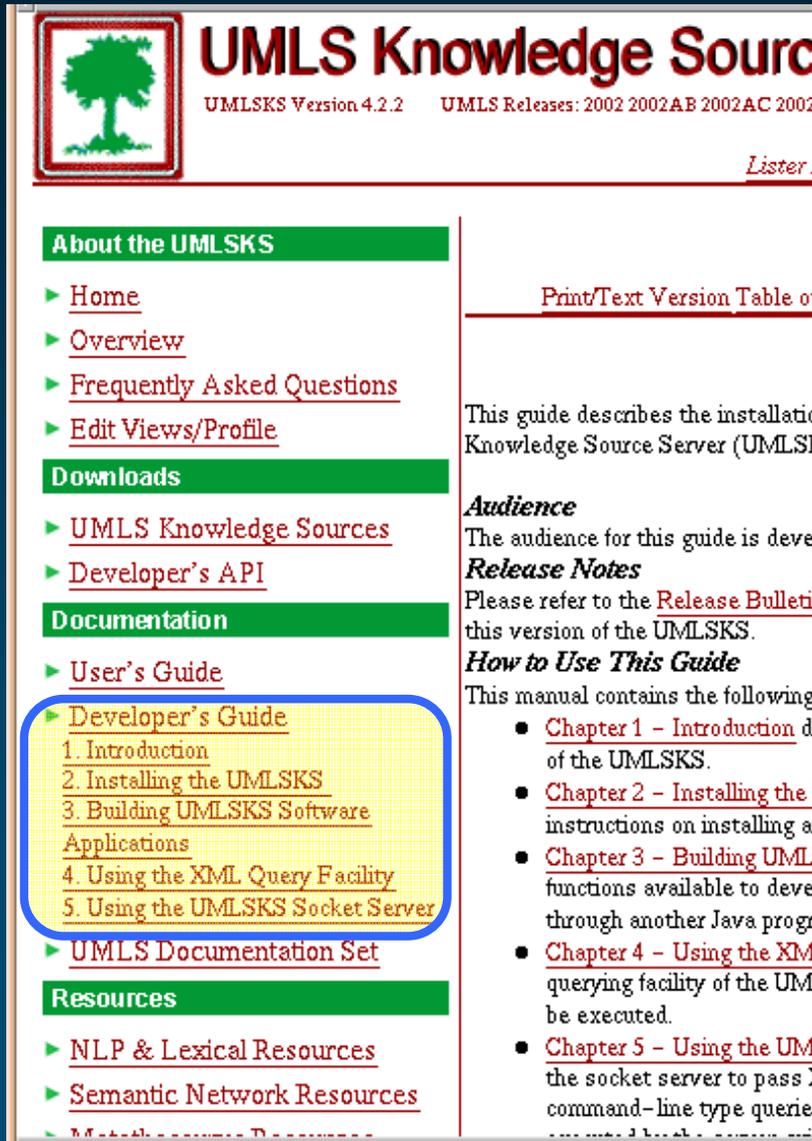
- ◆ Java-based applications
- ◆ Developer's Guide: Chapter 3
- ◆ Set of Java classes (part of the UMLS KS API download)
- ◆ Detailed *Javadoc* documentation online and with API download

TCP/IP socket

- ◆ XML-based queries
- ◆ Developer's Guide: Chapter 5
- ◆ XML schema
- ◆ Socket server
 - Host: umlsks.nlm.nih.gov
 - Port: 8042



Developer's Guide



UMLS Knowledge Source
UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002AD

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This guide describes the installation of the Knowledge Source Server (UMLSKS).

Audience

The audience for this guide is developers of UMLSKS applications using the UMLSKS API.

Release Notes

Please refer to the [Release Bulletin](#) for a detailed list of features, bug fixes, and known problems with this version of the UMLSKS.

How to Use This Guide

This manual contains the following chapters:

- [Chapter 1 - Introduction](#) describes the basic features and architecture of the UMLSKS.
- [Chapter 2 - Installing the UMLSKS](#) provides administrators instructions on installing and tailoring a UMLSKS installation.
- [Chapter 3 - Building UMLSKS Software Applications](#) describes the functions available to developers wanting to interface to the UMLSKS through another Java program.
- [Chapter 4 - Using the XML Query Facility](#) describes how to use the querying facility of the UMLSKS wherein users build XML queries to be executed.
- [Chapter 5 - Using the UMLSKS Socket Server](#) describes how to use the socket server to pass XML formatted commands or command-line type queries (e.g. ks -meta -c aids) that are to be executed by the server with the results passed back to the client.

MetamorphoSys

What is MetamorphoSys?

- ◆ Tool distributed with the UMLS
- ◆ Multi-platform Java software
- ◆ The UMLS installation and customization wizard
 - Installs Knowledge Sources to local storage
 - Subsets and customizes a local Metathesaurus

Why use MetamorphoSys?

Customize the Metathesaurus

- ◆ To remove terminology that is unhelpful, or even harmful, to your needs and purposes
- ◆ To comply with terms of license agreement

Changing Default Settings

- ◆ To alter the preferred name
- ◆ To alter suppressibility of specific source term types

References

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.