



Medinfo 2007  
Tutorial S009  
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MEDINFO 2007

BRISBANE AUSTRALIA

# The Unified Medical Language System

*What is it and how to use it?*



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# Outline

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## ◆ What is the UMLS?

- Introduction
- Overview through an example
- The three UMLS Knowledge Sources

## ◆ How to use the UMLS?

- Obtaining a license
- Remote access
- Local installation and customization
- A UMLS-based algorithm
- Benefits and limitations

# Part I

What is the UMLS?

# Outline

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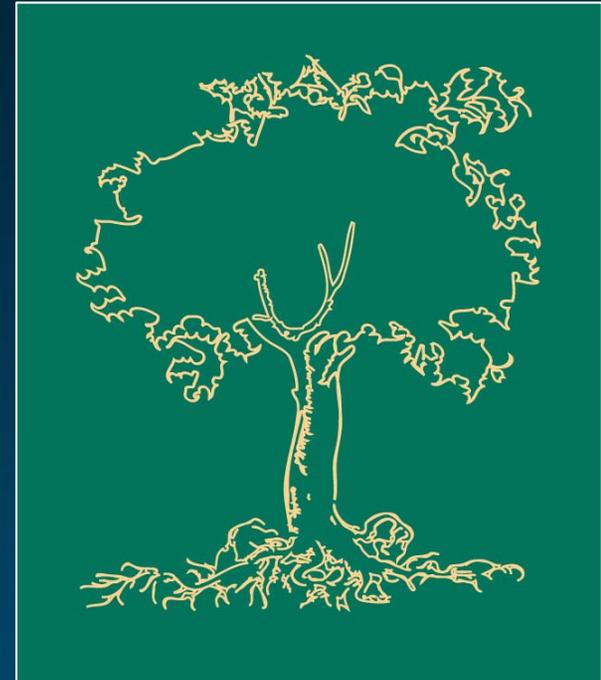
- ◆ Part I: *What is the UMLS?*
  - Introduction
  - Overview through an example
  - The three UMLS Knowledge Sources
    - UMLS Metathesaurus
    - UMLS Semantic Network
    - SPECIALIST Lexicon and lexical tools

Part I  
What is the UMLS?

*(1) Introduction*

# What does UMLS stand for?

- ◆ Unified
- ◆ Medical
- ◆ Language
- ◆ System



UMLS<sup>®</sup>  
Unified Medical Language System<sup>®</sup>  
UMLS Metathesaurus<sup>®</sup>

# Motivation

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- ◆ 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

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## ◆ 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)
- RRF browser (browsing subsets)



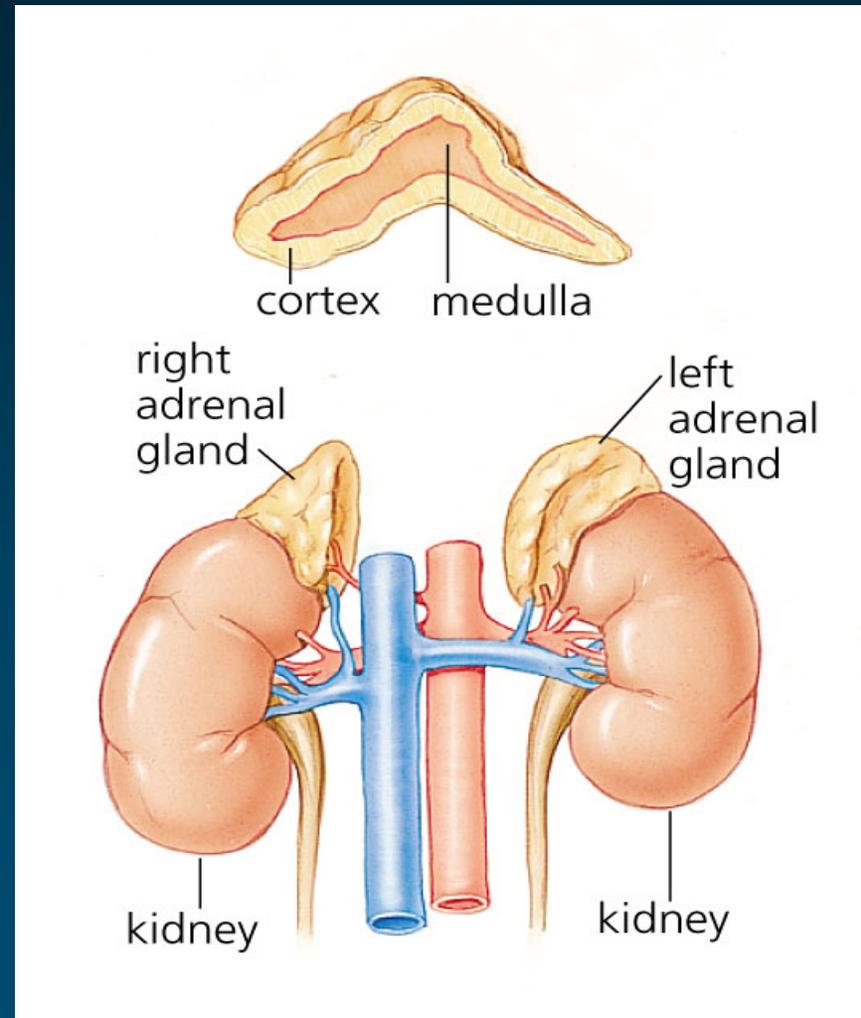
The UMLS is *not* an end-user application

Part I  
What is the UMLS?

*(2) Overview through an example*

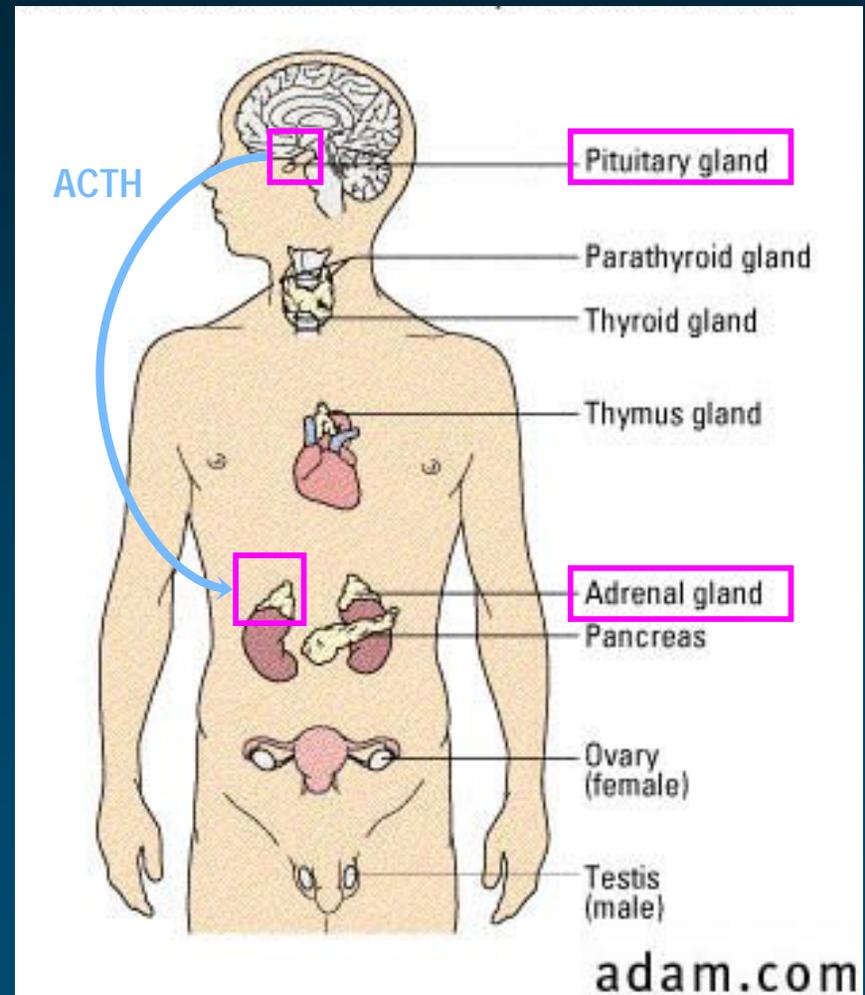
# Addison's disease

- ◆ 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

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- ◆ 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
  - Melasma addisonii
  - Asthenia pigmentosa
  - Primary adrenal deficiency
  - Primary adrenal insufficiency
  - Primary adrenocortical insufficiency
  - Chronic adrenocortical insufficiency
- } eponym
- } symptoms
- } clinical variants

## ◆ Contexts: different hierarchies



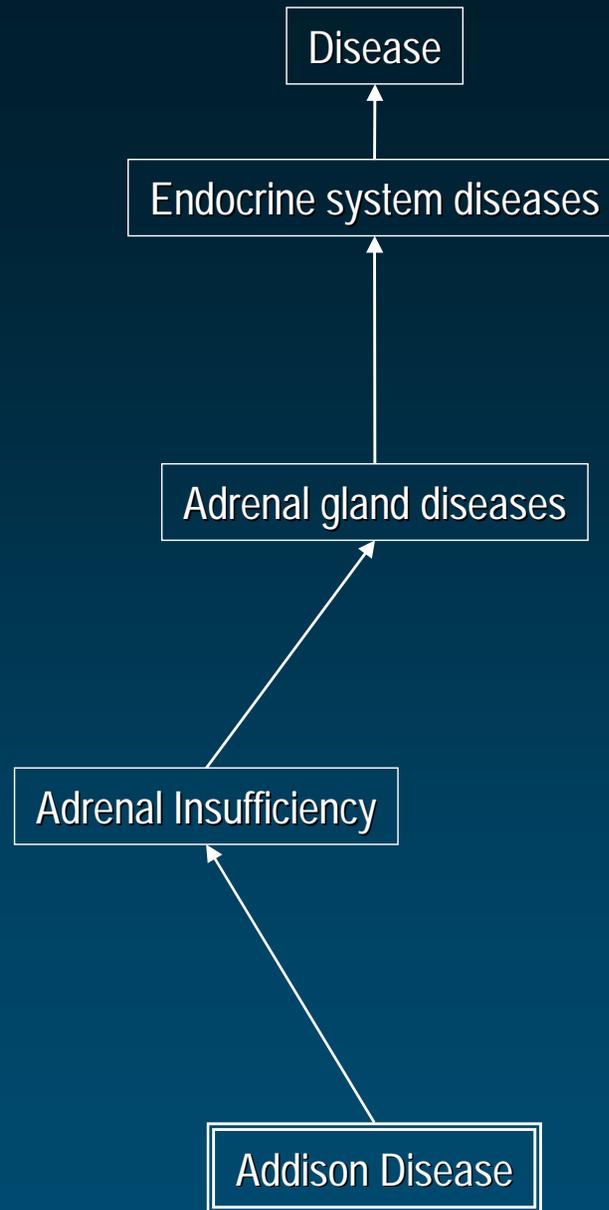
# Organize terms

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

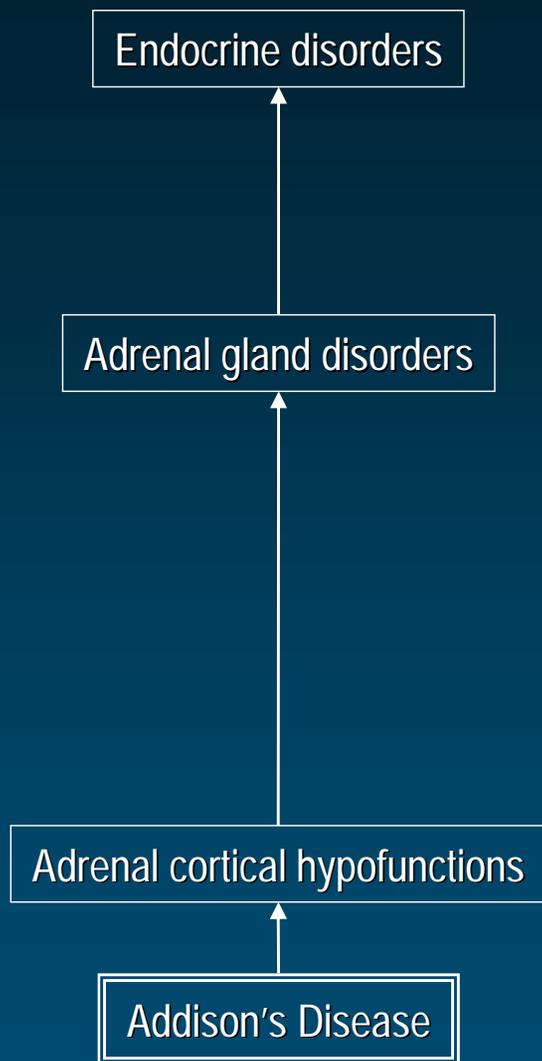
Addison Disease	MeSH	D000224
Primary hypoadrenalism	MedDRA	10036696
Primary adrenocortical insufficiency	ICD-10	E27.1
Addison's disease (disorder)	SNOMED CT	363732003

C0001403

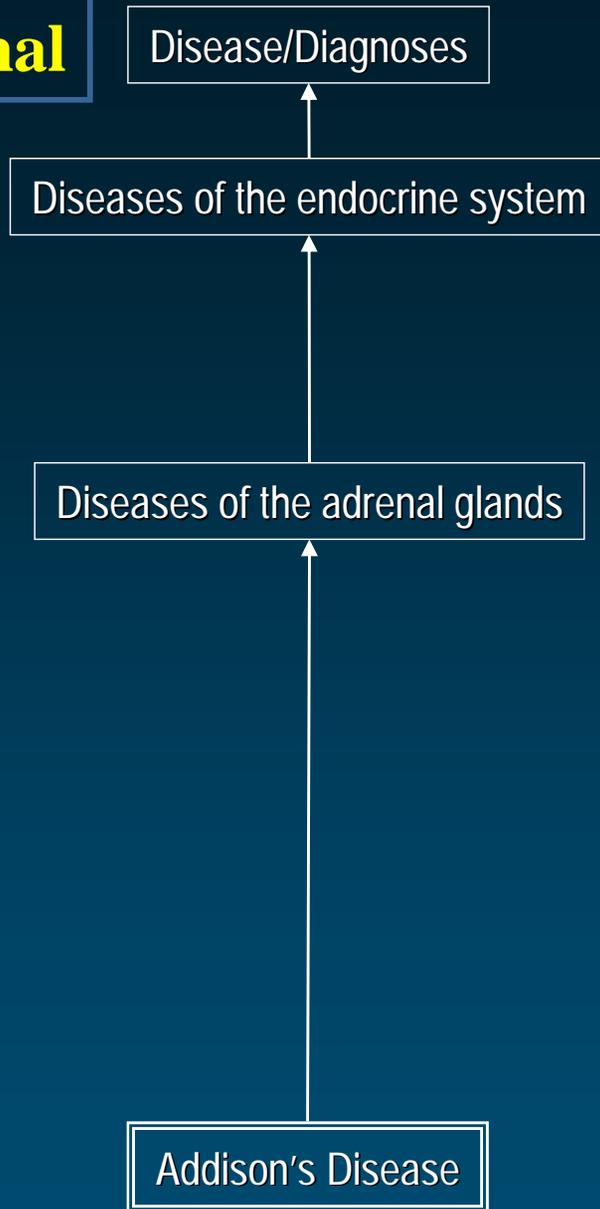
Addison's disease



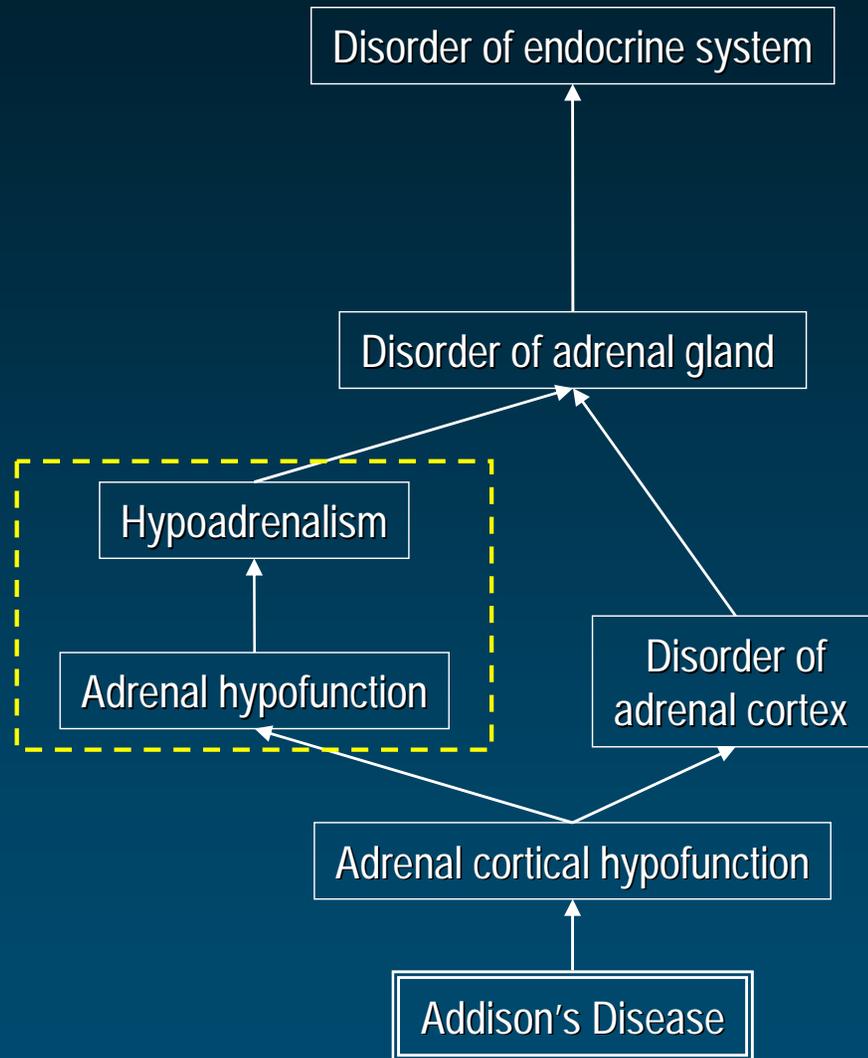
# MedDRA



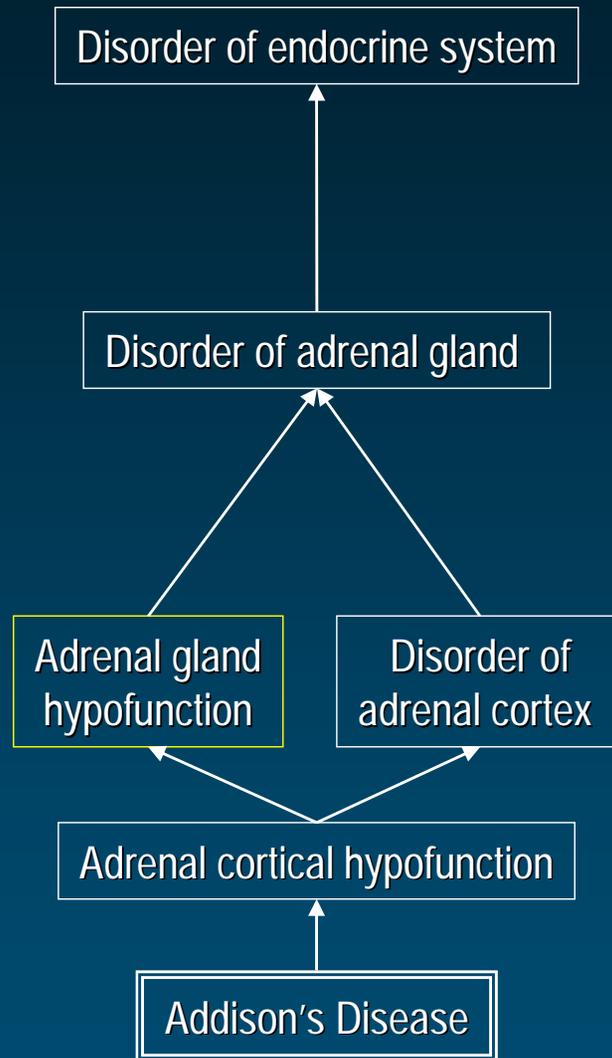
# SNOMED International



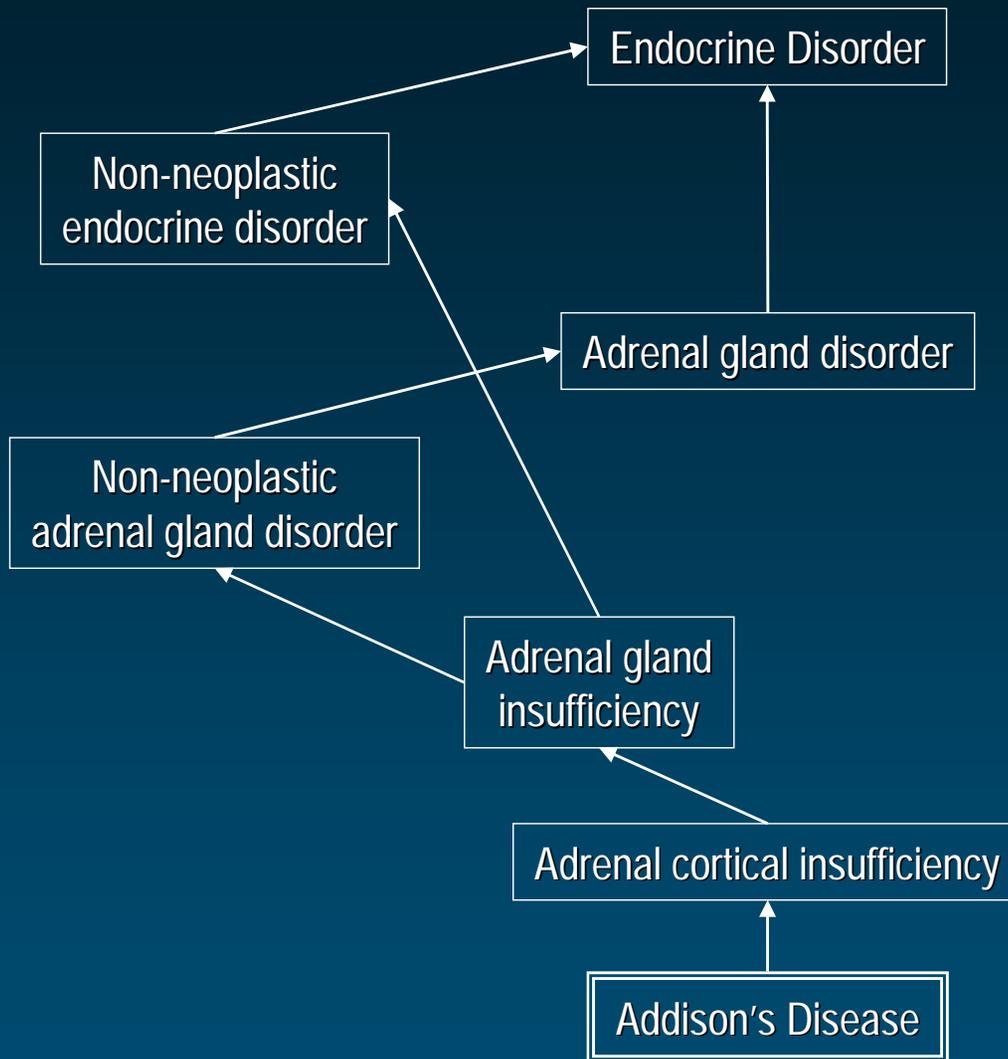
# SNOMED CT (native)



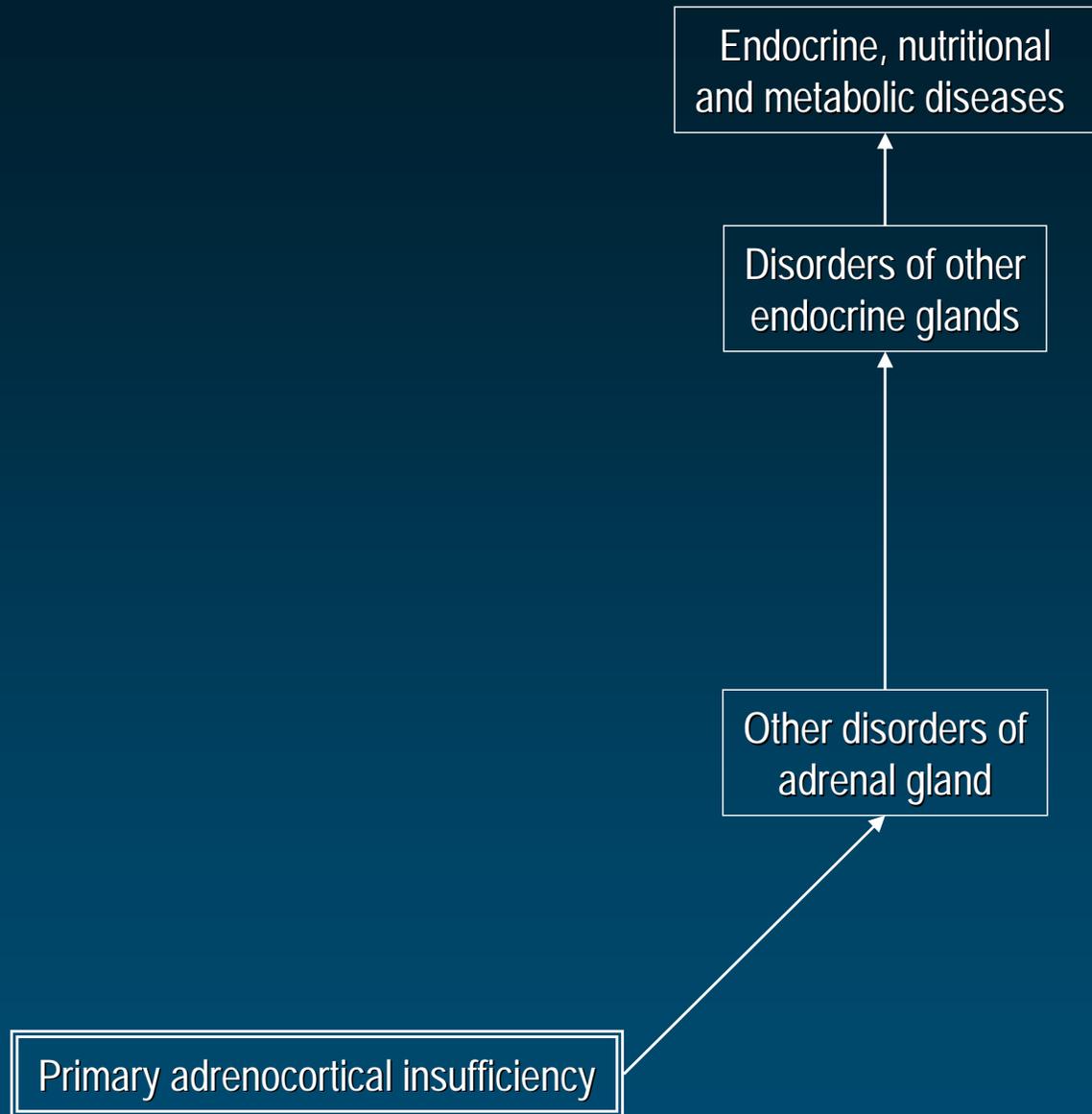
# SNOMED CT (UMLS view)



# NCI Thesaurus

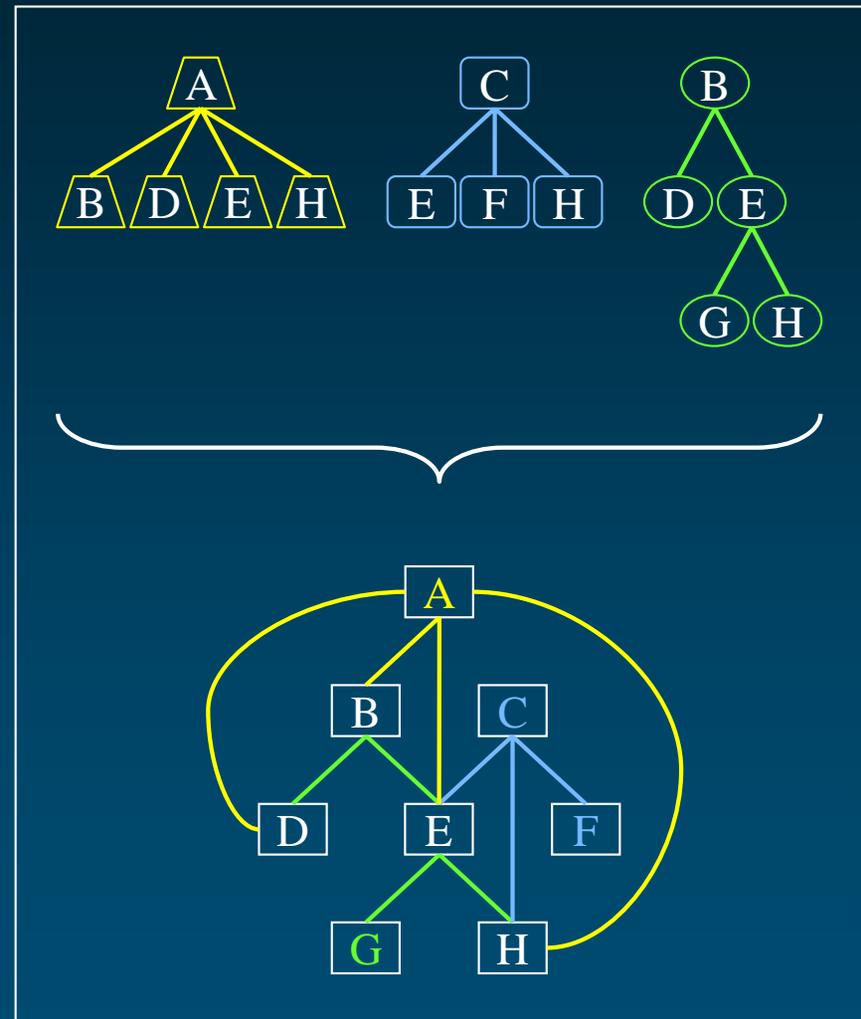


# ICD-10

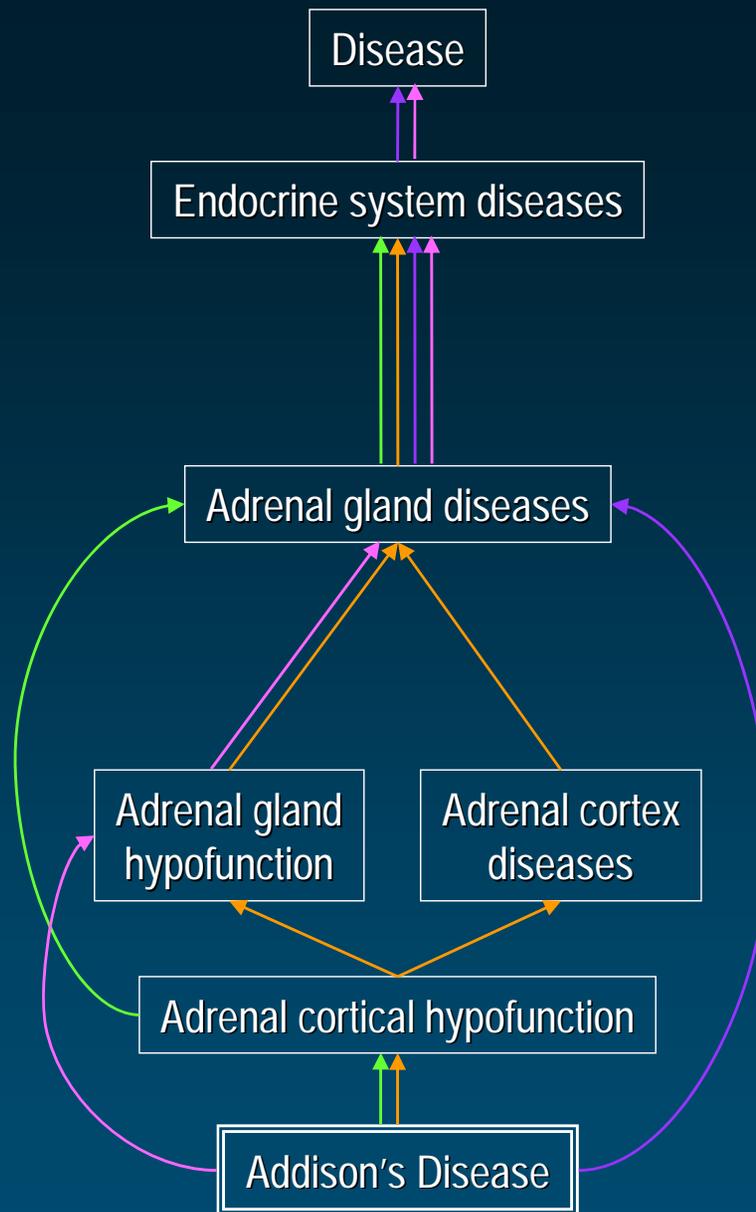


# Organize concepts

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

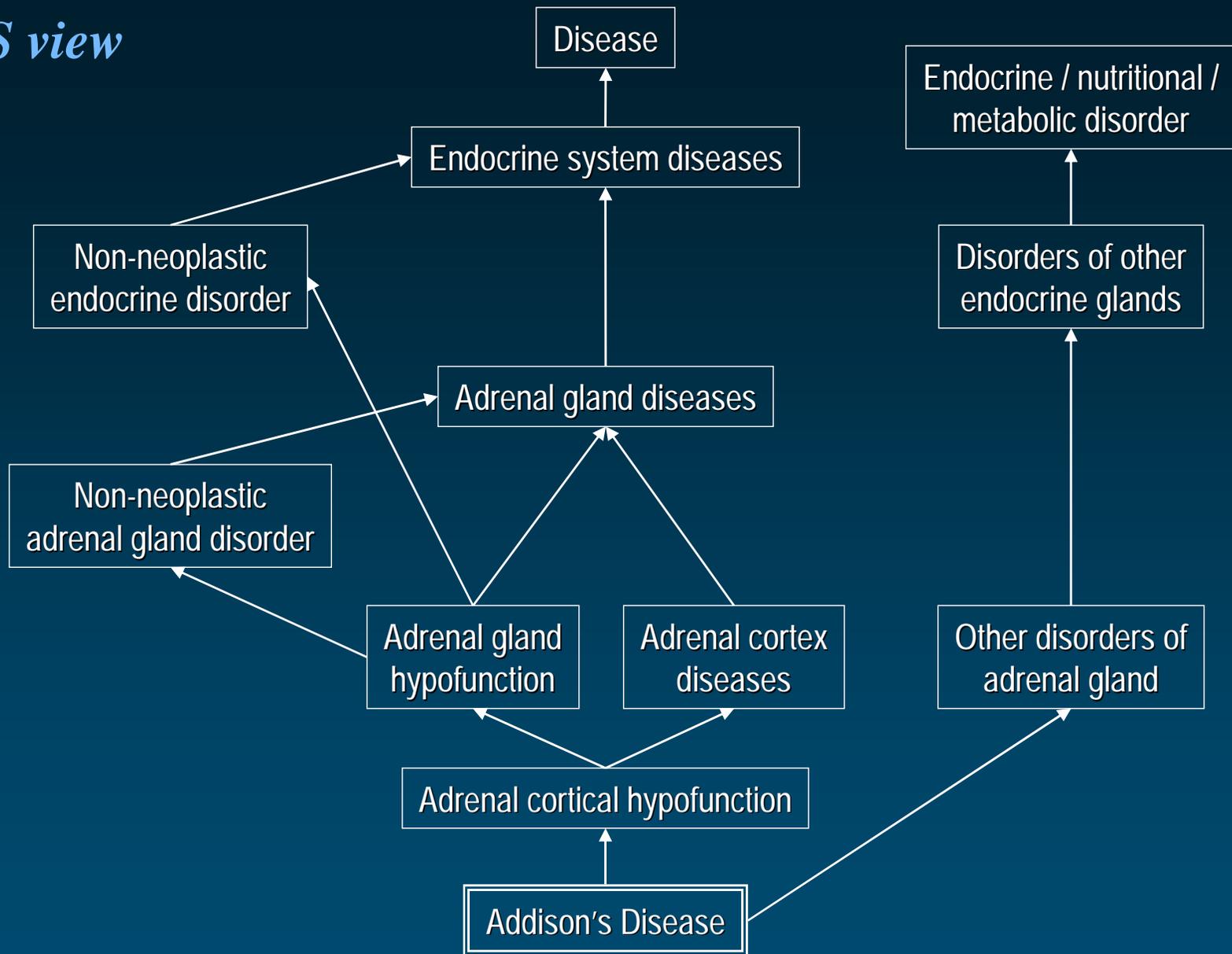


*organize concepts*

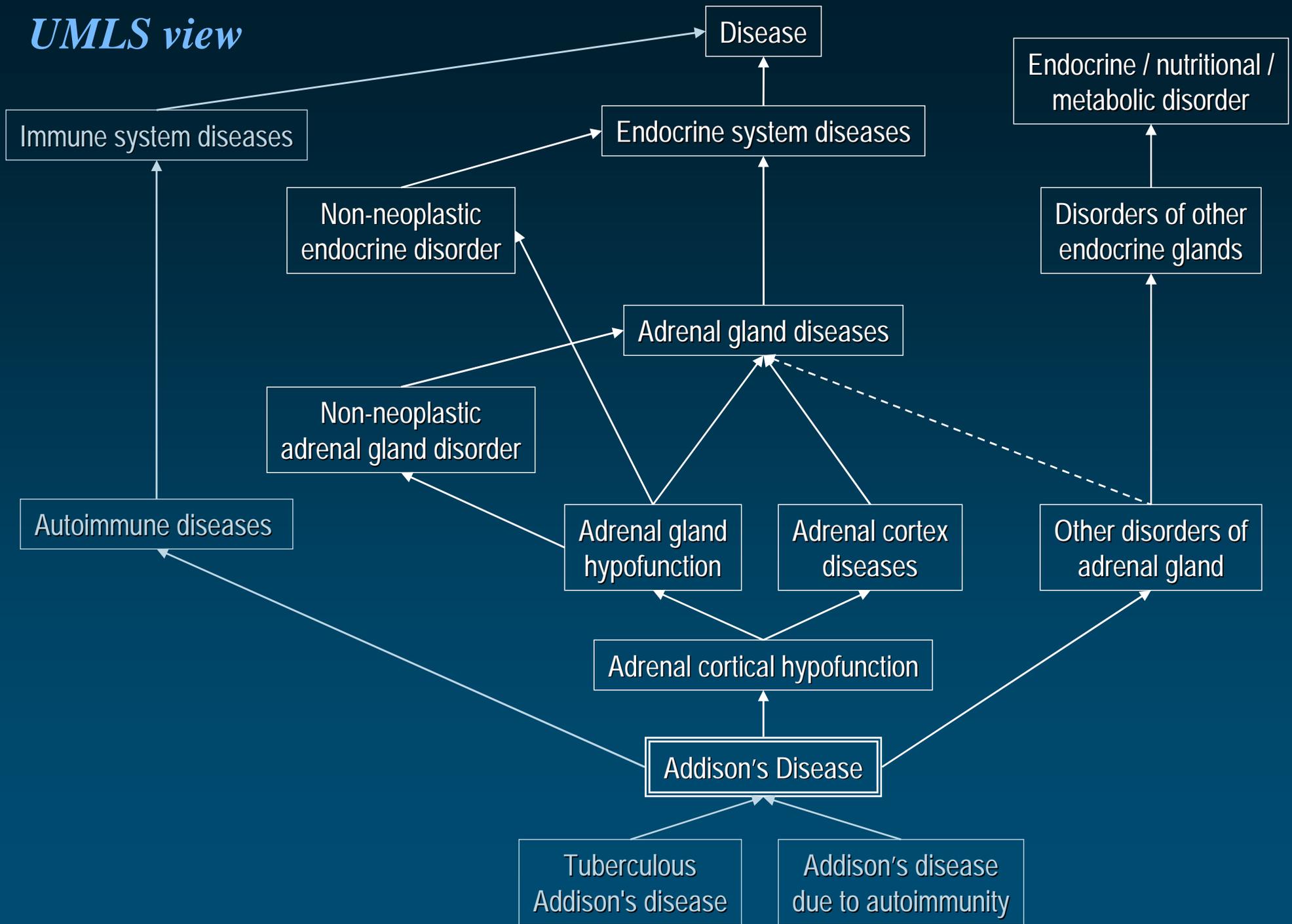


**SNOMED CT**  
**SNOMED Intl**  
**MeSH**  
**MedDRA**

*UMLS view*



*UMLS view*



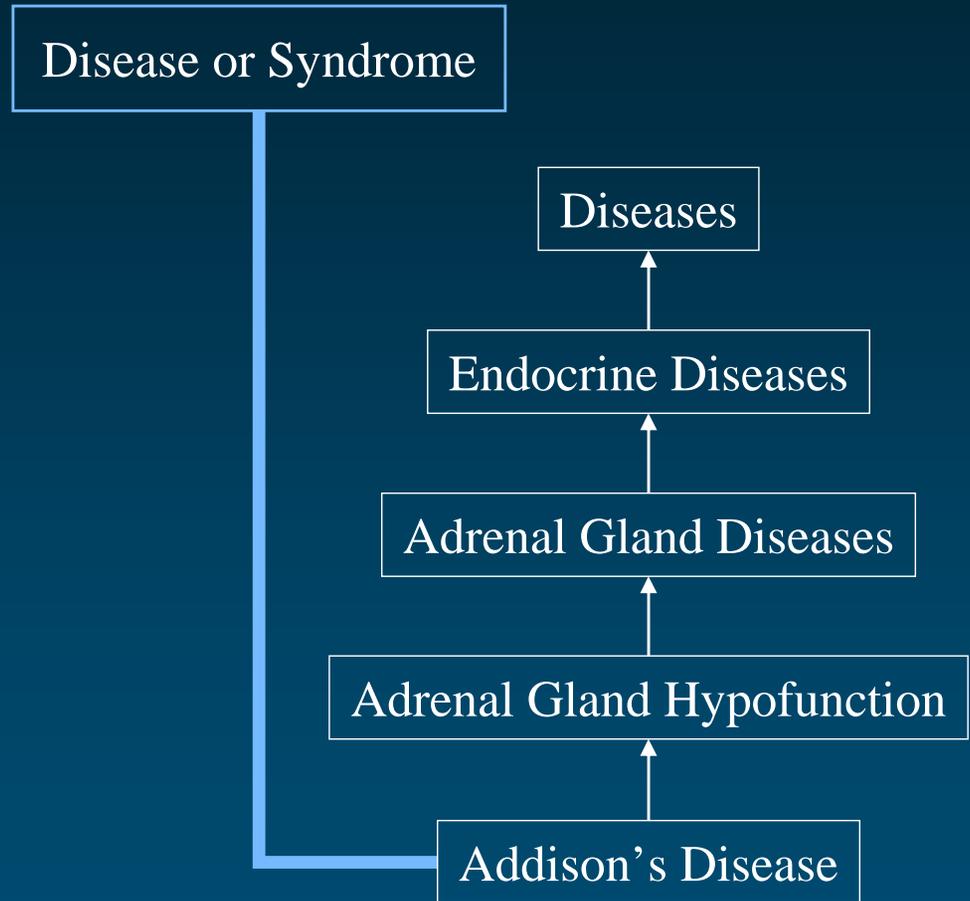
# Relate to other concepts

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

# 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?

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- ◆ Lexical knowledge
- ◆ Semantic pre-processing
- ◆ UMLS editors

# Lexical knowledge

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Adrenal gland diseases

Adrenal disorder

Disorder of adrenal gland

Diseases of the adrenal glands

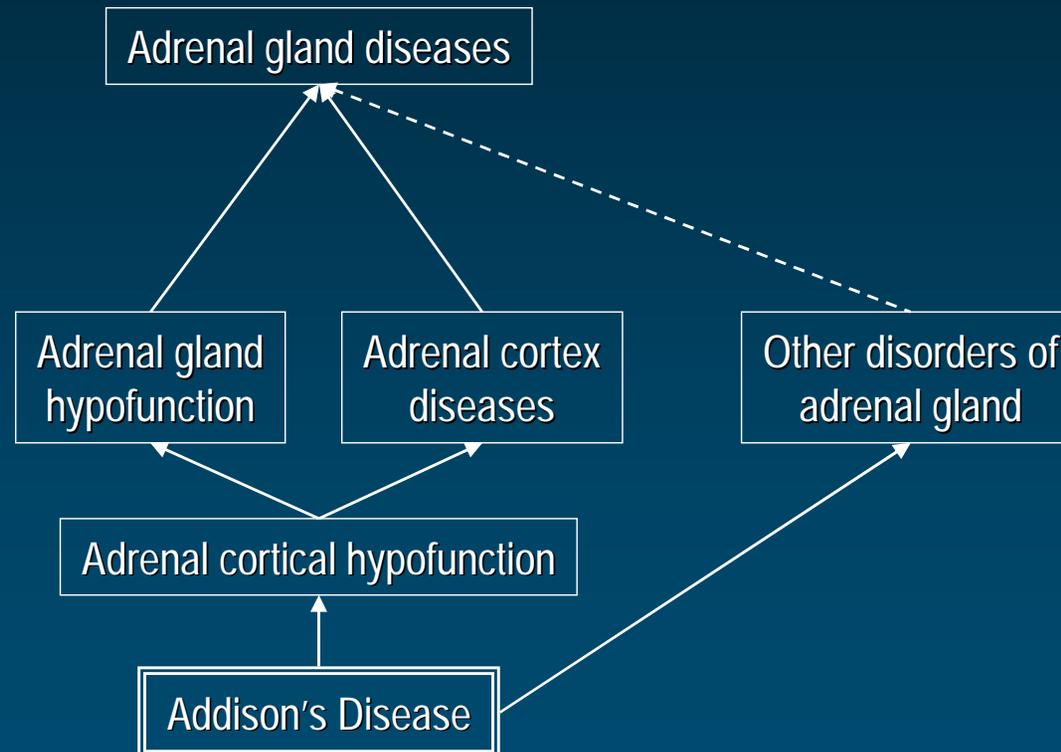
C0001621

# Semantic pre-processing

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- ◆ 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

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- ◆ Synonymous terms clustered into concepts
- ◆ Unique identifier
  
- ◆ Finer granularity
- ◆ Broader scope
- ◆ Additional hierarchical relationships
- ◆ Semantic categorization

Part I  
What is the UMLS?

*(3) UMLS Knowledge Sources*

# 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

# UMLS Metathesaurus

# Metathesaurus Basic organization

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## ◆ 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

(2007AB)

- ◆ 143 source vocabularies
  - 17 languages
- ◆ Broad coverage of biomedicine
  - 5.9M names
  - 1.4M concepts
  - 8M relations
- ◆ Common presentation

# Biomedical terminologies

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## ◆ 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)

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## ◆ Specialized vocabularies

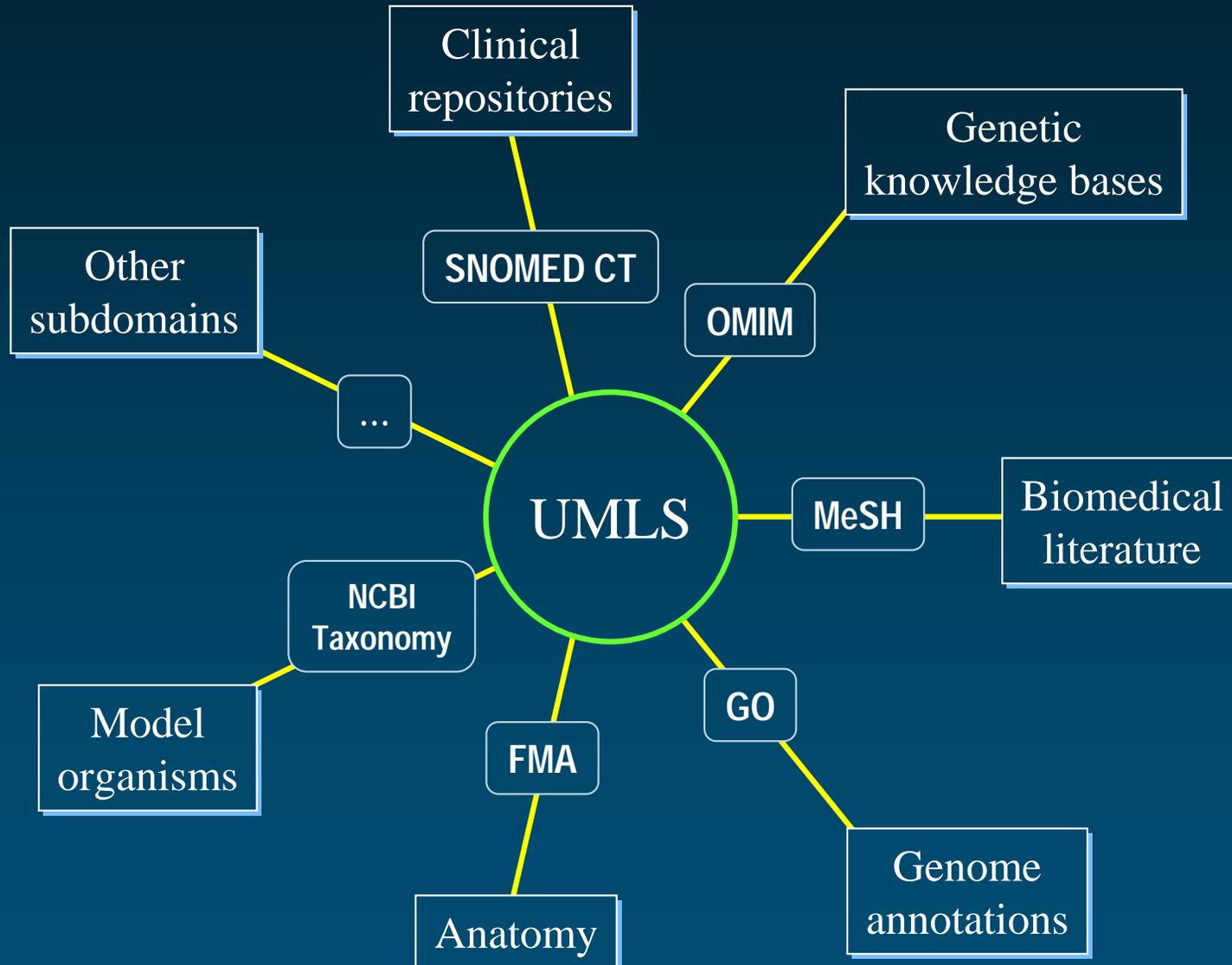
- nursing (NIC, NOC, NANDA, Omaha, PCDS)
- dentistry (CDT)
- oncology (PDQ)
- psychiatry (DSM, APA)
- adverse reactions (COSTART, WHO ART)
- primary care (ICPC)

## ◆ Terminology of knowledge bases (AI/Rheum, DXplain, QMR)

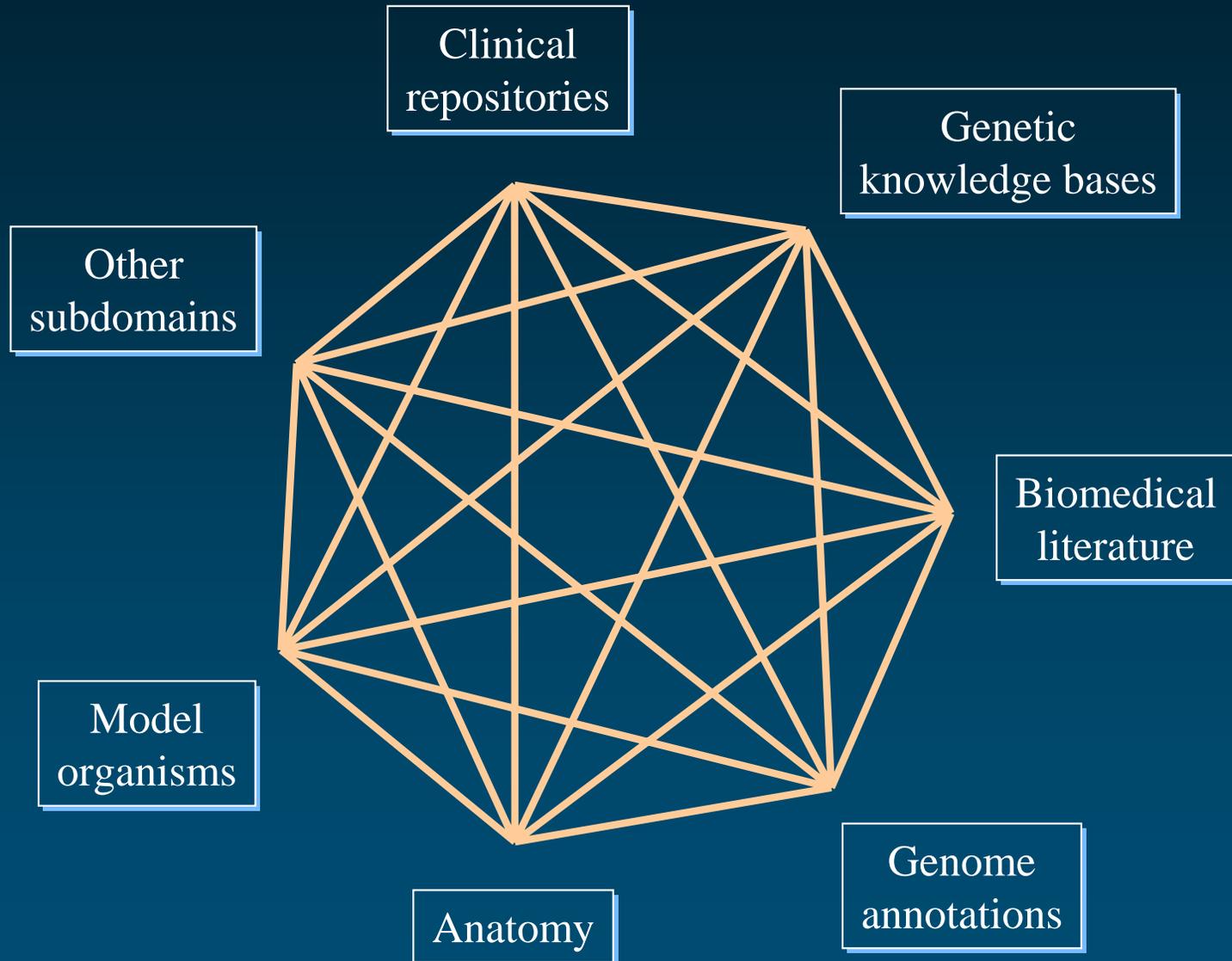
The UMLS serves as a vehicle for the regulatory standards (HIPAA, CHI)



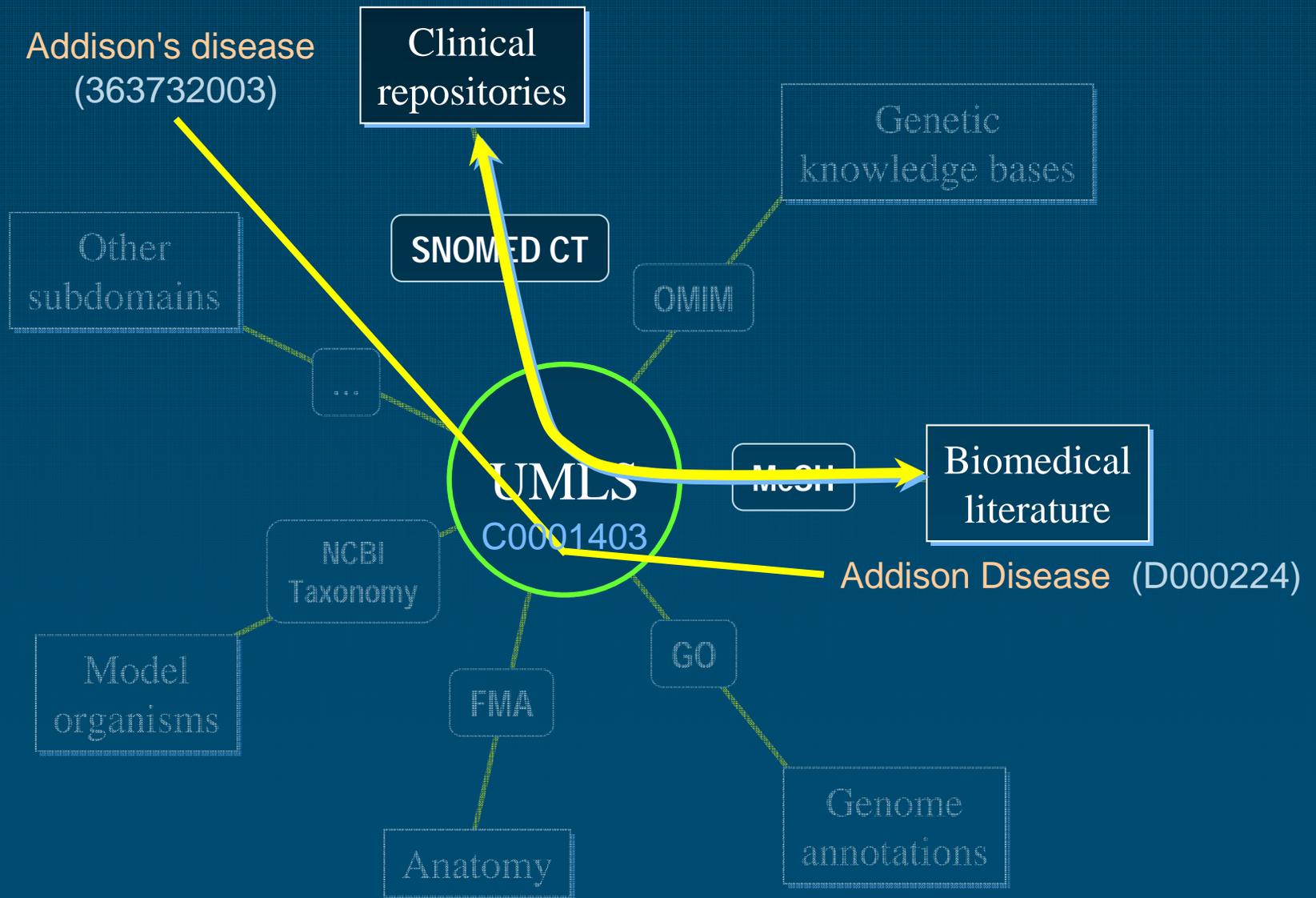
# Integrating subdomains



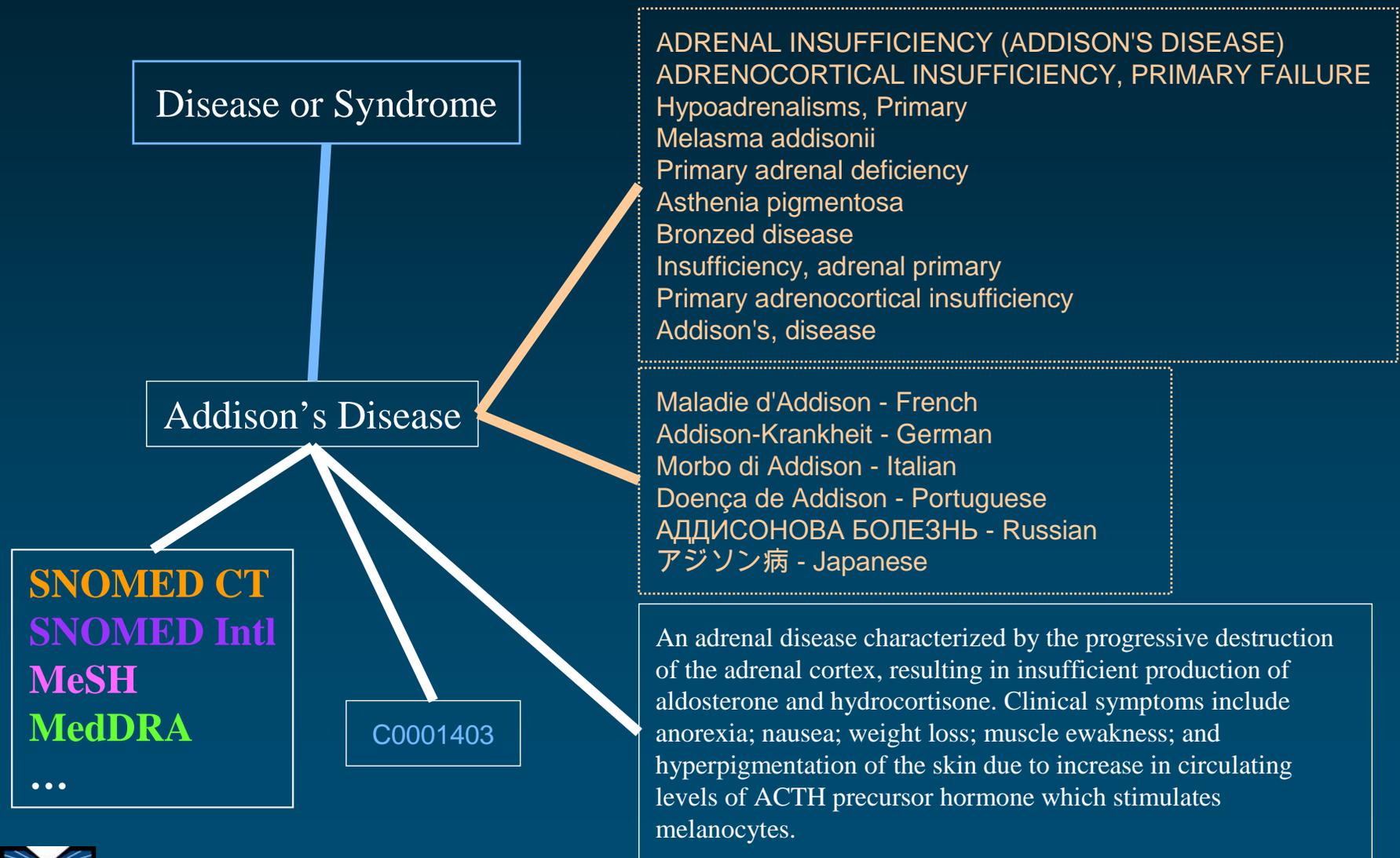
# Integrating subdomains



# Trans-namespace integration



# Addison's Disease: Concept



# Metathesaurus Concepts

(2007AB)

- ◆ Concept (~ 1.4M) CUI
  - Set of synonymous concept names
- ◆ Term (~ 5.3 M) LUI
  - Set of normalized names
- ◆ String (~ 5.9M) SUI
  - Distinct concept name
- ◆ Atom (~ 7.2M) AUI
  - Concept name in a given source

A0066000	Headache	(MeSH)
A0065992	Headache	(ICD-10)
<b>S0046854</b>		

A0066007	Headaches	(MedDRA)
A12003304	Headaches	(OMIM)
<b>S0046855</b>		

**L0018681**

A0540936	Cephalodynia	(MeSH)
<b>S0475647</b>		

**L0380797**

**C0018681**

# Cluster of synonymous terms

Concept  
C0001403

Term L0001403	<p>S0354372 <i>Addison's disease</i></p> <p>S0010794 Addison's Disease</p> <p>S0010792 Addison Disease</p> <p>S0010796 Addisons Disease</p> <p>S0033587 Disease, Addison</p> <p>S0469271 Addison's disease, NOS</p>	[...]
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>S5924573 Adrenal Insufficiency, Primary</p>	[...]
Term L0585243	<p>S5907343 <i>Primary Hypoadrenalism</i></p> <p>S0718109 Primary hypoadrenalism</p>	[...]
Term L3541031	<p>S4115514 <i>primary; hypoadrenocorticism</i></p> <p>S4090095 hypoadrenocorticism; primary</p>	[...]
Term L1229627	S1471573 <i>Addison-Krankheit</i>	GER
Term L5345155	S6107160 <i>Maladie d'Addison</i>	FRE

[...]



# Beyond concepts Descriptor level

- ◆ In some vocabularies, the unit of information is an aggregate of concepts
  - e.g., descriptors in MeSH, for indexing purposes

MeSH Heading	Hydrocortisone	← C0020268	
Tree Number	<a href="#">D04.808.745.745.654.600</a>		
Tree Number	<a href="#">D06.472.040.585.353.476</a>		
Tree Number	<a href="#">D06.472.040.585.478.392</a>		
Scope Note	The main glucocorticoid secreted by the <a href="#">ADRENAL CORTEX</a> . Its synthetic counterpart is used, either as an injection or to reduce inflammation, allergy, collagen diseases, asthma, adrenocortical deficiency, shock, and some neoplastic conditions.		
Entry Term	11-Epicortisol	← C0887247	
Entry Term	Cortifair		← C0699401
Entry Term	Cortisol	← C0020268	
Entry Term	Cortril		← C0699402
Entry Term	Epicortisol	← C0887247	
Entry Term	Hydrocortisone, (11 alpha)-Isomer	← C0887247	
Entry Term	Hydrocortisone, (9 beta,10 alpha,11 alpha)-Isomer		← C0887246

# Beyond concepts Descriptor level

A0070119 Hydrocortisone (MeSH)

A0043102 Cortisol (MeSH)

C0020268

A0066000 Epicortisol (MeSH)

A0016625 11-Epicortisol (MeSH)

A0055118 Hydrocortisone, (11 alpha)-Isomer (MeSH)

C0887247

A7801724 Hydrocortisone, (9 beta,10 alpha,11 alpha)-Isomer (MeSH)

C0887246

A7757592 Cortifair (MeSH)

C0699401

A7757595 Cortril (MeSH)

C0699402

D006854

# 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

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- ◆ 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

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## ◆ 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

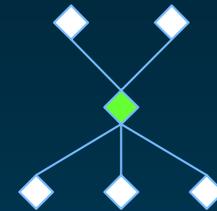
# Symbolic relationships Type

## ◆ 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

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## ◆ Hierarchical

- isa (is-a-kind-of)
- part-of

## ◆ Associative

- location-of
- caused-by
- treats
- ...

## ◆ Cross-references (mapping)

# Mapping relations

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## ◆ 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

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- ◆ Co-occurrence information (MRCOC)
  - Co- occurrence of MeSH descriptors in MEDLINE for the most part
- ◆ 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)
  - ...

Semantic Types

Anatomical Structure

Fully Formed Anatomical Structure

Embryonic Structure

Body Part, Organ or Organ Component

Disease or Syndrome

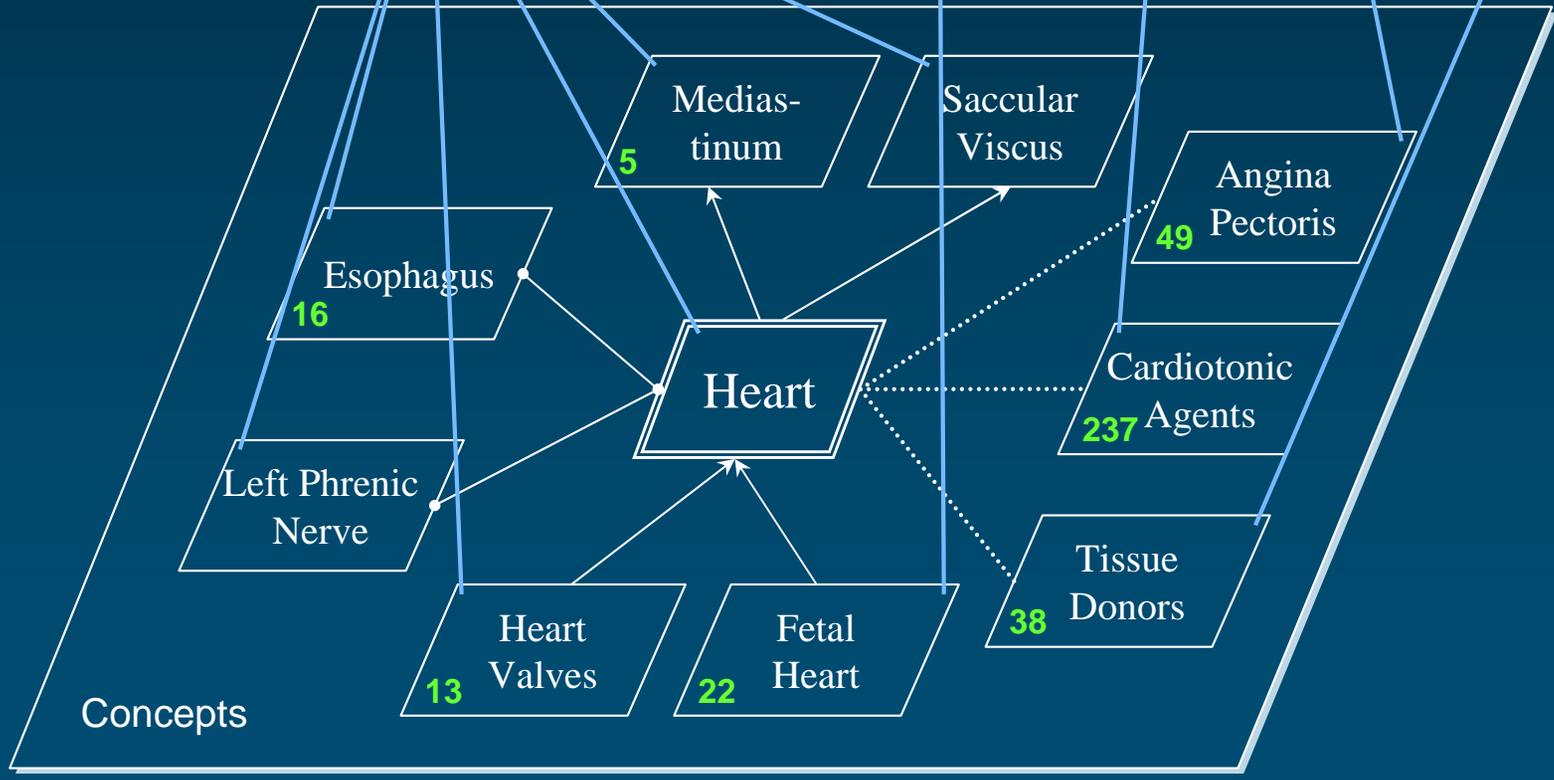
Pharmacologic Substance

Population Group

Semantic Network



Metathesaurus



# UMLS Semantic Network

# Semantic Network

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## ◆ Semantic types (135)

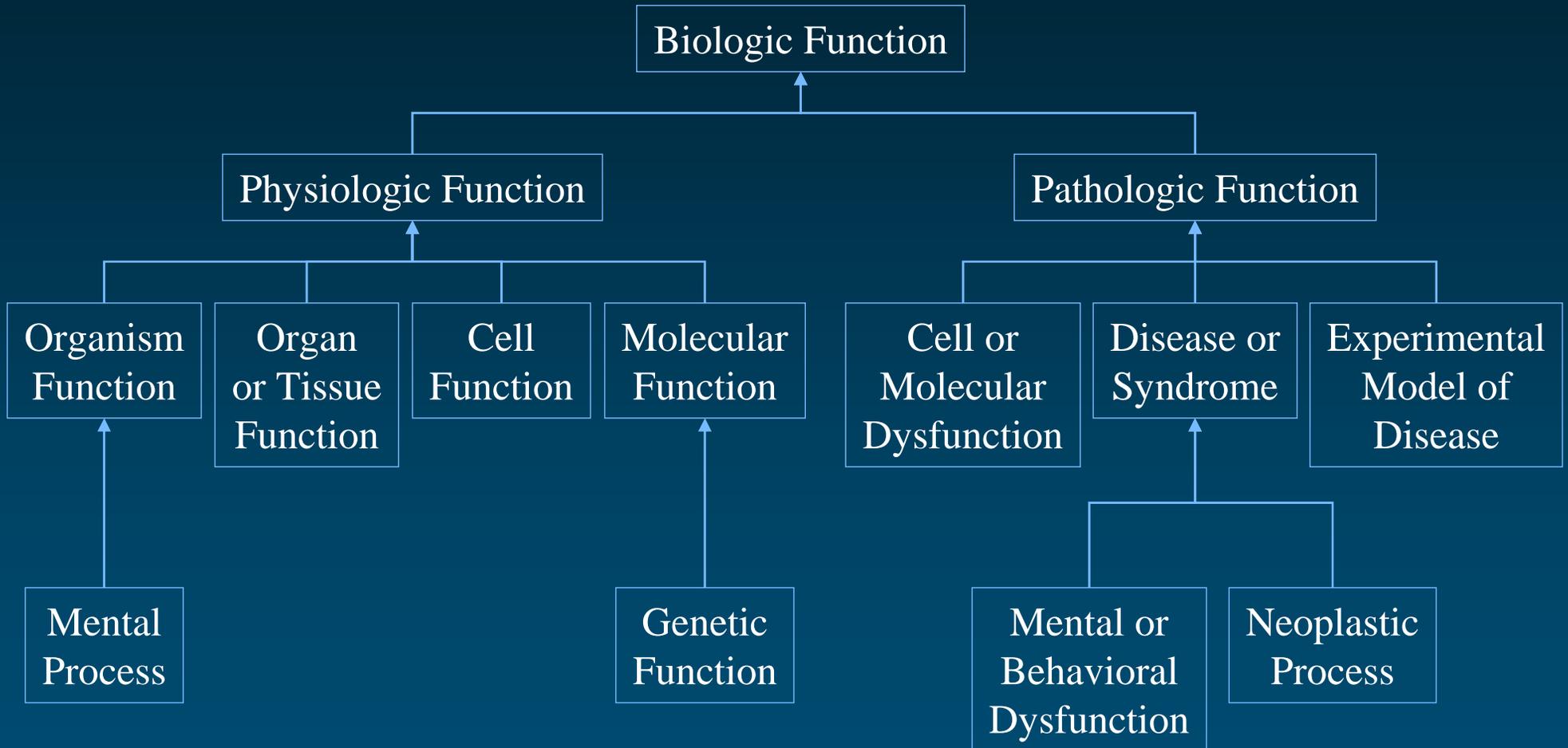
- tree structure
- 2 major hierarchies
  - Entity
    - Physical Object
    - Conceptual Entity
  - Event
    - Activity
    - Phenomenon or Process

# Semantic Network

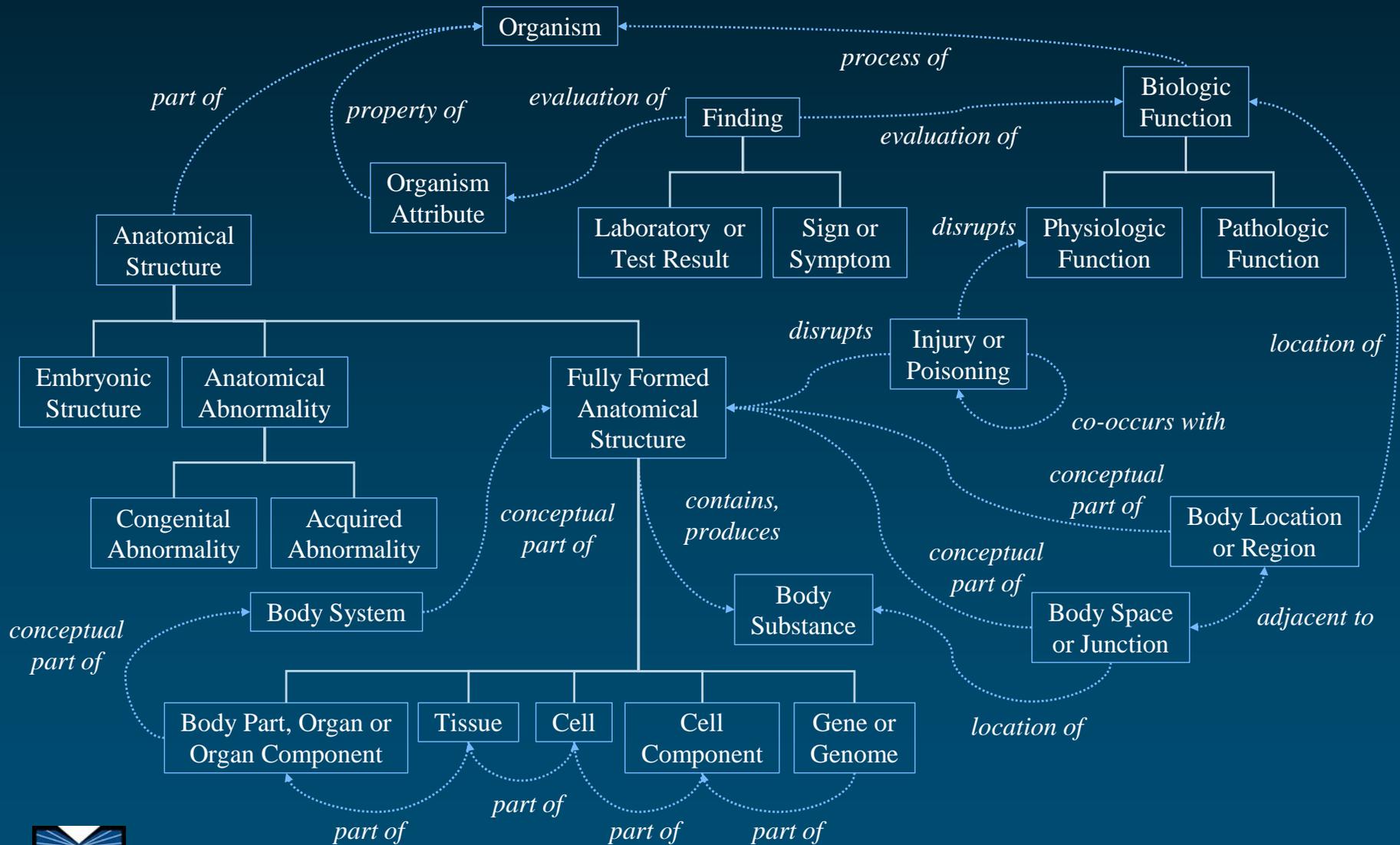
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- ◆ Semantic network relationships (54)
  - hierarchical (isa = is a kind of)
    - among types
      - *Animal isa Organism*
      - *Enzyme isa Biologically Active Substance*
    - among relations
      - *treats isa affects*
  - non-hierarchical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

# “Biologic Function” hierarchy (isa)



# Associative (non-isa) relationships

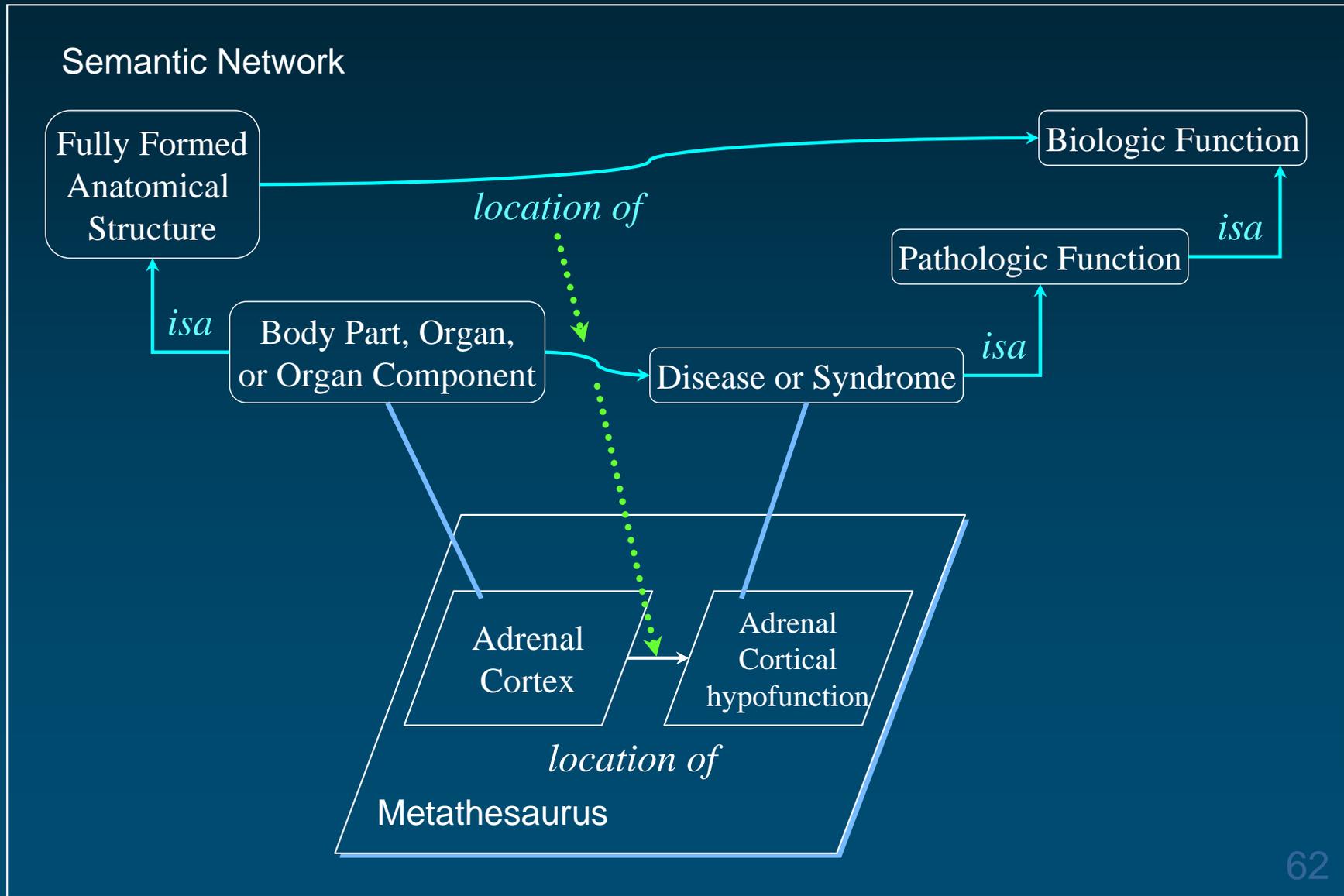


# Why a semantic network?

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- ◆ 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

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- ◆ Content
  - English lexicon
  - Many words from the biomedical domain
- ◆ 360,000 lexical items
- ◆ Word properties
  - morphology
  - orthography
  - syntax
- ◆ Used by the lexical tools

# Morphology

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## ◆ Inflection

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

## ◆ Derivation

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

# Orthography

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## ◆ Spelling variants

- oe/e oesophagus - esophagus
- ae/e anaemia - anemia
- ise/ize cauterise - cauterize
- genitive mark Addison's disease  
Addison disease  
Addisons disease

# Syntax

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## ◆ 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

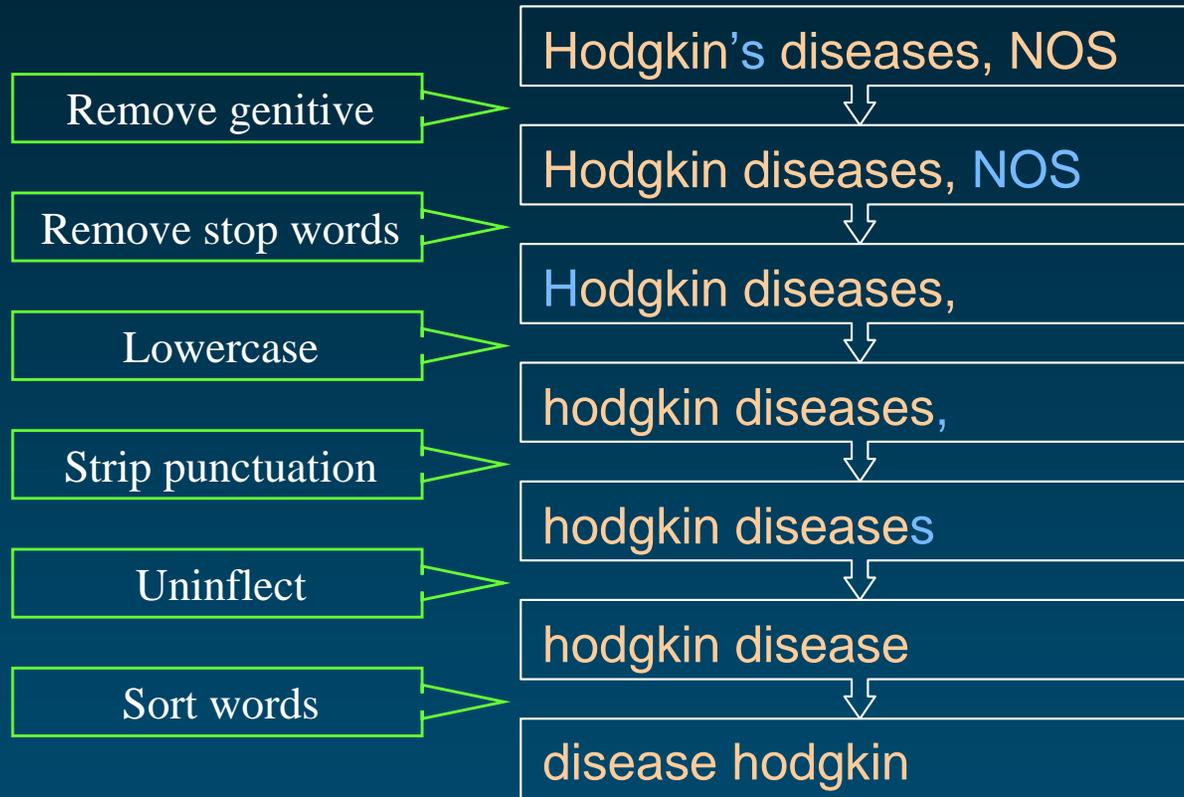
## ◆ Position for adjectives

# Lexical tools

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- ◆ 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

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- ◆ 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

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## ◆ 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

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- ◆ 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)

## Part II

How to use the UMLS?

# Outline

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- ◆ Part II: *How to use the UMLS?*
  - Obtaining a license
  - Remote access
    - Knowledge Source Server (UMLSKS)
    - UMLSKS Application programming interface (API)
  - Local installation and customization (MetamorphoSys)
  - A UMLS-based algorithm: *Restrict to MeSH*
  - Benefits and limitations

## Part II

### How to use the UMLS?

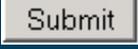
#### *(1) Obtaining a license*

# First step License agreement

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## ◆ Online Web-based license:

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

- Read license 
- Read appendix 1 and 2 
- Print a copy for your records 
- Complete the Web form 
- Verify:
  - receive e-mail from NLM; go to Web site within 72 hours and enter first and last name
- NLM official will countersign (turn-around time of a few days)
- Receive 2<sup>nd</sup> e-mail from NLM with new license number



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



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## Unified Medical Language System

[UMLS Home](#)

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**need to retain UMLS identifiers**

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special category for SNOMED CT

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Accept & continue

Accept and continue

Not accept

## APPENDIX A.1

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Sources are listed in order according to the abbreviations used in the UMLS Metathesaurus files. If additional restrictions and notices apply, the category of restrictions and the special notices appear under the name of the source. See the license agreement for an explanation of the categories of restrictions. Many sources publish printed editions and/or other explanatory information that may be essential to understanding the purpose and application of particular sources in data creation and retrieval. Contact information is provided for each source. Please address questions about permissions or license agreements for additional uses not covered by this Agreement, or other inquiries about individual sources, to the appropriate contacts.

NLM is working toward inclusion in the UMLS Metathesaurus of the complete, current edition of most of these vocabulary sources.

---

**AIR93** - AIR/RHEUM. Bethesda, (MD) : National Library of Medicine, Lister Hill Center, 1993.

Contact: May Cheh, Lister Hill National Center for Biomedical Communications, National Library of Medicine, Building 38A, Room 9E902, 8600 Rockville Pike, Bethesda, MD 20894; e-mail: [cheh@nlm.nih.gov](mailto:cheh@nlm.nih.gov).

---

**ALT2006** - Alternative Billing Concepts (Altlink). 7th Version. Las Cruces, NM, 2006.

[CATEGORY 3 RESTRICTIONS APPLY](#)

source restriction level

Contact: Alternative Link LLC, 6121 Indian School Road NE, Suite 131, Albuquerque, NM 87110; Phone: (505) 875-0001; Toll Free: (877)621-5465; Fax: (505) 875-0002; e-mail: [ail@alternativelink.com](mailto:ail@alternativelink.com)



**WHOFRE\_1997** - WHO Adverse Drug Reaction Terminology (WHOART). French Translation. Uppsala (Sweden): WHO Collaborating Centre for International Drug Monitoring, 1997.

[CATEGORY 2 RESTRICTIONS APPLY](#)

Contact: WHO Collaborating Centre for International Drug Monitoring, Stora Target 3, S-753 20 Uppsala, Sweden; fax: +46-18-656080

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**WHOGER\_1997** - WHO Adverse Drug Reaction Terminology (WHOART). German Translation. Uppsala (Sweden): WHO Collaborating Centre for International Drug Monitoring, 1997.

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**WHOSPA\_1997** - WHO Adverse Drug Reaction Terminology (WHOART). Spanish Translation. Uppsala (Sweden): WHO Collaborating Centre for International Drug Monitoring, 1997.

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## APPENDIX 2

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(2007AB)

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- ◆ Level 3 (25.4%)
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# Part II

## How to use the UMLS?

*(2) Remote access*

# Remote Access

---

- ◆ UMLS Knowledge Source Server:

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

- ◆ Web search interface
- ◆ Application Programming Interface (API)
  
- ◆ Coming soon: web services

Knowledge Source Server  
*Web search interface*

# UMLS<sup>KS</sup> Web search interface

---

- ◆ Logging in
- ◆ Basic searching
- ◆ Advanced searching

# UMLSKS Web search interface log in

**UMLS Knowledge Source Server (UMLSKS)**  
UMLSKS Version: 5.0 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB 2004AC 2005AA 2005AB 2005AC 2006AA 2006AB 2006AC 2006AD 2007AA 2007AB

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▶ [UMLS Metathesaurus®](#)  
▶ [Semantic Network](#)  
▶ [SPECIALIST Lexicon](#)

**Documentation**  
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Password:   
**Login**  
[Forgot your password?](#)

**What's New**  
▶ **The UMLS 2007AB files are now available for download and searching. (July 9, 2007, 8:00 AM)**  
▶ **Transfer of ownership of SNOMED CT® improves U.S. use rights, requires change to UMLS license. <more>**  
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Login ID:   
Password:   
Confirm Password:   
First Name:   
Last Name:   
Email:   
UMLS License #:   
UMLS Licensee First Name:   
UMLS Licensee Last Name:   
**Request Account**

◆ Returning users log in

◆ New users create account

# UMLS Knowledge Source Server Home Page

- ◆ Tabs across top access basic searching of 3 Knowledge Sources
- ◆ Advanced searching options on right-hand side

UMLS Knowledge Source Server (UMLSKS)

UMLSKS Version 5.0 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB 2004AC 2005AA 2005AB 2005AC 2006AA 2006AB 2006AC 2006AD 2007AA 2007AB

Metathesaurus Semantic Network SPECIALIST Lexicon

Logout

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- Mappings
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- Developer's API

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- Developer's Guide
- Developer's API Javadocs
- UMLS Documentation Set
- RxNorm Documentation

**Resources**

- NLP & Lexical Resources
- Semantic Network Resources
- Metathesaurus Resources

**Quick Search**

Select UMLS Release: 2007AB

Enter search value: addison's disease

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**

- The UMLS 2007AB files are now available for download and searching. (July 9, 2007, 8:00 AM)
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# UMLS Knowledge Source Server Home Page



## UMLS Knowledge Source Server (UMLSKS)

UMLSKS Version 5.0      UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB 2004AC 2005AA 2005AB 2005AC 2006AA 2006AB 2006AC 2006AD 2007AA 2007AB

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



Metathesaurus



Semantic N/W



SPECIALIST Lexicon

### Quick Search

Select UMLS Release:

Enter search value:

[Metathesaurus  
Concept  
Search](#)

[Search Tips...](#)

[Semantic  
Network  
Search](#)

[Search Tips...](#)

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#### [Semantic Network Browser](#)

Allows browsing of the hierarchies for the Semantic Network.

# Metathesaurus Basic Search

## *Addison's disease*

**UMLS Knowledge Source Server (UMLSKS)**  
UMLSKS Version 5.0 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB 2004AC 2005AA 2005AB 2005AC 2006AA 2006AB 2006AC 2006AD 2007AA 2007AB

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**Quick Search**  
Select UMLS Release: 2007AB  
Enter search value: addison's disease

**Advanced Searches**  
**Metathesaurus Advanced Search**  
Facilitates advanced searching of the UMLS Metathesaurus, including...

Select UMLS Release: 2007AB  
Enter search value: addison's disease

Metathesaurus Concept Search  
Semantic Network Search  
SPECIALIST Lexicon Search

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

◆ UMLS Release

◆ Search Term

◆ UMLS Knowledge Source

# Concept Report *Addison's disease*

**UMLS Knowledge Source Server (UMLS SKS)**  
UMLS SKS Version 5.0 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB 2004AC 2005AA 2005AB 2005AC 2006AA 2006AB 2006AC 2006AD 2007AA 2007AB

Metathesaurus Semantic Network SPECIALIST Lexicon

Home Advanced Search Logout

Metathesaurus Search for: **Addison's disease** in UMLS Release 2007AB

**Concept**  
 Definition  
 Synonyms  
 Other Languages  
 Suppressible Synonyms  
 Sources

**Context**  
 Ancestors  
 Parents  
 Siblings  
 Children

**Relations**  
 Narrower  
 Broader  
 Similar  
 Other  
 Related and possibly synonymous  
 Source asserted synonymy  
 Allowable Subheadings  
 Associated Expressions

**Co-occurring Concepts**  
 Co-occurring MeSH  
 Co-occurring AI/RHEUM

**Concept: Addison's disease**  
CUI: [C0001403](#)

**Semantic Type:** [Disease or Syndrome](#)

**Definition:**  
disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin; due to tuberculosis or autoimmune induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. ([CRISP Thesaurus](#))

En sjukdom kännetecknad av lågt blodtryck, viktninskning, anorexi, svaghet och ibland av en bronsliknande missfärgning av huden. Den orsakas av tuberkulos eller autoimmuna reaktioner i binjurarna som leder till aldosteron- och kortisonbrist. Utan behandling leder sjukdomen vanligtvis till döden. ([MeSH Swedish](#))

An adrenal disease characterized by the progressive destruction of the ADRENAL CORTEX, resulting in insufficient production of ALDOSTERONE and HYDROCORTISONE. Clinical symptoms include ANOREXIA; NAUSEA; WEIGHT LOSS; MUSCLE WEAKNESS; and HYPERPIGMENTATION of the SKIN due to increase in circulating levels of ACTH precursor hormone which stimulates MELANOCYTES. ([MeSH](#))

**Synonyms:**  
[Addison's disease](#)  
[Addison; disease or syndrome](#)  
[Addison's disease \(disorder\)](#)  
[ADDISON DIS](#)  
[adrenal cortex; deficiency, primary](#)

◆ Concept Name /CUI

◆ Semantic Type(s)

◆ Definition(s)

◆ Synonyms

# Display All

The screenshot shows the UMLSKS interface with a search for 'Addison's disease' in UMLS Release 2007AB. The interface includes a navigation bar with 'Home', 'Advanced Search', and 'Logout'. A search bar contains the text 'Metathesaurus Search for: Addison's disease in UMLS Release 2007AB'. Below the search bar, there are two buttons: 'Display' and 'Display All'. The 'Display All' button is highlighted with a blue box. A blue arrow points from the text '“Display” shows results for selected options' to the 'Display' button, and another blue arrow points from the text '“Display All” shows results for all available options' to the 'Display All' button. The main content area shows the concept 'Addison's disease' with its CUI: C0001403. A list of options is shown with checkboxes: 'Definition' (checked), 'Synonyms' (checked), 'Other Languages' (unchecked), 'Suppressible Synonyms' (unchecked), and 'Sources' (unchecked). The description of Addison's disease is visible, mentioning symptoms like weight loss, anorexia, weakness, and skin changes. A list of synonyms is also provided, including 'Addison's disease', 'Addison, disease or syndrome', 'Addison's disease (disorder)', 'ADDISON DIS', and 'adrenal cortex, deficiency, primary'.

“Display”  
shows results  
for selected  
options

“Display All”  
shows results  
for all available  
options

# Metathesaurus Basic Search

## *Adrenal gland insufficiency*

**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

### Metathesaurus Search

Select UMLS Release:

Enter a term or a concept unique identifier (CUI):

**Perform Concept Search**

Basic searching allows users to search for a concept by entering a term name or a

Select UMLS Release:

Enter a term or a concept unique identifier (CUI):

**Perform Concept Search**

1. The normalized string index is searched for the input string (English only).
2. If not found, then the normalized word index is searched for the input string (English only).
3. If not found, then a spelling check is performed on the input string.

**NOTE:** The basic searching algorithm is used primarily to locate English terms. To search for non-English terms, used the advanced search option **Focused Search** and select the "Word Index" and language

### ◆ Specify:

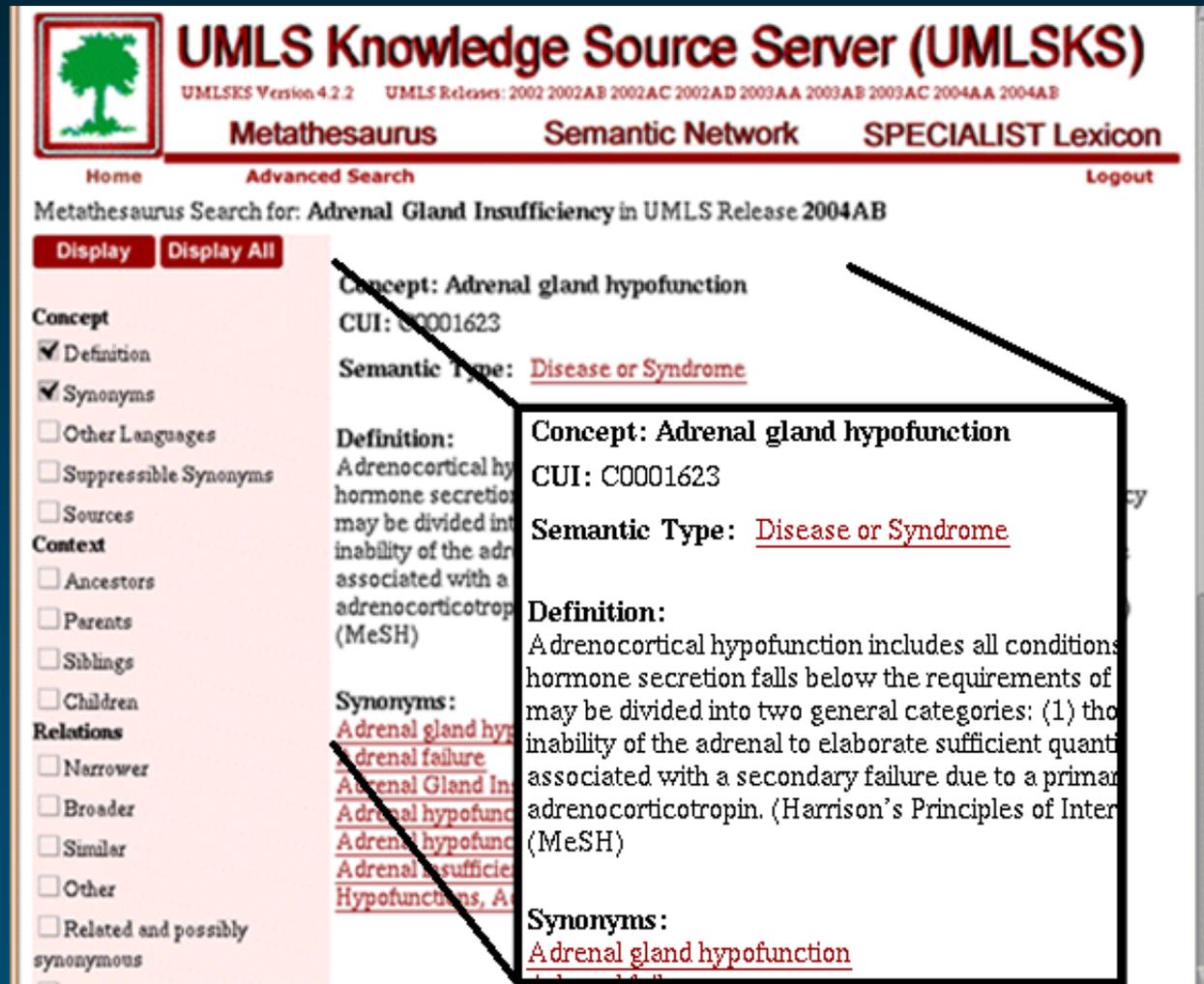
- UMLS Release
- Search term

### ◆ Algorithm:

- Search Normalized String
- Search Normalized Word
- Suggest Spelling

# Basic Concept Report

## *Adrenal gland insufficiency*



The screenshot displays the UMLS Knowledge Source Server (UMLS SKS) interface. The header includes the UMLS logo, the title "UMLS Knowledge Source Server (UMLS SKS)", and version information: "UMLS SKS Version 4.2.2" and "UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB". Navigation links for "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon" are present, along with "Home", "Advanced Search", and "Logout".

The search results show a "Metathesaurus Search for: Adrenal Gland Insufficiency in UMLS Release 2004AB". Two buttons, "Display" and "Display All", are visible. The left sidebar contains filters for "Concept", "Context", and "Relations".

The main content area shows the following details for the concept "Adrenal gland hypofunction" (CUI: C0001623):

- Concept:** Adrenal gland hypofunction
- CUI:** C0001623
- Semantic Type:** [Disease or Syndrome](#)
- Definition:** Adrenocortical hypofunction is a condition in which the secretion of the adrenal cortex may be divided into two general categories: (1) the inability of the adrenal to elaborate sufficient quantities of hormones associated with a secondary failure due to a primary failure of the pituitary gland to secrete adrenocorticotropic hormone (MeSH)
- Synonyms:**
  - [Adrenal gland hypofunction](#)
  - [Adrenal failure](#)
  - [Adrenal Gland Insufficiency](#)
  - [Adrenal hypofunction](#)
  - [Adrenal hypofunction](#)
  - [Adrenal insufficiency](#)
  - [Hypofunctions, Adrenal](#)

A black box highlights the concept information, and a black arrow points from the "Adrenal gland hypofunction" text in the main content area to the "Adrenal gland hypofunction" text in the highlighted box.

# Concept Report Display All *Adrenal Gland Insufficiency*

The screenshot shows the UMLS Knowledge Source Server (UMLSKS) interface. The search results for 'Adrenal gland insufficiency' in UMLS Release 2004AB are displayed. The main content area includes:

- Concept:** Adrenal gland hypofunction
- CUI:** C0001623
- Semantic Type:** [Disease or Syndrome](#)
- Definition:** Adrenocortical hypofunction includes all conditions in which adrenal steroid hormone secretion falls below the requirements of the body. Adrenal insufficiency may be divided into two general categories: (1) those associated with primary inability of the adrenal to elaborate sufficient quantities of hormone and (2) those associated with a secondary failure due to a primary failure in the elaboration of adrenocorticotropin. (Harrison's Principles of Internal Medicine, 13th ed, p1970) (MeSH)
- Synonyms:**
  - [Adrenal gland hypofunction](#)
  - [Adrenal failure](#)
  - [Adrenal Gland Insufficiency](#)
  - [Adrenal hypofunction](#)
  - [Adrenal hypofunction \(disorder\)](#)
  - [Adrenal insufficiency](#)
  - [Hypofunctions, Adrenal Gland](#)
- Other languages:**
  - Czech:** [NADLEDVINY - HYPOFUNKCE](#)
  - Dutch:**
    - [bijnierhypofunctie](#)
    - [Bijnierhypofunctie](#)
    - [bijnierinsufficiëntie](#)

The left sidebar contains navigation options such as 'Display', 'Display All', 'Concept', 'Definition', 'Synonyms', 'Other Languages', 'Context', 'Relations', 'Co-occurring Concepts', and 'Semantic Group'.

- ◆ Concept Name/CUI
- ◆ Semantic Type(s)
- ◆ Definition(s)
- ◆ Synonyms, including foreign languages
- ◆ Relations (broader, narrower, etc.)
- ◆ Co-occurrence data

# Concept Report Display All (continued)

Synonyms

Sources



Japanese  
副腎機能低下  
副腎機能低下症

Portuguese  
Hipofunção das Glândulas Supra-Renais  
HIPOFUNÇÃO SUPRA-RENAL  
Hipofunção supra-renal  
INSUFICIÊNCIA SUPRA-RENAL  
Insuficiência supra-renal  
Insuficiência supra-renal

Russian  
НАДПОЧЕЧНИКОВАЯ НЕДОСТАТОЧНОСТЬ

Spanish  
hipofunción adrenal  
hipofunción adrenal  
Hipofunción de las Glándulas Suprarrenales  
hipofunción suprarrenal  
Insuficiencia adrenal  
Insuficiencia suprarrenal  
INSUFICIENCIA SUPRARRENAL  
Insuficiencia suprarrenal  
SUPRARRENAL HIPOFUNCION

Swedish  
Binjurshypofunktion

Suppressible Synonyms:  
adrenal insufficiency <2>

Sources:  
Beth Israel Problem List  
Clinical Problem Statements  
COSTAR  
CRISP Thesaurus  
COSTART

Japanese  
副腎機能低下  
副腎機能低下症

Portuguese  
Hipofunção das Glândulas Supra-Renais  
HIPOFUNÇÃO SUPRA-RENAL  
Hipofunção supra-renal  
INSUFICIÊNCIA SUPRA-RENAL  
Insuficiência supra-renal  
Insuficiência supra-renal NE

Russian  
НАДПОЧЕЧНИКОВАЯ ГИПОФУНКЦИЯ

Spanish  
hipofunción adrenal  
hipofunción adrenal

# Concept Report Display All (continued)

Hierarchies

The screenshot displays a concept report with the following sections:

- Ancestors:**
  - MeSH**
    - [MeSH Descriptors](#) []
    - [Index Medicus Descriptor](#) []
    - [Diseases \(MeSH Category\)](#) [C]
    - [Endocrine Diseases](#) [C19]
    - [Adrenal Gland Diseases](#) [C19.053]
    - [Adrenal Gland Hypofunction](#) [C19.053.264]
  - MedDRA**
    - [Endocrine dis](#)
    - [Adrenal gla](#)
    - [Adrenal co](#)
    - [Adrenal in](#)
  - MedDRA**
    - [Metabolism a](#)
    - [Metabolism](#)
    - [Metabolic e](#)
    - [Adrenal](#)
  - NCI Thesaur**
    - [Diseases, Disorders and Findings](#) []
    - [Diseases and Disorders](#) []
    - [Disorder by Site](#) []
    - [Endocrine Disorder](#) []
    - [Adrenal Gland Disorder](#) []
    - [Non-Neoplastic Adrenal Gland Disorder](#) []
    - [Adrenal Gland Insufficiency](#) []
  - NCI Thesaurus**
    - [Diseases, Disorders and Findings](#) []
    - [Diseases and Disorders](#) []
    - [Disorder by Site](#) []

A callout box highlights the MeSH hierarchy:

- MeSH**
  - [MeSH Descriptors](#) []
  - [Index Medicus Descriptor](#) []
  - [Diseases \(MeSH Category\)](#) [C]
  - [Endocrine Diseases](#) [C19]
  - [Adrenal Gland Diseases](#) [C19.053]
  - [Adrenal Gland Hypofunction](#) [C19.053.264]



# Concept Report Display All (continued)

## Relations

The screenshot shows a web interface for a concept report. At the top, a list of concepts is displayed: THYROIDITIS [0420], THYROXINE DECREASED [1349], TSH DECREASED [1690], TSH INCREASED [1831], and VIRILISM [0421]. Below this, a section titled "Narrower Concepts:" lists several related terms with their source and relationship type. A yellow bracket on the left side of the slide highlights the "Relations" section, which is a zoomed-in view of the "Narrower Concepts:" section. The zoomed-in view shows the following text:

**Narrower Concepts:**

- Addison's disease (National Drug File – Reference Terminology) [Relation: isa]
- Addison's disease (Metathesaurus Names) [Relation: ]
- Hypoadosteronism (National Drug File – Reference Terminology) [Relation: isa]

Below the zoomed-in view, the original list of narrower concepts continues:

- Other adrenal hypofunction NOS (Metathesaurus Names) [Relation: ]
- Adrenoleukodystrophy (National Drug File – Reference Terminology) [Relation: inverse isa]
- Adrenal Gland Diseases (Metathesaurus Names) [Relation: ]
- Endocrine Diseases (Metathesaurus Names) [Relation: ]
- Endocrine Diseases (WHO Adverse Reaction Terms) [Relation: ]
- Adrenal cortical hypofunction (SNOMED Clinical Terms)

# Concept Report Display All (continued)

psychology  
radiography  
radionuclide imaging  
radiotherapy  
rehabilitation  
surgery  
therapy  
urine  
veterinary  
ultrasonography  
virology

Associated Expressions: None found.

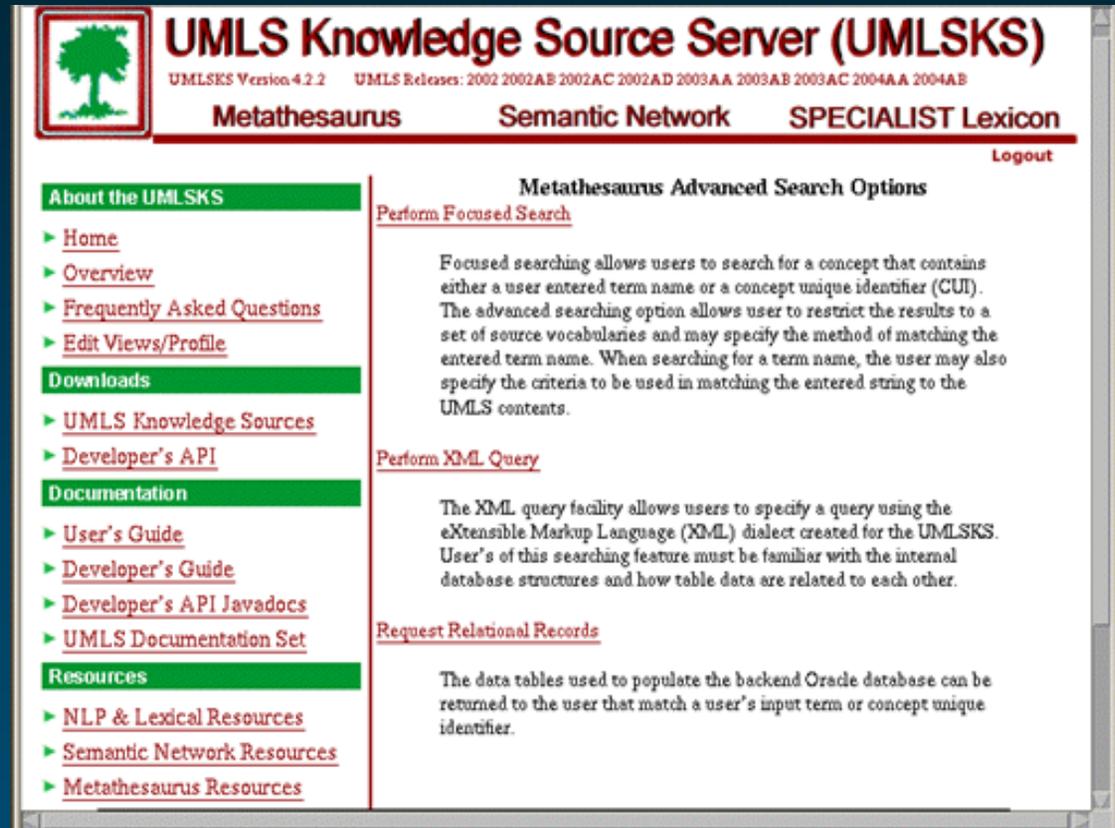
Co-occurring MeSH Terms:

[64] <u>Corticotropin</u>	[64] <u>Corticotropin</u>
[57] <u>Hydrocortisone</u>	[57] <u>Hydrocortisone</u>
[38] <u>Glucocorticoids</u>	[38] <u>Glucocorticoids</u>
[31] <u>Esophageal achalasia</u>	[31] <u>Esophageal achalasia</u>
[30] <u>Dog Diseases</u>	[30] <u>Dog Diseases</u>
[28] <u>Adrenal Glands</u>	[28] <u>Adrenal Glands</u>
[27] <u>Anti-Inflammatory Agents</u>	
[26] <u>DNA-Binding Proteins</u>	
[25] <u>Repressor Proteins</u>	
[25] <u>TRANSCRIPTION FACTORS</u>	
[24] <u>Retinoic Acid Receptors</u>	
[23] <u>Pituitary-Adrenal Axis</u>	
[21] <u>Lacrimal Apparatus</u>	
[20] <u>Hypothalamic-Hypophysial Axis</u>	
[19] <u>Postoperative Complications</u>	
[18] <u>Adrenal Gland Neoplasms</u>	

Co-occurrence  
data

# Metathesaurus Advanced Search Options

- ◆ Focused Search
- ◆ Raw Relational Records



The screenshot displays the UMLS Knowledge Source Server (UMLSKS) interface. At the top, there is a logo of a tree and the title "UMLS Knowledge Source Server (UMLSKS)". Below the title, it shows the version "UMLSKS Version 4.2.2" and a list of releases: "UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB". The main navigation bar includes "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon", with a "Logout" link on the right.

The left sidebar contains a menu with the following sections:

- 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

The main content area is titled "Metathesaurus Advanced Search Options" and contains three sections:

- Perform Focused Search**: A paragraph explaining that focused searching allows users to search for a concept containing a user-entered term name or a concept unique identifier (CUI). It notes that the advanced option allows restricting results to a set of source vocabularies and specifying matching criteria.
- Perform XML Query**: A paragraph stating that the XML query facility allows users to specify a query using the eXtensible Markup Language (XML) dialect created for the UMLSKS. It requires familiarity with internal database structures.
- Request Relational Records**: A paragraph explaining that data tables from the backend Oracle database can be returned to the user matching their input term or CUI.

# Metathesaurus Advanced Search Feature

## Focused Search

The screenshot shows the UMLS Knowledge Source Server (UMLSKS) interface. The page title is "UMLS Knowledge Source Server (UMLSKS)" with a version of 5.0. The navigation menu includes "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". The main content area is titled "Metathesaurus Focused Search:" and contains the following steps:

- 1) Select UMLS Release: 2007AB
- 2) Enter a term or a concept unique identifier (CUI): [Text input field]
- 3) Restrict source vocabulary to:
  - All Source Vocabularies
  - Restrict to selected sources:
- 4) String Matching: Normalized string index [Dropdown menu]
- 5) Language: All [Dropdown menu]

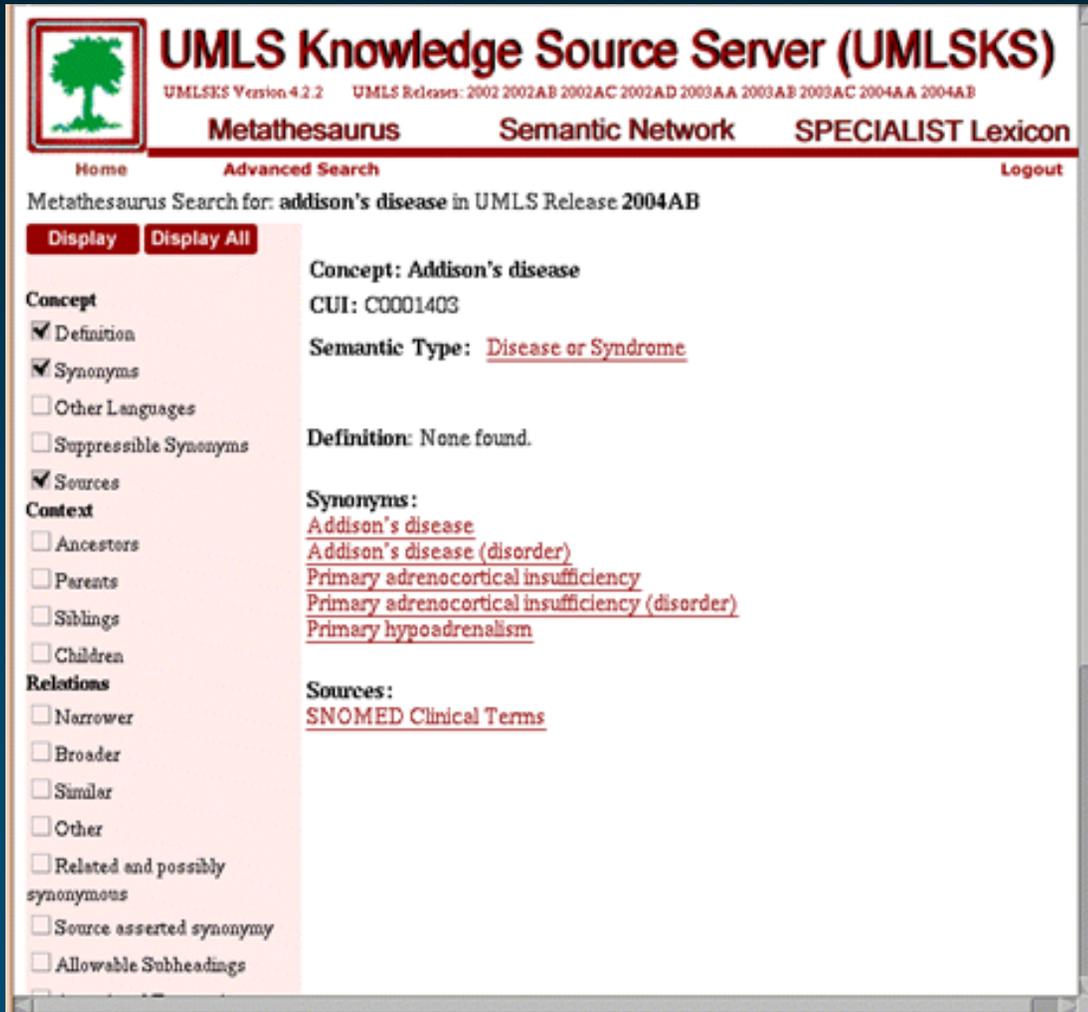
There are two buttons: "Perform Concept Search" and "Perform Term Search". Below the search form, there is a paragraph explaining focused searching and a note about non-English search language.

Done

- ◆ UMLS Release
- ◆ Search Term
- ◆ Source Vocabularies
- ◆ String Criteria
  - Exact Match
  - Normalized string & word
  - Word
  - Truncation (left/right)
  - Approximate Match
- ◆ Language

# Restricted Source Concept Report

## *Addison's Disease*



The screenshot displays the UMLS Knowledge Source Server (UMLSKS) interface. At the top, it shows the UMLSKS logo and version information (4.2.2). Below this, there are navigation links for 'Metathesaurus', 'Semantic Network', and 'SPECIALIST Lexicon'. The search results for 'addison's disease' in UMLS Release 2004AB are shown. The report includes the following information:

- Concept:** Addison's disease
- CUI:** C0001403
- Semantic Type:** [Disease or Syndrome](#)
- Definition:** None found.
- Synonyms:**
  - [Addison's disease](#)
  - [Addison's disease \(disorder\)](#)
  - [Primary adrenocortical insufficiency](#)
  - [Primary adrenocortical insufficiency \(disorder\)](#)
  - [Primary hypoadrenalism](#)
- Sources:**
  - [SNOMED Clinical Terms](#)

The left sidebar contains various filters and options, including 'Display', 'Display All', 'Concept', 'Context', and 'Relations'.

- ◆ UMLS Release:  
2004AB
- ◆ Search Term:  
addison's disease
- ◆ Source Vocabulary:  
SNOMED CT
- ◆ String Criteria:  
Normalized string
- ◆ Language:  
English

# Addison's disease in SNOMED CT

## Preferred Term and Code



**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](#)

[Home](#)   [Advanced Search](#)   [Logout](#)

Metathesaurus Search for: **Addison's disease** in UMLS Release **2004AB**

**Display**

Term Name: Addison's disease  
CUI: [C0001403](#)  
Term UI: L0001403

**Terms**

Term Variants  
 MeSH Entry Terms  
 MeSH Attributes

Source: SNOMED Clinical Terms  
TTY: PT  
ID: 363732003

Users are responsible for compliance with [UMLS copyright restrictions](#)

- ◆ TTY: Term Type
- ◆ ID: Source Code Descriptor

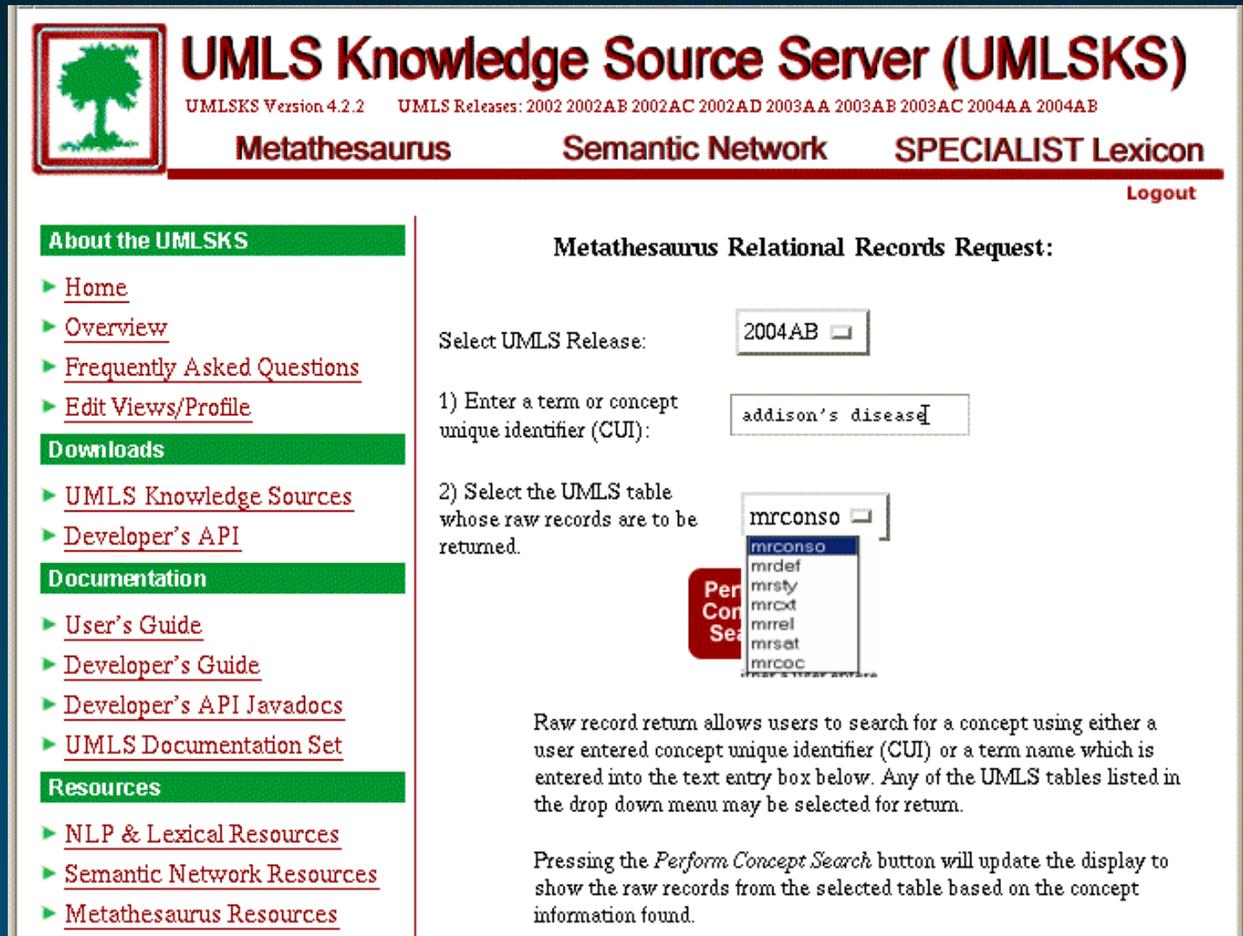
# Metathesaurus Advanced Search Feature

## Relational Record Request

◆ UMLS  
Release

◆ Search Term

◆ UMLS  
Relational  
Table



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

**Metathesaurus Semantic Network SPECIALIST Lexicon** [Logout](#)

**About the UMLS SKS**

- ▶ [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](#)

**Metathesaurus Relational Records Request:**

Select UMLS Release:

1) Enter a term or concept unique identifier (CUI):

2) Select the UMLS table whose raw records are to be returned.

**Per  
Cor  
Se**

- mrconso
- mrdef
- mrsty
- mrct
- mrrel
- mrset
- mrcc

Raw record return allows users to search for a concept using either a user entered concept unique identifier (CUI) or a term name which is entered into the text entry box below. Any of the UMLS tables listed in the drop down menu may be selected for return.

Pressing the *Perform Concept Search* button will update the display to show the raw records from the selected table based on the concept information found.

# Relational Records MRCNSO.RRF

```
CUI|LAT|TS|LUI|STT|SUI|ISPREF|AUI|SAUI|SCUI|SDUI|SAB|TTY|CODE|STR|SUPPRESS|CVF|
C0001403|CZE|P|L3180742|PF|S3708232|Y|A3910108|||D000224|MSHCZE|MH|D000224|ADDISONOVA NEMOC|
C0001403|DUT|P|L2048638|PF|S2386860|N|A6566810|||10001130|MDRDUT|LT|10001130|Addison, ziekte
C0001403|DUT|P|L2048638|PF|S2386860|Y|A3931189|||D000224|MSHDUT|MH|D000224|Addison, ziekte v
C0001403|DUT|S|L2048637|PF|S2386859|Y|A3931188|||D000224|MSHDUT|SY|D000224|Addison, syndroom
C0001403|DUT|S|L2528364|PF|S2985131|Y|A3966882|||D000224|MSHDUT|SY|D000224|Ziekte van Addisc
C0001403|DUT|S|L3205108|PF|S3732602|N|A5146733||E27.1||ICD10DUT|PT|E27.1|Primaire bijniersch
C0001403|DUT|S|L3205108|PF|S3732602|Y|A3970882|||D000224|MSHDUT|SY|D000224|Primaire bijniers
C0001403|DUT|S|L4999233|PF|S5686738|Y|A6627443|||10036696|MDRDUT|LT|10036696|primair hypoadr
C0001403|DUT|S|L4999270|PF|S5686775|N|A6627493|||10052381|MDRDUT|LT|10052381|primaire bijnie
C0001403|DUT|S|L4999270|PF|S5686775|Y|A6627494|||10052381|MDRDUT|PT|10052381|primaire bijnie
C0001403|DUT|S|L5012413|PF|S5699917|Y|A6645695|||10013096|MDRDUT|LT|10013096|ziekte van Addi
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388276|||A0D|DE|0000006012|Addison's disease|0||
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388277|||0060-3321|CSP|PT|0060-3321|Addison's diseas
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388279|||LCH|PT|U000061|Addison's disease|0||
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388280|||10001390|MDR|LT|10001130|Addison's disease|
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388281|||RCD|PT|C1541|Addison's disease|3||
```

# Semantic Network Searching

- ◆ Select Tab along top
- ◆ Quick search
- ◆ Advanced Search on right-hand side

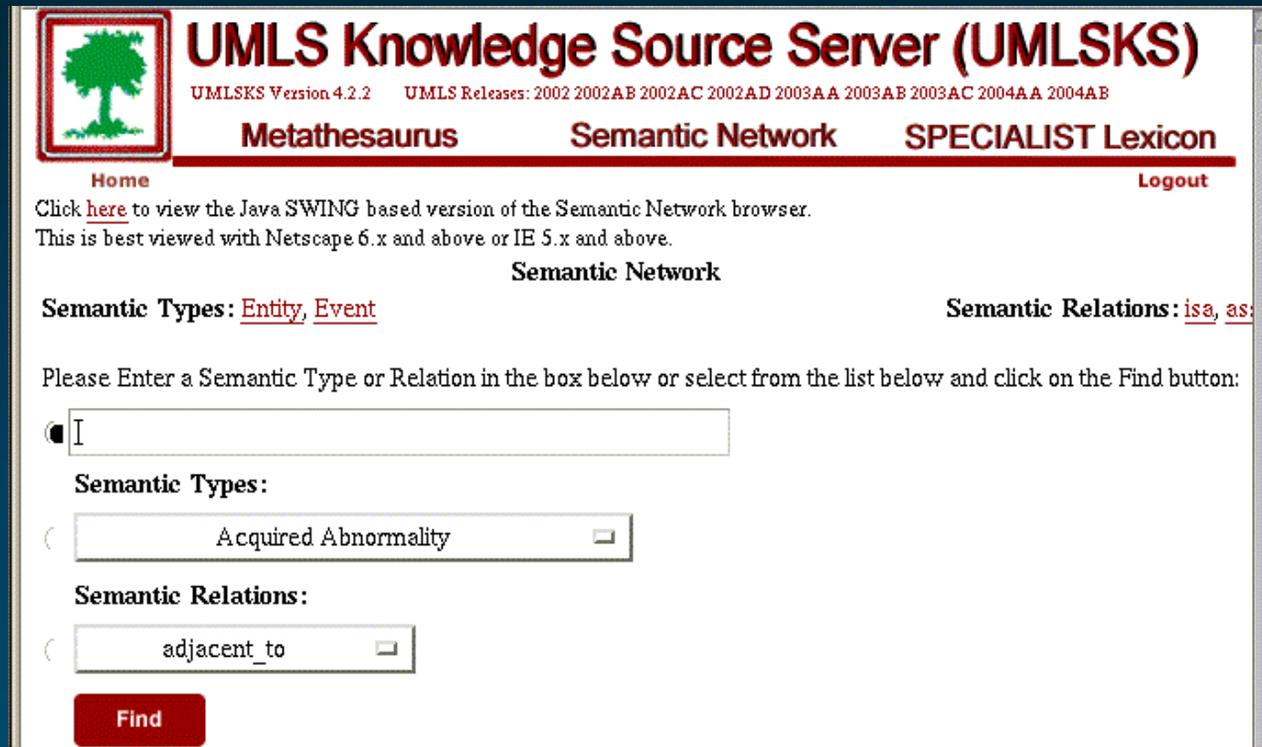
The screenshot shows the UMLS Knowledge Source Server (UMLSKS) website. The browser address bar displays <http://umlsks.nlm.nih.gov/kss/servlet/Turbine/action/KssLogin>. The page title is "UMLS Knowledge Source Server (UMLSKS)". Below the title, there is a navigation bar with tabs for "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". The "Semantic Network" tab is currently selected. The page is divided into several sections:

- About the UMLSKS:** Includes links for Home, Overview, Frequently Asked Questions, Edit Views/Profile, Downloads, UMLS Knowledge Sources, RxNorm Files, SNOMED CT®, Mappings, VA/KP Problem List, and Developer's API.
- Documentation:** Includes links for User's Guide, Developer's Guide, Developer's API Javadocs, UMLS Documentation Set, and RxNorm Documentation.
- Resources:** Includes links for NLP & Lexical Resources, Semantic Network Resources, and Metathesaurus Resources.
- Quick Search:** Features a search form with a dropdown for "Select UMLS Release:" (set to 2007AB) and a text input for "Enter search value:" (containing "addison's disease"). Below the form are three buttons: "Metathesaurus Concept Search", "Semantic Network Search", and "SPECIALIST Lexicon Search".
- Advanced Searches:** Includes a link for "Metathesaurus Advanced Search" and a link for "Semantic Network Browser".
- What's New:** Contains two news items: "The UMLS 2007AB files are now available for download and searching. (July 9, 2007, 8:00 AM)" and "Transfer of ownership of SNOMED CT® improves U.S. use rights, requires change to UMLS license. <more>".

At the bottom of the page, there is a footer with the following text: "Users are responsible for compliance with UMLS copyright restrictions", "Lister Hill National Center for Biomedical Communications", "U.S. National Library of Medicine (NLM), 3600 Rockville Pike, Bethesda, MD 20894", "National Institutes of Health (NIH)", "Department of Health & Human Services", "Copyright Privacy Accessibility Freedom of Information Act", and "Site Last Updated: 15 May 2007 Comments/Suggestions? Email [outserv@nlm.nih.gov](mailto:outserv@nlm.nih.gov) with your input."



# Semantic Network Search



The screenshot shows the UMLS Knowledge Source Server (UMLSKS) interface. At the top, there is a logo of a tree and the title "UMLS Knowledge Source Server (UMLSKS)". Below the title, it says "UMLSKS Version 4.2.2" and "UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB". The main navigation bar includes "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". There are links for "Home" and "Logout".

Click [here](#) to view the Java SWING based version of the Semantic Network browser.  
This is best viewed with Netscape 6.x and above or IE 5.x and above.

**Semantic Network**

Semantic Types: [Entity](#), [Event](#)      Semantic Relations: [isa](#), [as](#)

Please Enter a Semantic Type or Relation in the box below or select from the list below and click on the Find button:

Semantic Types:

(  )

Semantic Relations:

(  )

◆ Enter search string

-or-

◆ Select semantic type

-or-

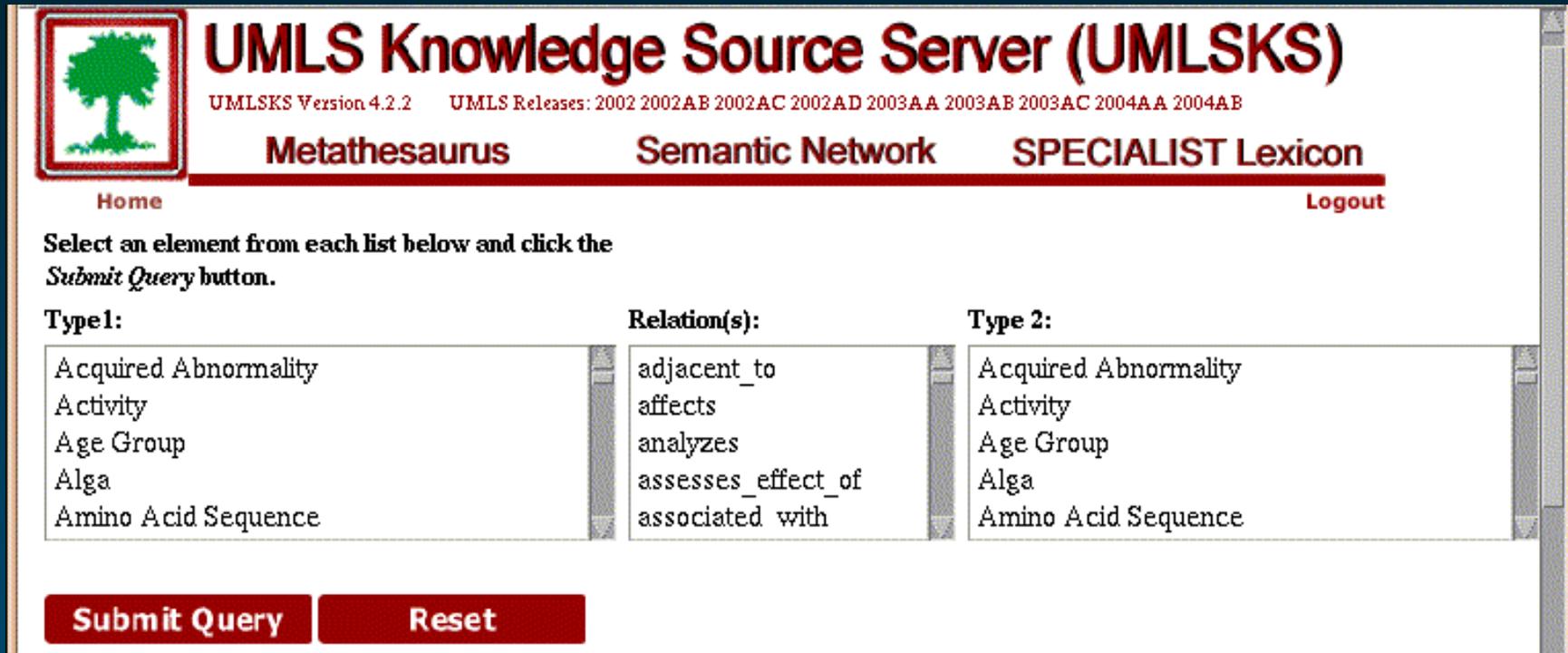
◆ Select semantic relation

# Semantic Type Clinical Drug

The screenshot displays the 'Metathesaurus Semantic Network SPECIALIST Lexicon' interface. At the top, there are navigation links for 'Home', 'Logout', and 'Click here to view the Java SWING based version of the Semantic Network browser'. Below this, there are search options: 'Meta Concepts', 'Relations', and 'Raw Records'. The main content area shows the 'Semantic Type: Clinical Drug' with a TUI of 'T200' and a definition: 'A pharmaceutical preparation as produced by the manufacturer. The name usually includes the substance, its strength, and the form, but may include the substance and only one of the other two items.' To the left, there is a search input field and a 'Find' button. Below the search field, there are sections for 'Semantic Types' (with 'Entity' and 'Event' listed) and 'Semantic Relations' (with 'adjacent\_to' selected). A hierarchical tree of 'Entity' types is visible on the left, including 'Physical Object', 'Organism', 'Plant', 'Alga', 'Fungus', 'Virus', 'Rickettsia or Chlamydia', 'Bacterium', 'Animal', 'Invertebrate', 'Vertebrate', 'Amphibian', 'Bird', 'Fish', 'Reptile', and 'Mammal'.

- ◆ Browse ST hierarchy
- ◆ View Concepts with ST
- ◆ View Relations valid for the ST
- ◆ View Raw Relational Records

# Show Relations Between Types



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

[Home](#) [Logout](#)

[Metathesaurus](#) [Semantic Network](#) [SPECIALIST Lexicon](#)

Select an element from each list below and click the *Submit Query* button.

Type 1:	Relation(s):	Type 2:
Acquired Abnormality	adjacent_to	Acquired Abnormality
Activity	affects	Activity
Age Group	analyzes	Age Group
Alga	assesses_effect_of	Alga
Amino Acid Sequence	associated with	Amino Acid Sequence

[Submit Query](#) [Reset](#)

- ◆ Validates whether a selected Semantic Relationship (SR) holds between two selected Semantic Types (ST)

# SPECIALIST Lexicon Searching

◆ Select Tab along top

◆ Quick search

The screenshot shows the UMLS Knowledge Source Server (UMLSKS) website. The browser address bar displays <http://umlsks.nlm.nih.gov/kss/servlet/Turbine/action/KssLogin>. The page title is "UMLS Knowledge Source Server (UMLSKS)". Below the title, there is a navigation bar with tabs for "Metathesaurus", "Semantic Network", and "SPECIALIST Lexicon". The "SPECIALIST Lexicon" tab is selected. The page content is organized into several sections:

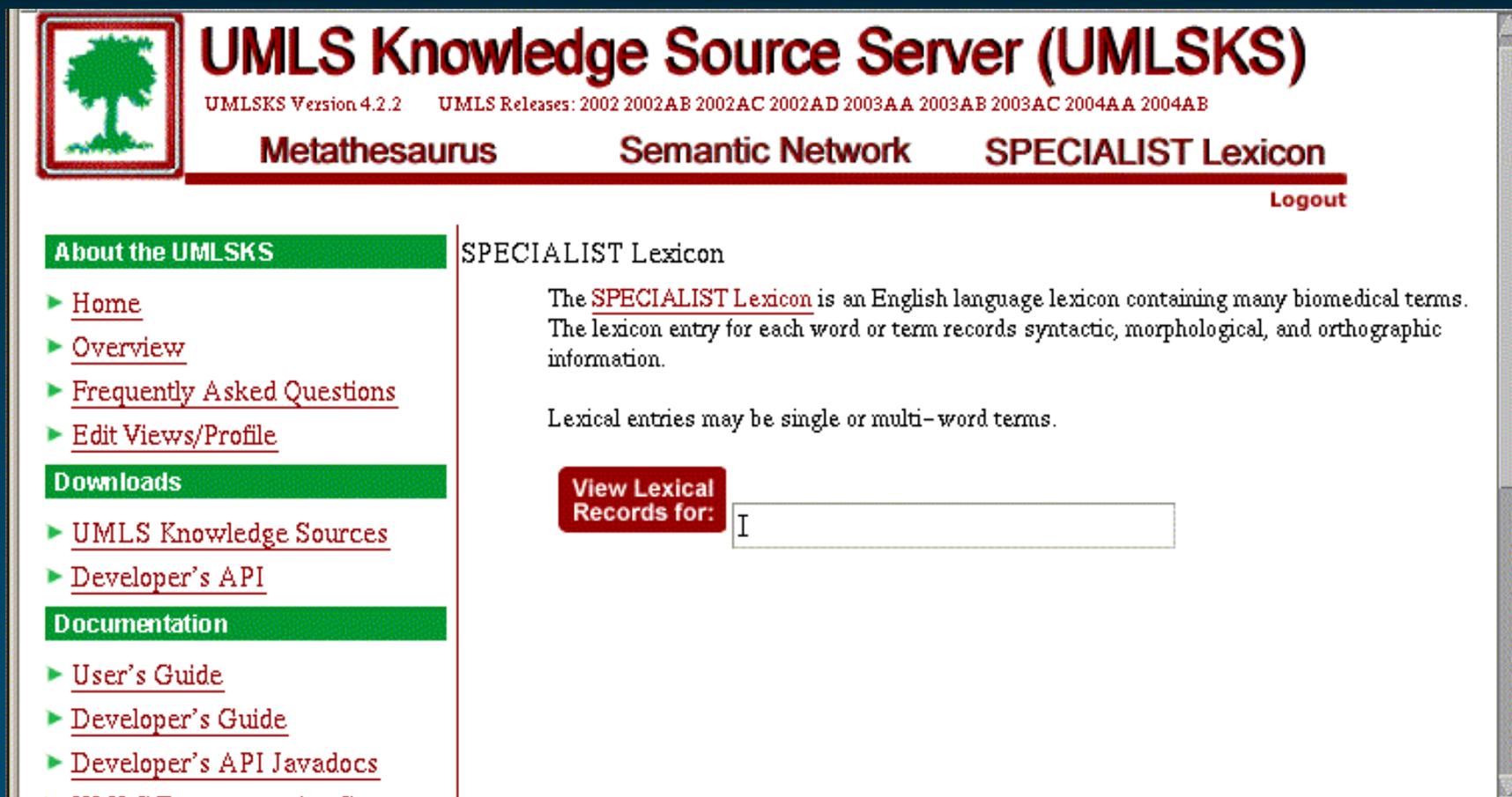
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# SPECIALIST Lexicon Search



**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](#)

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**SPECIALIST Lexicon**

The **SPECIALIST Lexicon** is an English language lexicon containing many biomedical terms. The lexicon entry for each word or term records syntactic, morphological, and orthographic information.

Lexical entries may be single or multi-word terms.

**View Lexical Records for:**

# SPECIALIST Lexical Record



## UMLS Knowledge Source Server (UMLSKS)

UMLSKS Version 4.2.2    UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB

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## Specialist Lexical Record

```
{base=Addison's disease
entry=E0000160
cat=noun
variants=uncount
variants=reg
}
```

View "Addison's disease" in relational format.

---

# UMLS Resources

## ◆ NLP & Lexical Resources

- MetaMap Transfer (MMTx)
- Word Sense Disambiguation (WSD) Test Collection

## ◆ Semantic Network

- Semantic Navigator
- Semantic Groups

## ◆ Metathesaurus

- String Properties

UMLS Knowledge Source Server (UMLSKS)

UMLSKS Version 5.0 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB 2004AC 2005AA 2005AB 2005AC 2006AA 2006AB 2006AC 2006AD 2007AA 2007AB

Metathesaurus Semantic Network SPECIALIST Lexicon

Logout

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- ▶ [RxNorm Documentation](#)

**Resources**

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- ▶ [Metathesaurus Resources](#)

**Quick Search**

Select UMLS Release: 2007AB

Enter search value: addison's disease

[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**

- ▶ The UMLS 2007AB files are now available for download and searching. (July 9, 2007, 8:00 AM)
- ▶ Transfer of ownership of SNOMED CT® improves U.S. use rights, requires change to UMLS license. [<more>](#)

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### Resources

- ▶ [NLP & Lexical Resources](#)
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- ▶ [Metathesaurus Resources](#)



# The “new” UMLSKS (coming soon)

The screenshot displays the UMLS Knowledge Source Server (UMLSKS) website. At the top, there is a red header with the UMLS logo (a tree) and the title "UMLS Knowledge Source Server (UMLSKS)". Navigation links include "Home", "Help", and "Log Off". Below the header, a secondary navigation bar contains buttons for "Home", "Metathesaurus", "SPECIALIST Lexicon", "Semantic Network", "My UMLSKS", and "Source View". The main content area is titled "Hello, Basic Umls-user" and features a "Change Content" link. The page is organized into three main columns:

- Left Column:** Contains three sections: "Downloads" (with links to UMLS Knowledge Sources, RxNorm Files, and Developer's API), "Documentation" (with links to Overview, Frequently Asked Questions, UMLSKS User's Guide, Developer's API Javadocs, UMLS Documentation, and RxNorm Documentation), and "Resources" (with links to Lexical Tools, MetaMap Transfer, WSD Collection, and Semantic Navigator).
- Center Column:** Features an "Overview" section with the heading "Unified Medical Language System Knowledge Source Server (UMLSKS) Fact Sheet Version 4.x". Below this is a "Background" section with two paragraphs of text. The first paragraph states: "The Unified Medical Language System® (UMLS) approach involves the development of a set of widely distributed Knowledge Sources (Metathesuarus, Semantic Network, and SPECIALIST Lexicon) that can be used by a variety of applications to compensate for differences in the way concepts are expressed in a variety of computerized biomedical sources." The second paragraph states: "The UMLS Knowledge Source Server (UMLSKS) is a computer application that provides Internet access to the Knowledge Sources and other related resources made available by developers using the UMLS. Its purpose is to make UMLS data more accessible to users, and in particular to system developers. The system architecture allows remote site users (individuals as well as computer programs) to send requests to a server at the National Library of Medicine (NLM). Access to the system is provided through the World Wide Web, an Extensible Markup Language (XML)-based socket programming interface, and through an Application Programmer Interface (API)."
- Right Column:** Contains a "What's New" section with a bulleted list: "Web Services Implementation of the API." and "Web Interface based on Portal technology."



# UMLS Knowledge Source Server (UMLSKS)

Home Help Log Off

Home Metathesaurus SPECIALIST Lexicon Semantic Network My UMLSKS Source View

Hello, Basic Umls-user

Choose a Section:

Metathesaurus: Default View

Metathesaurus: Lexical View

Metathesaurus: Raw View

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## Resources

Lexical Tools  
MetaMap Transfer

## Metathesaurus

Change

### Metathesaurus Search

Enter term

Term:

C0001403

OK

Release:

2007AA

Index:

Exact Match

Sources:

All sources

### Basic Concept

Concept Information - UMLS Release 2007AA

[ C0001403 ] Addison's disease

Definition:

CRISP Thesaurus/A0388277

MeSH/A6954527

An adrenal disease characterized by the progressive destruction of the ADRENAL CORTEX, resulting in insufficient production of ALDOSTERONE and HYDROCORTISONE. Clinical symptoms include ANOREXIA; NAUSEA; WEIGHT LOSS; MUSCLE WEAKNESS; and HYPERPIGMENTATION of the SKIN due to increase in circulating levels of ACTH precursor hormone which stimulates MELANOCYTES.

Semantic Types:

Disease or Syndrome

Atoms: (179)

Addison's disease[ A0388276/Alcohol and Other Drug Thesaurus/DE/0000006012 ]

ADDISON DISEASE[ A0385542/Clinical Problem Statements/PT/0022753 ]

ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)[ A0385630/COSTAR/PT/U000087 ]

Addison's disease[ A0388277/CRISP Thesaurus/PT/0060-3321 ]

Printer-Fri



# UMLS Knowledge Source Server (UMLSKS)

Hello, Basic Umls-user

Choose a Section:

**Metathesaurus: Default View**

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## Metathesaurus

[Change](#)

### Metathesaurus Search

Enter term

Term:

C0001403

Release:

2007AA

Index:

Exact Match

Sources:

MedDRA Japanese

### Basic Concept

[Printer-Friendly](#)

- Concept Information - UMLS Release 2007AA
  - [ C0001403 ] Addison's disease
  - Semantic Types:
    - Atoms: (12)
      - アジソン病[ A11403670/MedDRA Japanese/LT/10001130 ]
      - アジソン病[ A11505319/MedDRA Japanese/OL/10013096 ]
      - アジソン病[ A11538038/MedDRA Japanese/PT/10001130 ]
      - 原発性副腎機能低下症[ A11549716/MedDRA Japanese/LT/10036696 ]
      - 原発性副腎機能不全[ A11513345/MedDRA Japanese/LT/10052381 ]
      - 原発性副腎機能不全[ A11520927/MedDRA Japanese/PT/10052381 ]
      - ケンバ\*グエイカジンキノウゲン[ A11493725/MedDRA Japanese/LT/10052381 ]
      - ケンバ\*グエイカジンキノウゲン[ A11500944/MedDRA Japanese/PT/10052381 ]
      - アジ ソビ ヨ[ A11443237/MedDRA Japanese/LT/10001130 ]
      - アジ ソビ ヨ[ A11525328/MedDRA Japanese/OL/10013096 ]
      - アジ ソビ ヨ[ A11538039/MedDRA Japanese/PT/10001130 ]
      - ケンバ\*グエイカジンキノウゲン[ A11529874/MedDRA Japanese/LT/10036696 ]

# UMLS Semantic Navigator

---

- ◆ Web-based
  - <http://mor.nlm.nih.gov/perl/semnav.pl>
- ◆ Concept- and relation-centric
- ◆ Displays contexts graphically
- ◆ Displays all relations simultaneously
- ◆ Excludes hierarchical cycles in the UMLS graph
- ◆ Search
  - By CUI
  - By word



# RRF Browser

---

- ◆ Distributed with the UMLS
  - Along with MetamorphoSys
- ◆ Standalone
- ◆ Can browse particular subsets of the Metathesaurus
- ◆ Search
  - By code
  - By CUI
  - By word

# RRF Browser

Rich Release Format Browser 2007AB C0001403

File Edit Options Help

Monospaced 11 D:\2007AB\META

CUI Search Code Search Tree Browser Word Search

Enter search terms: (English)

addison's disease

Search..

Select a concept. (3 results)

- C0001403 Addison Disease
- C0002892 Anemia, Pernicious
- C0162309 Adrenoleukodystrophy

Raw View Report View

- Concept:** [CUI C0001403] Addison Disease
- Semantic Type**  
Disease or Syndrome
- Definition**  
MSH/MH|An adrenal disease characterized by the progressive destruction of the ADRENAL CORTEX, resulting in insufficient production of ALDOSTERONE and HYDROCORTISONE. Clinical symptoms include ANOREXIA; NAUSEA; WEIGHT LOSS; MUSCLE WEAKNESS; and HYPERPIGMENTATION of the SKIN due to increase in circulating levels of ACTH precursor hormone which stimulates MELANOCYTES.
- Atoms (17):** (Sorted by Source, String)
  - ADDISON DIS [A12078968/MSH/DEV/D000224] Y
  - ADDISONS DIS [A12075312/MSH/DEV/D000224] Y
  - Addison Disease [A6954527/MSH/MH/D000224]
  - Addison's Disease [A6954528/MSH/EN/D000224]
  - Addisons Disease [A0019742/MSH/PM/D000224]
  - Adrenal Insufficiency, Primary [A6993206/MSH/PM/D000224]
  - Adrenocortical Insufficiencies, Primary [A6993209/MSH/PM/D000224]
  - Adrenocortical Insufficiency, Primary [A6993210/MSH/PM/D000224]
  - Disease, Addison [A0049628/MSH/PM/D000224]

Knowledge Source Server  
*Application Programming Interface*

# UMLSKS 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 UMLSKS 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](http://umlsks.nlm.nih.gov)
  - Port: 8042

# Developer's Guide

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## Developer's Guide

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### About This Guide

This guide describes the installation and usage of the programmatic interface for the UMLS 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 a client listening to the socket.

# Documentation Java API



## UMLS Knowledge Source Server (UMLSKS)

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### UMLSKS API Download

The following instructions describe the procedures for downloading and installing the UMLSKS API. The sections include

- ◆ [Downloading the UMLSKS API](#)
- ◆ [Building the Example . java Files](#)
- ◆ [Running the Client](#)
- ◆ [Running the ExpertClient](#)
- ◆ [Running the SocketClient](#)
- ◆ [Running the StandardQueryClient](#)
- ◆ [Available Documentation](#)
- ◆ [Sample Output and XML Query Examples](#)

### Downloading the UMLSKS API

The complete set of files for the UMLSKS API may be downloaded by clicking on the link below. Clicking the

# Documentation Javadocs

All Classes

Packages

- [gov.nih.nlm.kss.api](#)
- [gov.nih.nlm.kss.mode](#)

All Classes

- [AdjEntry](#)
- [AdvEntry](#)
- [AssocExprsExecutor](#)
- [AssociatedExp](#)
- [AssociatedExpVector](#)
- [AssociativeRelExister](#)
- [AssociativeRelation](#)
- [AssociativeRelationVe](#)
- [Attr](#)
- [AttrVector](#)
- [AttributeContext](#)
- [AttributeValue](#)
- [AuxEntry](#)
- [BasicConceptPropsE](#)
- [COCContext](#)
- [CUIExecutor](#)
- [CatEntry](#)
- [CompareUtil](#)
- [Concept](#)
- [ConceptAttribute](#)
- [ConceptAttributeVect](#)
- [ConceptAttrsExecutor](#)
- [ConceptDelta](#)
- [ConceptId](#)
- [ConceptIdVector](#)
- [ConceptNameExecute](#)
- [ConceptPropsExecute](#)
- [ConceptVector](#)
- [ConstantSettings](#)
- [Context](#)

**Overview** Package Class Use [Tree](#) [Deprecated](#) [Index](#) [Help](#)

PREV NEXT [FRAMES](#) [NO FRAMES](#)

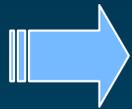
## Packages

<a href="#">gov.nih.nlm.kss.api</a>	
<a href="#">gov.nih.nlm.kss.models</a>	
<a href="#">gov.nih.nlm.kss.models.lex</a>	
<a href="#">gov.nih.nlm.kss.models.meta</a>	
<a href="#">gov.nih.nlm.kss.models.meta.assocExp</a>	
<a href="#">gov.nih.nlm.kss.models.meta.attribute</a>	
<a href="#">gov.nih.nlm.kss.models.meta.concept</a>	
<a href="#">gov.nih.nlm.kss.models.meta.context</a>	
<a href="#">gov.nih.nlm.kss.models.meta.cooccurrence</a>	
<a href="#">gov.nih.nlm.kss.models.meta.deltas</a>	
<a href="#">gov.nih.nlm.kss.models.meta.locator</a>	
<a href="#">gov.nih.nlm.kss.models.meta.meshentry</a>	
<a href="#">gov.nih.nlm.kss.models.meta.relation</a>	
<a href="#">gov.nih.nlm.kss.models.meta.source</a>	
<a href="#">gov.nih.nlm.kss.models.sem</a>	
<a href="#">gov.nih.nlm.kss.models.sem.rels</a>	
<a href="#">gov.nih.nlm.kss.models.sem.units</a>	
<a href="#">gov.nih.nlm.kss.query</a>	
<a href="#">gov.nih.nlm.kss.query.lex</a>	
<a href="#">gov.nih.nlm.kss.query.meta</a>	
<a href="#">gov.nih.nlm.kss.query.sem</a>	
<a href="#">gov.nih.nlm.kss.util</a>	
<a href="#">gov.nih.nlm.nls.lexCheck.Lib</a>	



# Sample XML query (1) Current version

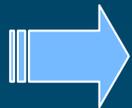
```
<?xml version="1.0"?>  
<getCurrentUMLSVersion version="1.0"/>
```



```
<?xml version="1.0"?>  
<CurrentUMLSYear version="1.0">  
    2007AB  
</CurrentUMLSYear>
```

## Sample XML query (2) Concepts by string

```
<?xml version="1.0"?>
<findCUI version="1.0">
<conceptName>appendectomy</conceptName>
<language>ENG</language>
<exact/>
<noSuppressibles/>
</findCUI>
```



```
<?xml version="1.0"?>
<ConceptIdCollection version="1.0">
  <release>2004AB</release>
  <conceptId>
    <cui>C0003611</cui>
    <cn>Appendectomy</cn>
  </conceptId>
</ConceptIdCollection>
```



# Sample XML query (3) Concepts properties

```
<?xml version="1.0"?>
<getSemanticType version="1.0">
<cui>C0033572</cui>
</getSemanticType>
```

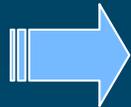


```
<?xml version="1.0"?>
<SemanticTypeCollection version="1.0">
<release>2004AB</release>
<cui>C0033572</cui>
<cn>Prostate</cn>
  <semanticType>
    <tui>T023</tui>
    <sty>Body Part, Organ,
      or Organ Component</sty>
  </semanticType>
</SemanticTypeCollection>
```



# Sample XML query (4) Relationships

```
<?xml version="1.0"?>
<getRelations version="1.0">
<cui>C0033572</cui>
<rel>RO</rel>
</getRelations>
```



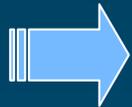
```
<?xml version="1.0"?>
<RelationCollection version="1.0">
[...]
```

```
  <relation>
    <aui>A3188910</aui>
    <sab>SNOMEDCT</sab>
    <relSource>
      <cui>C0007112</cui>
      <cn>Adenocarcinoma of prostate</cn>
      <aui>A3318222</aui>
      <rel>RO</rel>
      <rui>R54806623</rui>
      <rela>has_finding_site</rela>
    </relSource>
  </relation>
[...]
```



# Sample XML query (5) All semantic type IDs

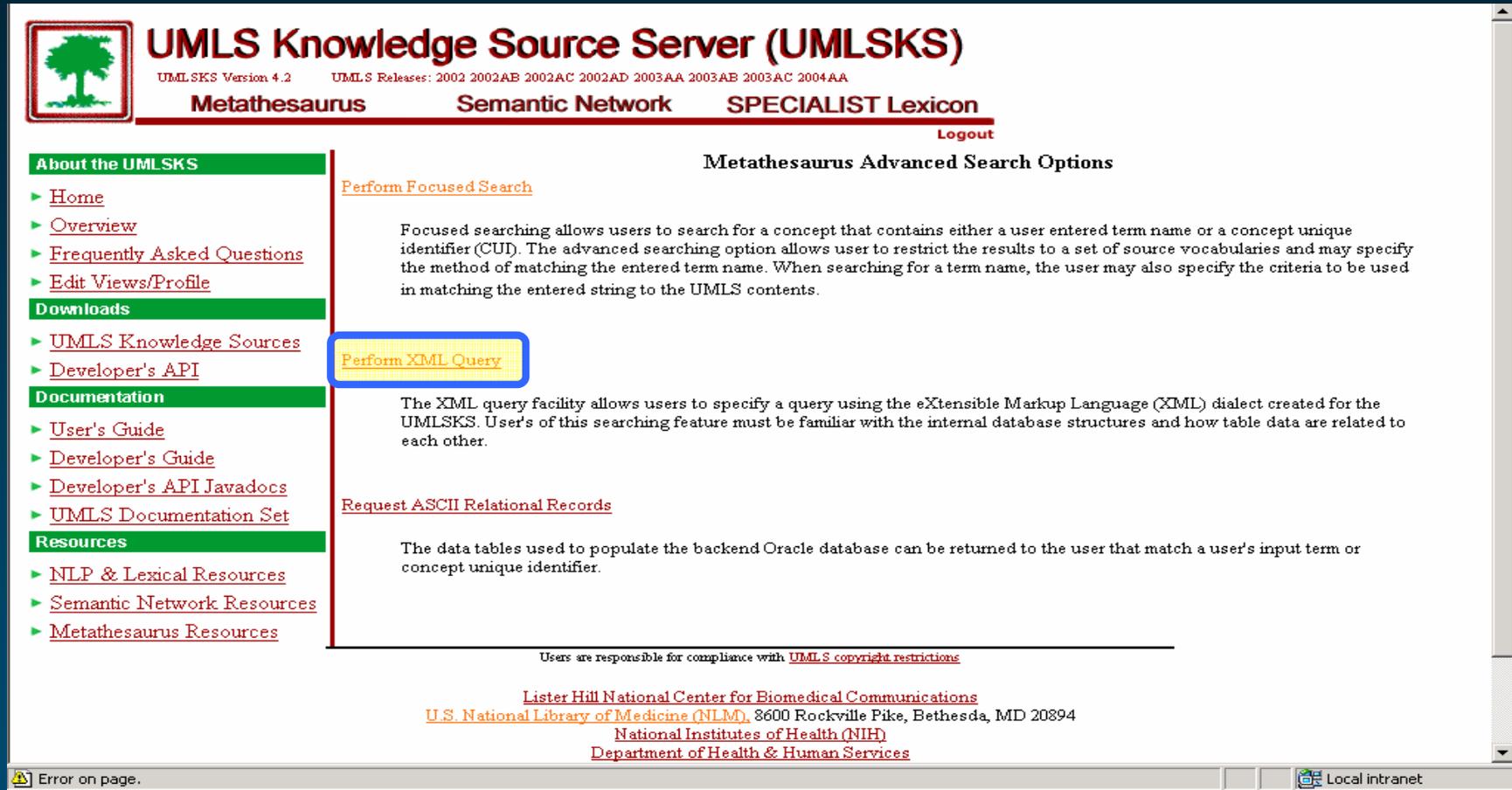
```
<?xml version="1.0"?>  
<listSemTypeIds version="1.0">  
</listSemTypeIds>
```



```
<?xml version="1.0"?>  
<SemNetIdCollection version="1.0">  
  <release>2004AB</release>  
  <semnetId>  
    <name>Acquired Abnormality</name>  
    <ui>T020</ui>  
    <semtype/>  
  </semnetId>  
  <semnetId>  
    <name>Activity</name>  
    <ui>T052</ui>  
    <semtype/>  
  </semnetId>
```

[...]

# Performing XML queries from UMLSKS



**UMLS Knowledge Source Server (UMLSKS)**  
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[Metathesaurus](#) [Semantic Network](#) [SPECIALIST Lexicon](#) [Logout](#)

### Metathesaurus Advanced Search Options

- [Perform Focused Search](#)

Focused searching allows users to search for a concept that contains either a user entered term name or a concept unique identifier (CUI). The advanced searching option allows user to restrict the results to a set of source vocabularies and may specify the method of matching the entered term name. When searching for a term name, the user may also specify the criteria to be used in matching the entered string to the UMLS contents.
- [Perform XML Query](#)**

The XML query facility allows users to specify a query using the eXtensible Markup Language (XML) dialect created for the UMLSKS. User's of this searching feature must be familiar with the internal database structures and how table data are related to each other.
- [Request ASCII Relational Records](#)

The data tables used to populate the backend Oracle database can be returned to the user that match a user's input term or concept unique identifier.

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Error on page. Local intranet

# Performing XML queries from UMLSKS

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- [Metathesaurus Resources](#)

**Metathesaurus XML Query**

The UMLSKS provides a facility for interpreting two forms of XML:

- Standard API commands
- Specialized requests

"Standard" API commands are a set of XML documents in which each of the standard API commands (all but the query) are represented in the XML. For examples on the structure of this type of document, see the [examples](#)

"Specialized" requests are those XML documents that are structured for use with the `kssretriever.query` method. These types of queries are describe in [Chapter 4](#) of the UMLSKS User's Guide. User's of this type of query must be familiar with the internal database structures and how table data are related to each other.

Either type/paste the XML query into the text entry area or browse your local file system to locate an XML query file to upload to the UMLSKS. Once the query is entered, click the *Find* button to execute the query

Enter the XML query:

```
<?xml version="1.0"?>  
<getRelations>  
<cui>C0033572</cui>  
<rel>RO</rel>  
</getRelations>
```

## Part II

### How to use the UMLS?

*(3) Installing the UMLS locally and  
Customizing the Metathesaurus  
using 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

# Using MetamorphoSys

---

- ◆ Simple to use
- ◆ Screens and tabs lead you through process
- ◆ Installs NLM data format files to local storage



# 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

# Why use MetamorphoSys?

---

## *Changing Default Settings*

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

# Customization is Critical

---

- ◆ Requires a clear understanding of:
  - Characteristics of source vocabularies
  - License arrangements
  - User's functional requirements
  - User's purpose and perspective
- ◆ Technical expertise

**... and requires a  
multidisciplinary technical team**



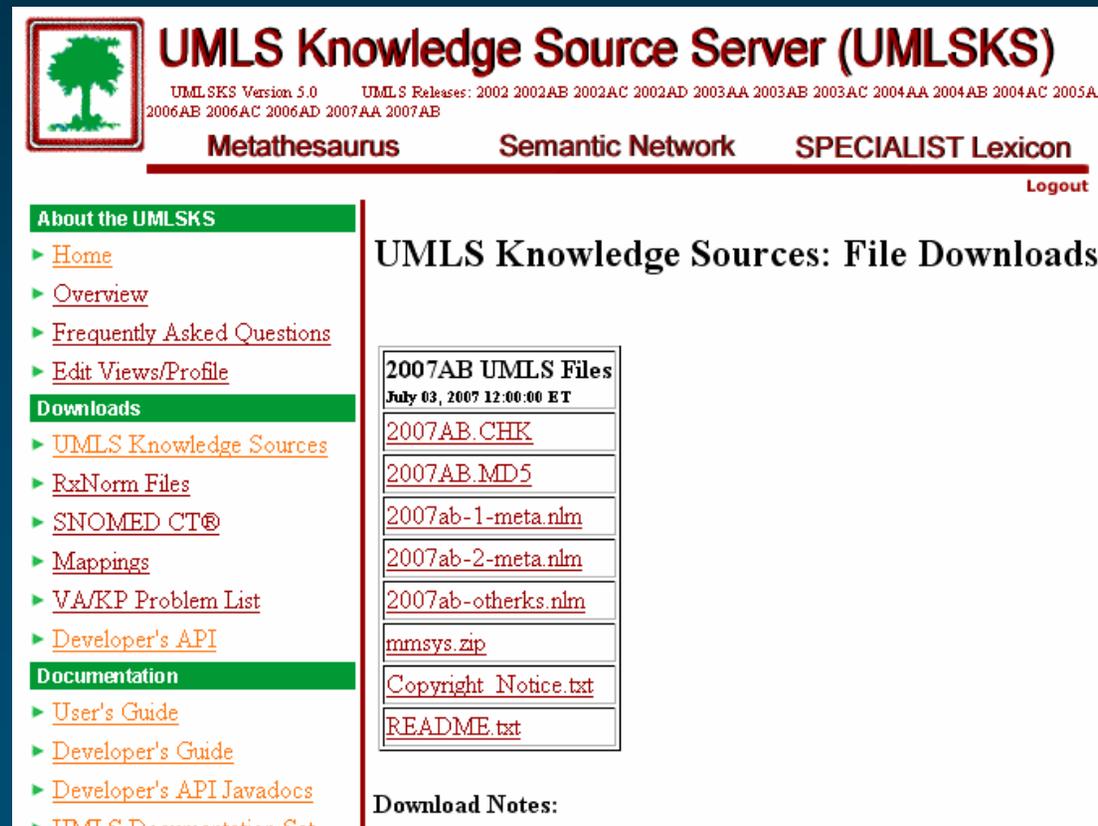
# Machine Requirements

---

- ◆ A fast CPU – 1 GHz or higher
- ◆ 1 GB RAM recommended (512 MB min.)
- ◆ 6x (or better) DVD drive
- ◆ 22 GB minimum free disk space
  
- ◆ Runs on Sun Solaris 8 & 9, Windows XP, NT, and 2000, Linux, and Mac
- ◆ 1-10 hours run time on platforms tested

# Download from UMLSKS ...

- ◆ High speed Internet connection required
- ◆ Read the README file for the release



**UMLS Knowledge Source Server (UMLSKS)**  
UMLSKS Version 5.0 UMLS Releases: 2002 2002AB 2002AC 2002AD 2003AA 2003AB 2003AC 2004AA 2004AB 2004AC 2005AA 2006AB 2006AC 2006AD 2007AA 2007AB

[Metathesaurus](#)   [Semantic Network](#)   [SPECIALIST Lexicon](#)   [Logout](#)

**About the UMLSKS**

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

**Downloads**

- ▶ [UMLS Knowledge Sources](#)
- ▶ [RxNorm Files](#)
- ▶ [SNOMED CT®](#)
- ▶ [Mappings](#)
- ▶ [VA/KP Problem List](#)
- ▶ [Developer's API](#)

**Documentation**

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

### UMLS Knowledge Sources: File Downloads

<b>2007AB UMLS Files</b> July 03, 2007 12:00:00 ET
<a href="#">2007AB.CHK</a>
<a href="#">2007AB.MD5</a>
<a href="#">2007ab-1-meta.nlm</a>
<a href="#">2007ab-2-meta.nlm</a>
<a href="#">2007ab-otherks.nlm</a>
<a href="#">mmsys.zip</a>
<a href="#">Copyright Notice.txt</a>
<a href="#">README.txt</a>

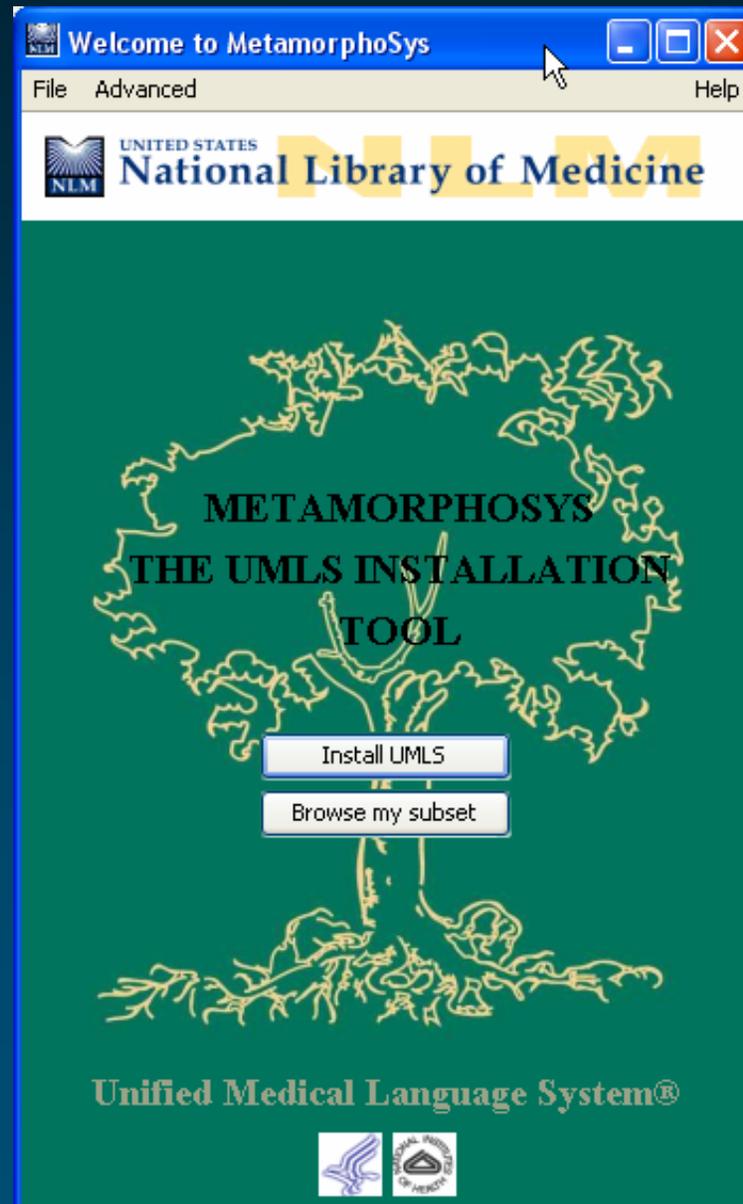
**Download Notes:**

## ...or DVD?

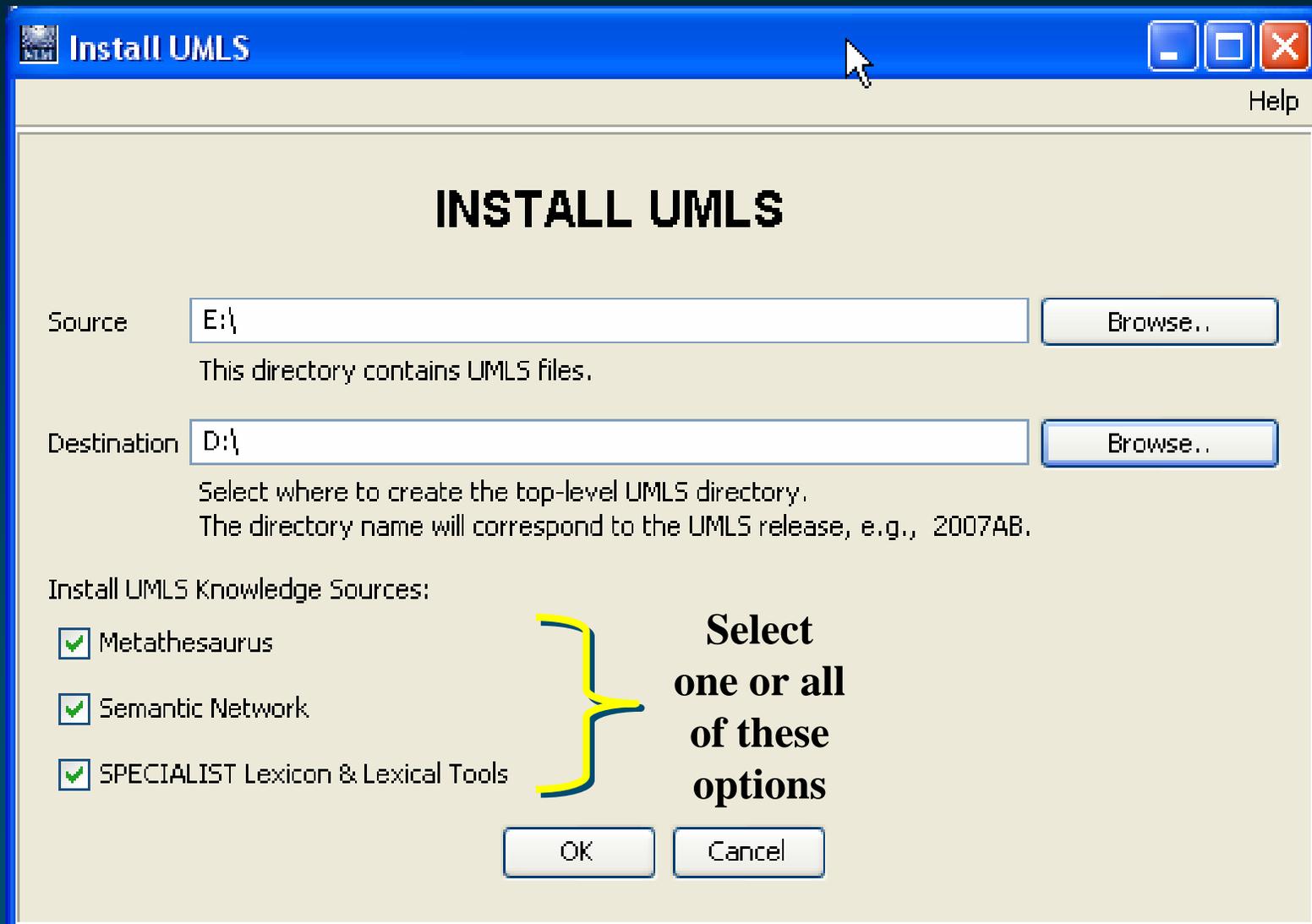
---

- ◆ Order at: [umls\\_support@nlm.nih.gov](mailto:umls_support@nlm.nih.gov)
- ◆ **Include your license number**
  
- ◆ Run MetamorphoSys from DVD
  - Windows
    - Autorun; or go to root directory and click on “windows\_mmsys.bat”
  
  - Linux, Solaris, Macintosh
    - open a terminal window, change to the root directory and type appropriate command: `./linux_mmsys.sh`, `./solaris_mmsys.sh`, `./macintosh_mmsys.sh`

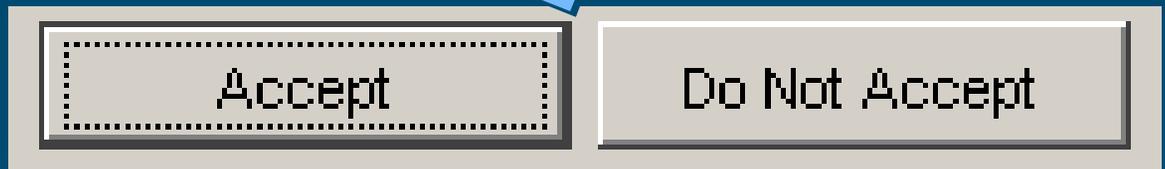
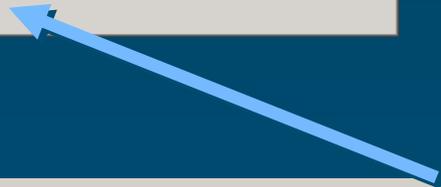
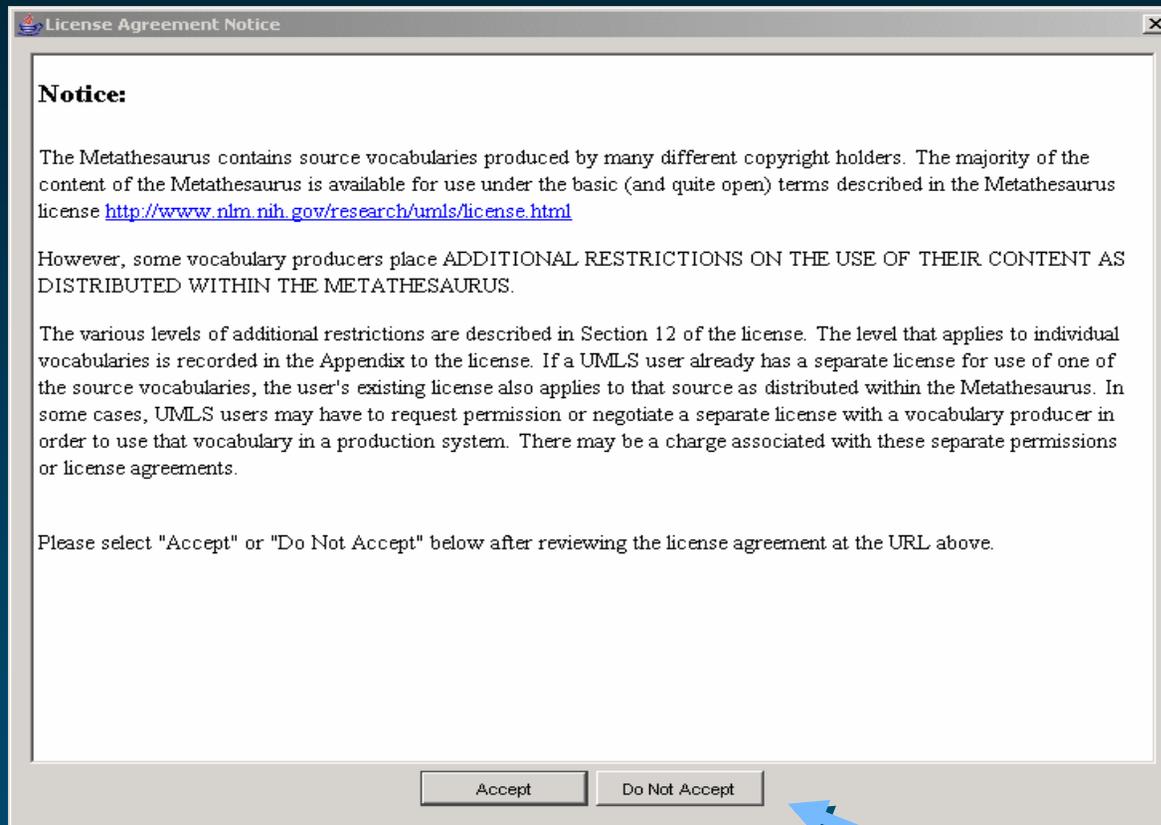
# Welcome Screen



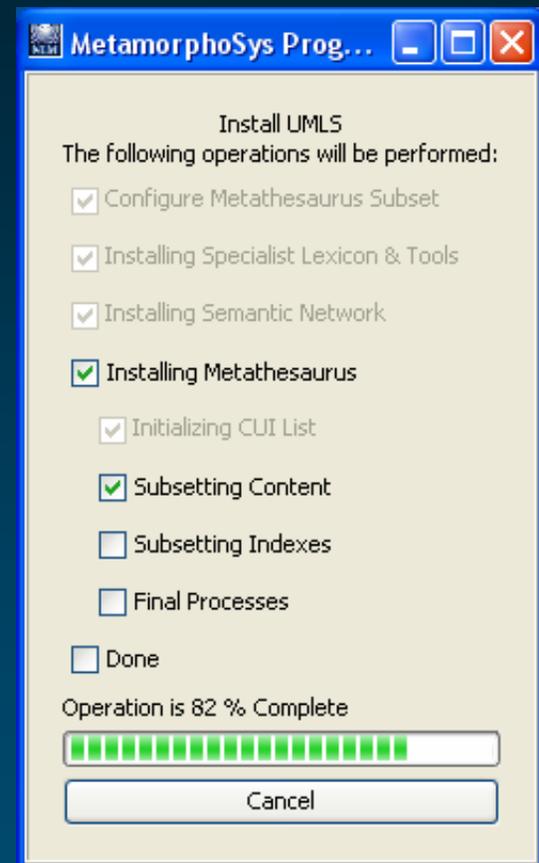
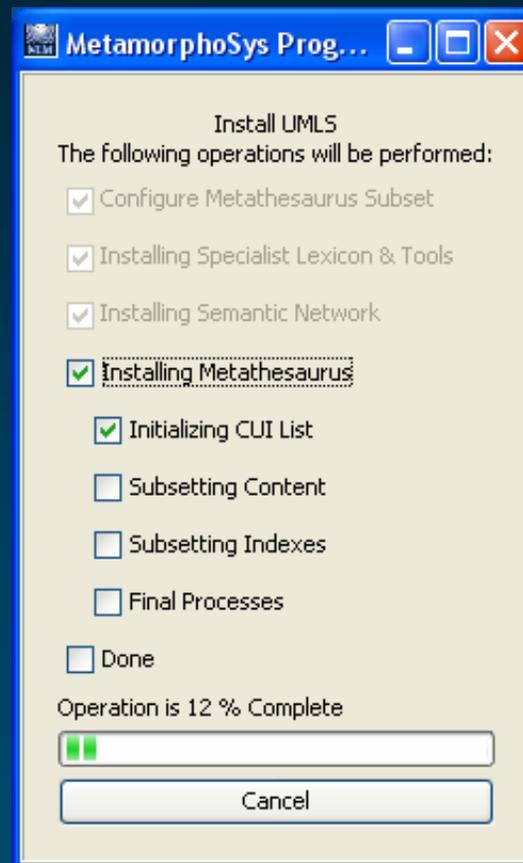
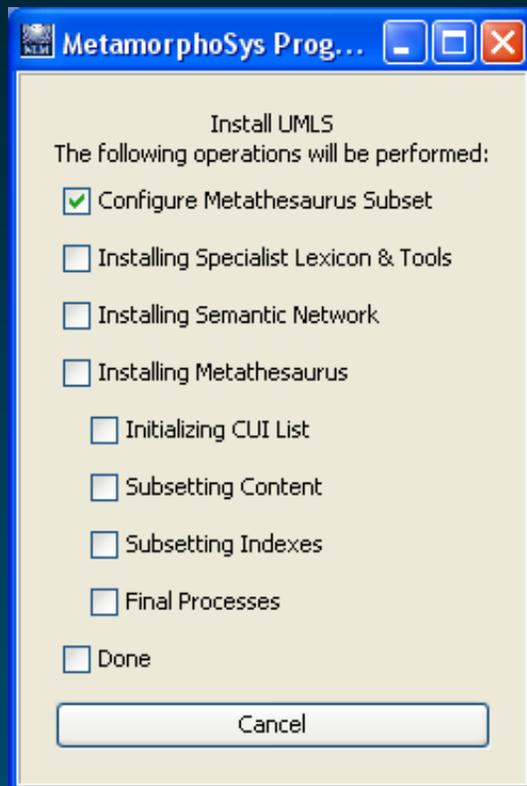
# Install UMLS



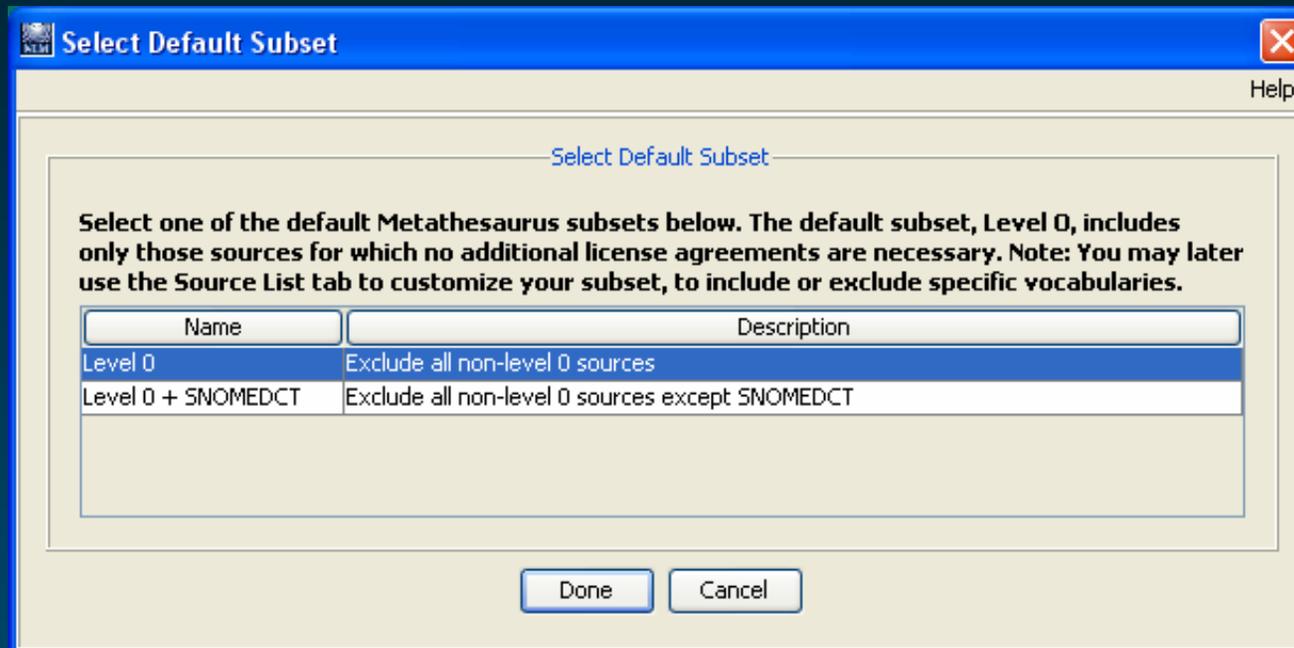
# UMLS License Notice



# Installation progress monitor



# Select a default subset



Level 0 → no separate additional license agreements

Level 0 + SNOMEDCT → Users from non-IHTSDO member countries must have separate license agreements

# Input Options Tab

The screenshot shows a window titled "UMLS Metathesaurus Configuration 2007AB" with a menu bar containing "File", "Edit", "Options", "Reset", "Done", and "Help". Below the menu bar are five tabs: "Input Options", "Output Options", "Source List", "Precedence", and "Suppressibility". The "Input Options" tab is active and contains the following text:

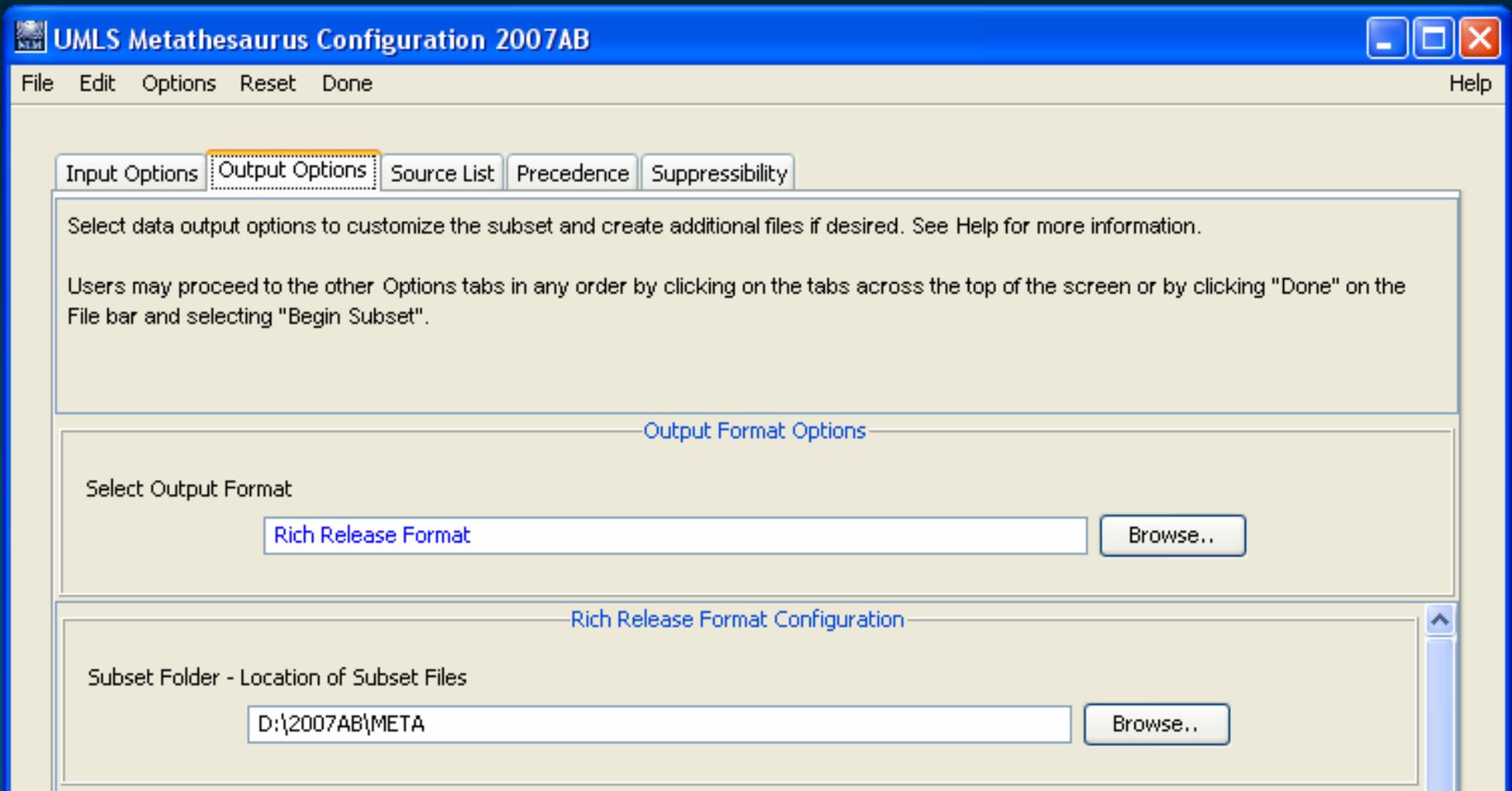
Select the format in which subset data was saved and indicate the location of the source folder that contains the existing subset configuration. See Help for more information.

Users may proceed to the other Options tabs in any order by clicking on the tabs across the top of the screen or by clicking "Done" on the File bar and selecting "Begin Subset".

The "Input Format Options" section includes a "Select Input Format" label, a text box containing "NLM Data File Format", and a "Browse.." button.

The "NLM Data File Format Configuration" section includes a "Source Folder - Location of Metathesaurus Files" label, a text box containing "E:\", and a "Browse.." button.

# Output Options Tab



# Output Options Tab

## Write Database Load Scripts

Write Oracle load script.

Write MySQL load script

## Source Abbreviation Format

Output versioned source abbreviations rather than versionless source abbreviations.

## Maximum Field Length

Truncate long fields to  characters.

## Eliminate Extended Unicode Characters

Remove records containing extended UTF-8 characters.

## Remove MTH only concepts

Remove concepts containing only MTH atoms.

## Calculate MD5 values for output files

Calculate MD5s for output files - writes mmsys.md5 file.

# Source List Tab

UMLS Metathesaurus Configuration 2007AB

File Edit Options Reset Done Help

Input Options Output Options **Source List** Precedence Suppressibility

Indicate below to INCLUDE or EXCLUDE selected sources.  
SELECTED sources appear on a dark background, e.g., AI/RHEUM, 1993 .

To undo selections and return to default source list, select "Reset Source List" from Reset menu at top.

Select sources to EXCLUDE from subset  
 Select sources to INCLUDE in subset

Sources to Exclude

* Full Source Name	Source Abbreviation	Source Family	Lang...	Level	Concepts
AI/RHEUM, 1993	AIR93	AIR	ENG	0	630
Alternative Billing Concepts, 2006	ALT2006	ALT	ENG	3	4661
Alcohol and Other Drug Thesaurus, 2000	AOD2000	AOD	ENG	0	15900
Authorized Osteopathic Thesaurus, 2003	AOT2003	AOT	ENG	0	278
Beth Israel Vocabulary, 1.0	BI98	BI	ENG	2	939
Canonical Clinical Problem Statement System, 1999	CCP5599	CCPSS	ENG	3	15266
Clinical Classifications Software, 2005	CCS2005	CCS	ENG	0	1107
Current Dental Terminology 2007-2008 (CDT-7/8), 2007-2008	CDT2007-2008	CDT	ENG	3	582
COSTAR, 1989-1995	COSTAR_89-95	COSTAR	ENG	0	3073
Medical Entities Dictionary, 2003	CPM2003	CPM	ENG	2	3078
Physicians' Current Procedural Terminology, Spanish Transla...	CPT015P	CPT	SPA	3	7632
Current Procedural Terminology, 2007	CPT2007	CPT	ENG	3	8847
CRISP Thesaurus, 2006	CSP2006	CSP	ENG	0	16702
COSTART, 1995	CST95	CST	ENG	0	3847
Common Terminology Criteria for Adverse Events, 2003	CTCAEV3	CTCAE	ENG	0	5598
Diseases Database, 2000	DDB00	DDB	ENG	3	169
German translation of ICD10, 1995	DMDICD10_1995	ICD10	GER	1	11308

Highlighted rows are excluded from the subset.

# Precedence Tab

- Ranks names by types of terms within sources
- Highest ranking name determines the Preferred Name

This screen sets the relative ranking of source/term type combinations. The ranking of sources and term types will affect the output subset. In particular, the name of a concept will be determined by the highest ranking term type in that concept. To move rows, either cut and paste rows using keyboard shortcuts, or drag and drop. See Help for more information.

Users may proceed to the other Options tabs in any order by clicking on the tabs across the top of the screen or by clicking "Done" on the File bar and selecting "Begin Subset".

Full Source Name	Source Abbreviation	Term Type
UMLS Metathesaurus	MTH	PN
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	MH
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	TQ
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	PEP
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	PEN
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	EP
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	EN
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	XQ
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	PXQ
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	NM
RxNorm Vocabulary, 07AA_070503F	RXNORM_07AA_070503F	SCD
RxNorm Vocabulary, 07AA_070503F	RXNORM_07AA_070503F	SBD
RxNorm Vocabulary, 07AA_070503F	RXNORM_07AA_070503F	SY

Cut and  
paste rows  
to alter the  
preferred  
name

# Suppressibility Tab

This screen contains the Suppressibility Filter, which specifies source/term type combinations to be suppressed. Users can customize the subset by selecting and deselecting source/term type combinations. See Help for more information.

Users may proceed to the other Options tabs in any order by clicking on the tabs across the top of the screen or by clicking "Done" on the File bar and selecting "Begin Subset".

Select One or More Suppressible Term Types

Source	Source Abbreviation	Term Type
Logical Observation Identifier Names and Codes, 219	LNC219	LN
Logical Observation Identifier Names and Codes, 219	LNC219	LO
Logical Observation Identifier Names and Codes, 219	LNC219	LPDN
Logical Observation Identifier Names and Codes, 219	LNC219	LPN
Logical Observation Identifier Names and Codes, 219	LNC219	LS
Logical Observation Identifier Names and Codes, 219	LNC219	LX
Logical Observation Identifier Names and Codes, 219	LNC219	OLX
Logical Observation Identifier Names and Codes, 219	LNC219	OOSN
Logical Observation Identifier Names and Codes, 219	LNC219	OSN
Logical Observation Identifier Names and Codes, 219	LNC219	SN
Logical Observation Identifier Names and Codes, 219	LNC219	SX
Logical Observation Identifier Names and Codes, 219	LNC219	XM
McMaster University Epidemiology Terms, 1992	MCM92	PT
McMaster University Epidemiology Terms, 1992	MCM92	RT
MedlinePlus Health Topics_2004_08_14, 20040814	MEDLINEPLUS_20040814	ET
MedlinePlus Health Topics_2004_08_14, 20040814	MEDLINEPLUS_20040814	PT
MedlinePlus Health Topics_2004_08_14, 20040814	MEDLINEPLUS_20040814	XM
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	CE
Medical Subject Headings, 2007_2007_05_01	MSH2007_2007_05_01	DEV

Highlighted source term types will be marked as suppressible

# File menu



# Edit menu

UMLS Metathesaurus Configuration 2007AB

File Edit Options Reset Done Help

Undo Suppressibility action Ctrl+Z

Redo Ctrl+Y

Increase Font Size Ctrl+Close Bracket

Decrease Font Size Ctrl+Open Bracket

Suppressibility

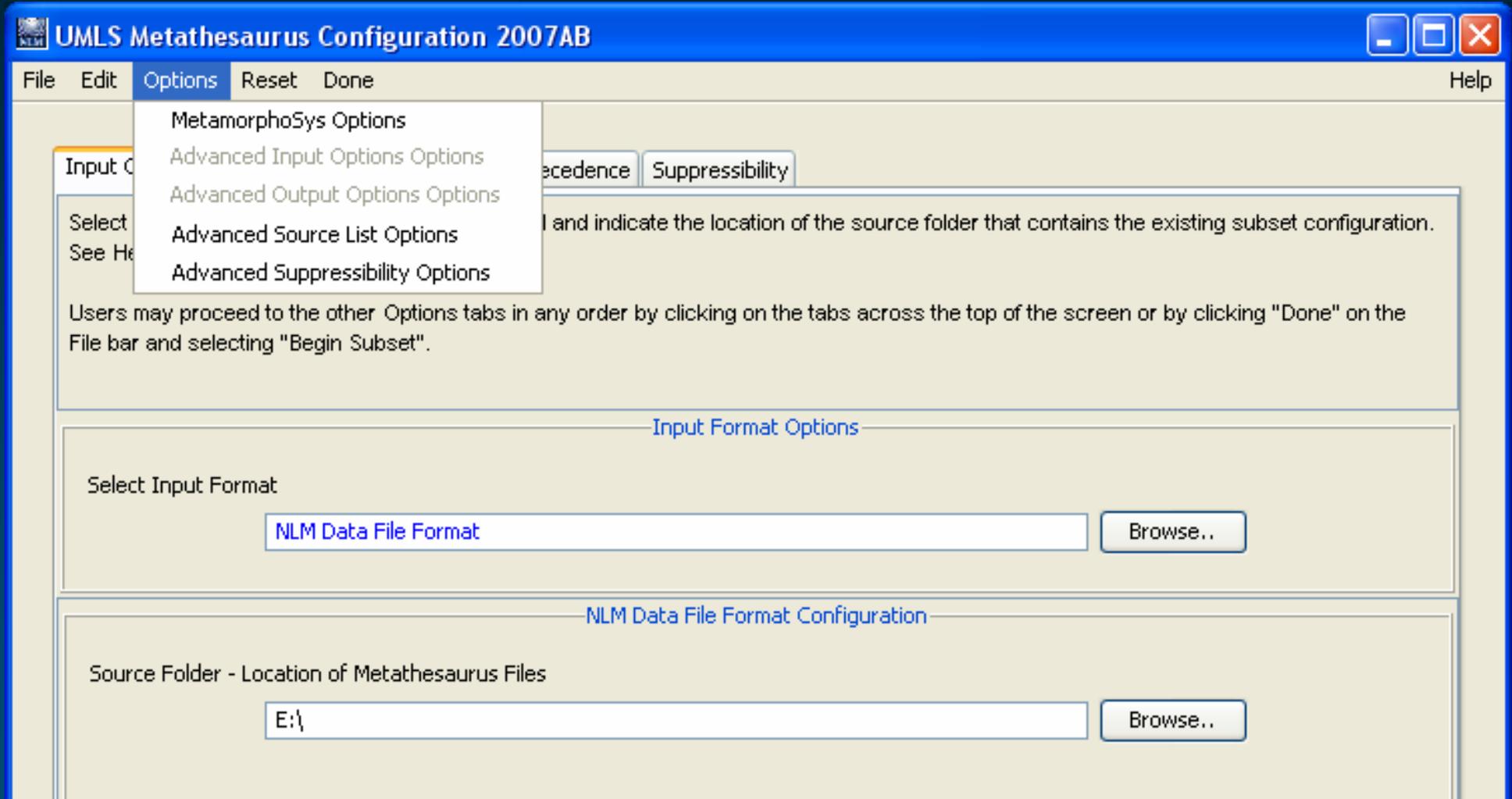
Specifies source/term type combinations to be suppressed. Users can customize the combinations. See Help for more information.

Users may proceed to the other Options tabs in any order by clicking on the tabs across the top of the screen or by clicking "Done" on the File bar and selecting "Begin Subset".

Select One or More Suppressible Term Types

Source	Source Abbreviation	Term Type
Logical Observation Identifier Names and Codes, 219	LNC219	LN
Logical Observation Identifier Names and Codes, 219	LNC219	LO
Logical Observation Identifier Names and Codes, 219	LNC219	LPDN
Logical Observation Identifier Names and Codes, 219	LNC219	LPN
Logical Observation Identifier Names and Codes, 219	LNC219	LS
Logical Observation Identifier Names and Codes, 219	LNC219	LX
Logical Observation Identifier Names and Codes, 219	LNC219	OLX
Logical Observation Identifier Names and Codes, 219	LNC219	OOSN
Logical Observation Identifier Names and Codes, 219	LNC219	OSN
Logical Observation Identifier Names and Codes, 219	LNC219	SN
Logical Observation Identifier Names and Codes, 219	LNC219	SX

# Options menu

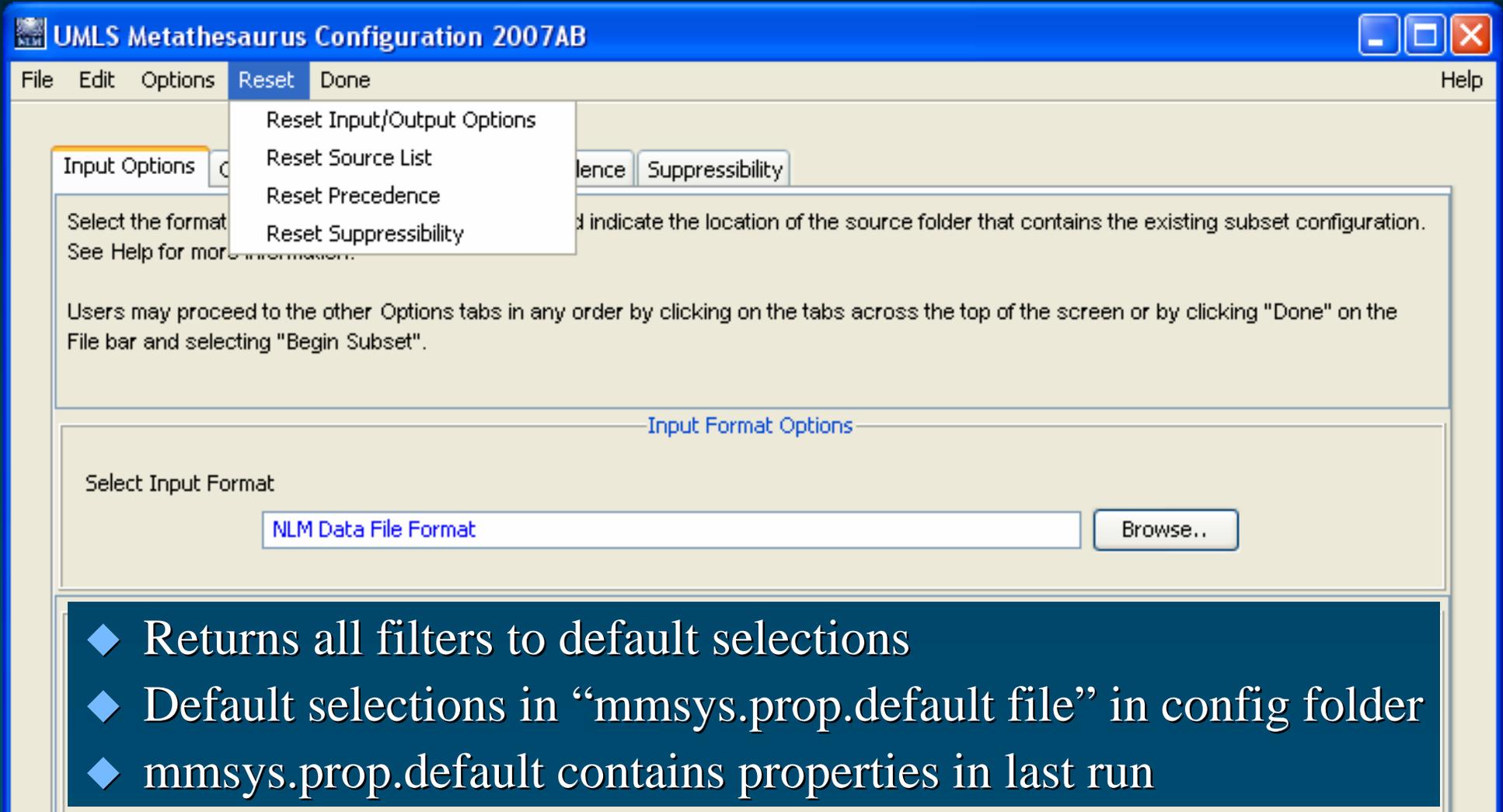


The screenshot shows the 'UMLS Metathesaurus Configuration 2007AB' application window. The 'Options' menu is open, displaying the following items:

- MetamorphoSys Options
- Advanced Input Options Options
- Advanced Output Options Options
- Advanced Source List Options
- Advanced Suppressibility Options

The main window content includes a 'File' menu with 'Reset' and 'Done' options, and a 'Help' button. The main area contains a text box with instructions: 'Users may proceed to the other Options tabs in any order by clicking on the tabs across the top of the screen or by clicking "Done" on the File bar and selecting "Begin Subset".' Below this, there are two sections: 'Input Format Options' with a 'Select Input Format' dropdown set to 'NLM Data File Format' and a 'Browse..' button; and 'NLM Data File Format Configuration' with a 'Source Folder - Location of Metathesaurus Files' dropdown set to 'E:\' and a 'Browse..' button.

# Reset menu



The screenshot shows the 'UMLS Metathesaurus Configuration 2007AB' application window. The 'Reset' menu is open, displaying the following options:

- Reset Input/Output Options
- Reset Source List
- Reset Precedence
- Reset Suppressibility

The application window includes a menu bar with 'File', 'Edit', 'Options', 'Reset', and 'Done'. The main area contains several tabs: 'Input Options', 'Precedence', and 'Suppressibility'. Below the tabs, there is a text area with instructions: 'Select the format... See Help for more information...' and 'Users may proceed to the other Options tabs in any order by clicking on the tabs across the top of the screen or by clicking "Done" on the File bar and selecting "Begin Subset".' Below this is a section titled 'Input Format Options' with a text field containing 'NLM Data File Format' and a 'Browse..' button.

- ◆ Returns all filters to default selections
- ◆ Default selections in “mmsys.prop.default file” in config folder
- ◆ mmsys.prop.default contains properties in last run

# Done – Begin Subset

UMLS Metathesaurus Configuration 2007AB

File Edit Options Reset Done Help

Begin Subset Ctrl+B

Input Options Output Options Source List Precedence Suppressibility

Select the format in which subset data was saved and indicate the location of the source folder that contains the existing subset configuration. See Help for more information.

Users may proceed to the other Options tabs in any order by clicking on the tabs across the top of the screen or by clicking "Done" on the File bar and selecting "Begin Subset".

Input Format Options

Select Input Format

NLM Data File Format Browse..

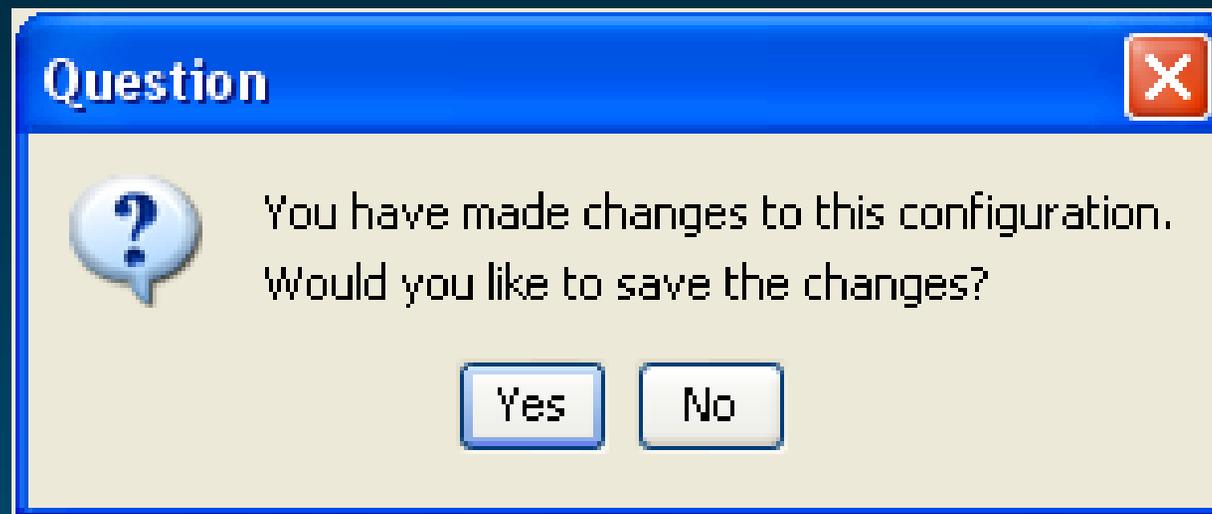
NLM Data File Format Configuration

Source Folder - Location of Metathesaurus Files

E:\ Browse..

# Save configuration for next installation

---



# How MetamorphoSys Works

---

- ◆ Removes all information from relational files in excluded vocabularies
  - atoms, strings, relationships, attributes, mappings, etc.
- ◆ Applies additional options selected by user
  - such as adding source term suppressibility or altering precedence
- ◆ Produces a full set of Metathesaurus files
  - relational files with customized data
  - reflecting other user criteria

# MetamorphoSys log

**MetamorphoSys Subset Log**

 MetamorphoSys Version:.....MMSYS-2007AB-20070621  
MetamorphoSys Build Date:.....2007\_06\_21\_18\_35\_25  
UMLS Build Date:.....2007\_06\_29\_11\_50\_02  
Release Version:.....2007AB  
Release Date:.....20070616  
Release Description:.....Base Release for Spring 2007  
Metathesaurus Source paths:.....E:\  
Subsetted Metathesaurus folder:.....D:\2007AB\META  
Configuration file used:.....D:\2007AB\ob-mesh.properties  
Start at:.....Fri Jul 27 16:28:35 EDT 2007  
Initialize CUI List completed:.....Fri Jul 27 16:33:00 EDT 2007  
Subset Metathesaurus completed:.....Fri Jul 27 17:28:55 EDT 2007  
Subset Index Files completed:.....Fri Jul 27 17:36:34 EDT 2007  
Subset Release Metadata completed:....Fri Jul 27 17:36:56 EDT 2007  
Finished at:.....Fri Jul 27 17:39:02 EDT 2007  
Concepts in source:.....1436586  
Concepts in subset:.....278431  
Time elapsed:.....01:10:26

Details about MetamorphoSys Input Handler

# MetamorphoSys log

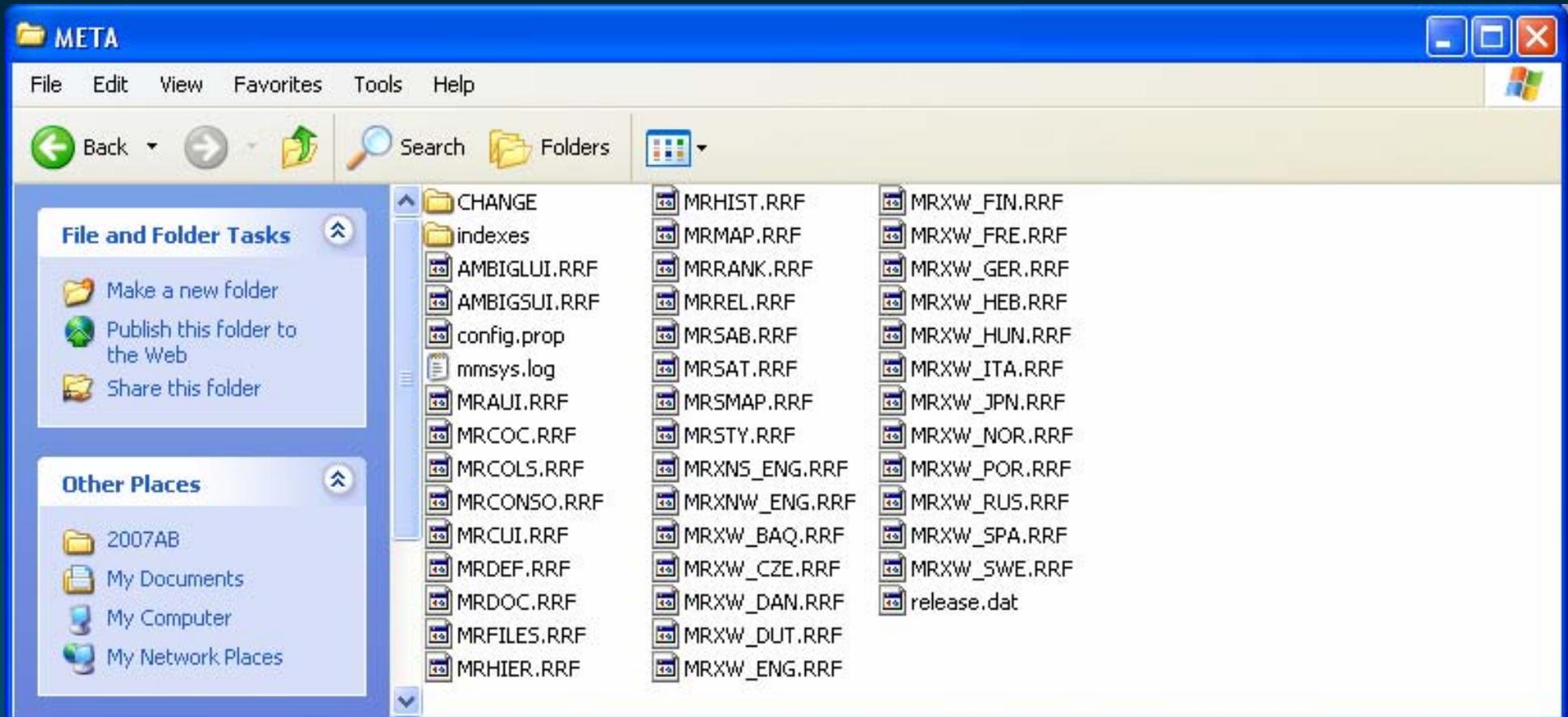
```
mmsys.log - Notepad
File Edit Format View Help
MetamorphoSys Version:.....MMSYS-2007AB-20070621
MetamorphoSys Build Date:.....2007_06_21_18_35_25
UMLS Build Date:.....2007_06_29_11_50_02
Release Version:.....2007AB
Release Date:.....20070616
Release Description:.....Base Release for Spring 2007
Metathesaurus Source paths:.....E:\
Subsetted Metathesaurus folder:.....D:\2007AB\META
Configuration file used:.....D:\2007AB\ob-mesh.properties
Start at:.....Fri Jul 27 16:28:35 EDT 2007
Initialize CUI List completed:.....Fri Jul 27 16:33:00 EDT 2007
Subset Metathesaurus completed:.....Fri Jul 27 17:28:55 EDT 2007
Subset Index Files completed:.....Fri Jul 27 17:36:34 EDT 2007
Subset Release Metadata completed:....Fri Jul 27 17:36:56 EDT 2007
Finished at:.....Fri Jul 27 17:39:02 EDT 2007
Concepts in source:.....1436586
Concepts in subset:.....278431
Time elapsed:.....01:10:26

Details about MetamorphoSys Input Handler
-----
MetamorphoSys Input: NLM Data File Format

Details about MetamorphoSys Output Handler
-----
Metathesaurus Output: Rich Release Format
Long fields were not truncated.
Source Abbreviations were written out with a versionless (root)
representation.
Fields containing UTF-8 characters were not removed.
UTF-8 BOM characters were not added to output files.
Md5s were not calculated.
Concepts containing only MTH atoms were not removed.

Excluded Sources
  AI/RHEUM, 1993
```

# Output directory contents



## Part II

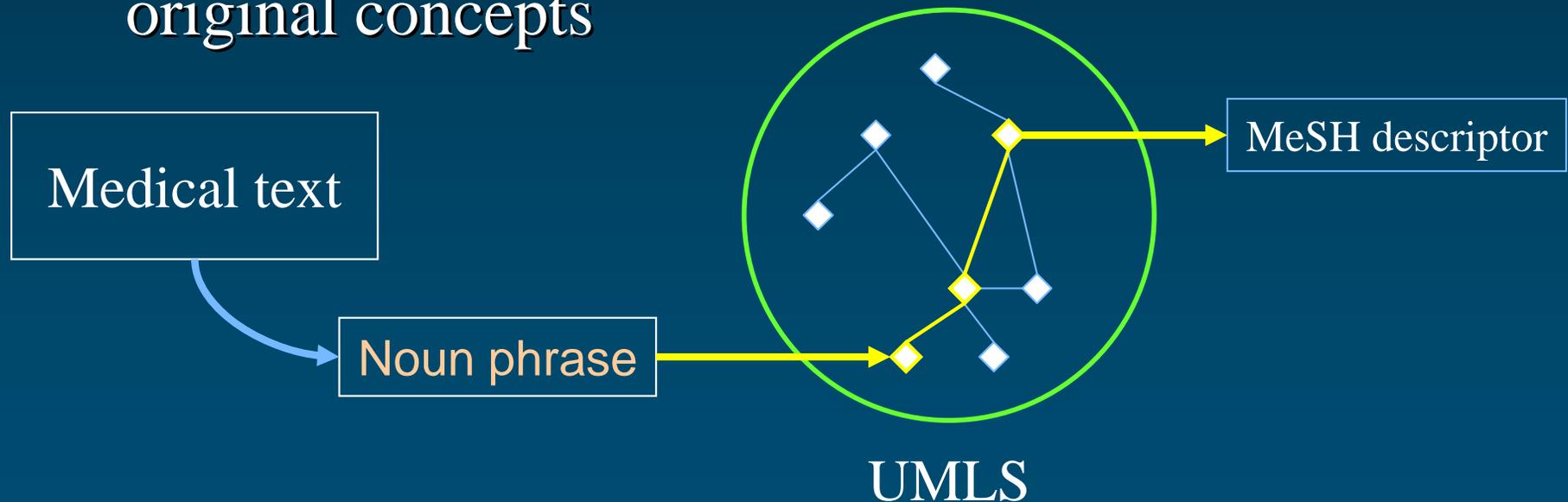
### How to use the UMLS?

*(4) A UMLS-based algorithm*

# Indexing Initiative

[Aronson & al., *AMIA*, 2000]

- ◆ 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



# Restrict to MeSH

[Bodenreider & al., *AMIA*, 1998]

- ◆ Based on the principle of semantic locality
- ◆ Use different components of the UMLS
- ◆ 4 techniques of increasing aggressiveness
  - Use Synonymy **MRCONSO**
  - Use Associated expressions (ATXs) **MRATX + MRREL**
  - Explore the Ancestors **MRREL + SN**
  - Explore the Other related concepts **MRREL + SN**

# Restrict to MeSH Synonymy

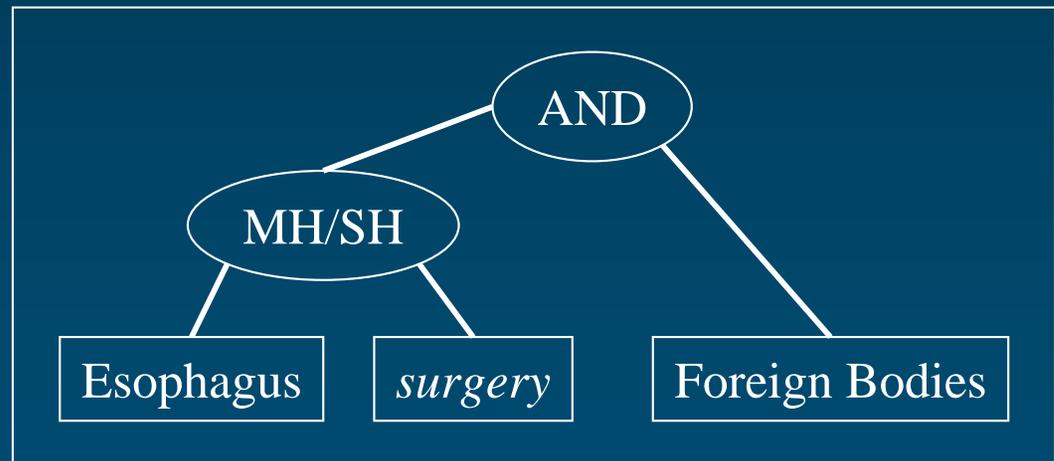
---

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

# Restrict to MeSH Assoc. expressions

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

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 (**MRCNSO**)

# Restrict to MeSH Example

Vein of neck, NOS

There is a MeSH term in the synonyms of SC

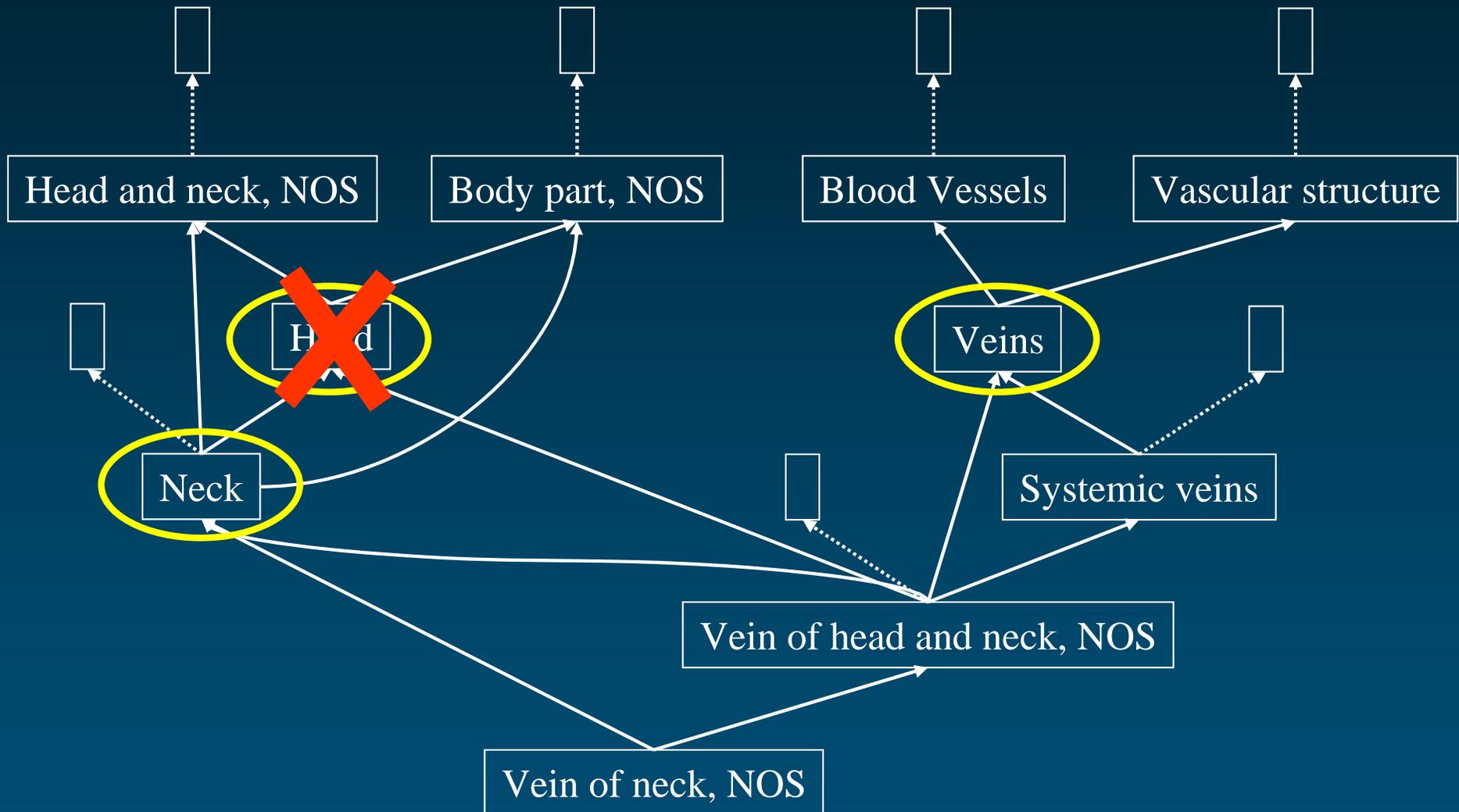
SC is described by a combination of MeSH terms (ATX)

The ancestors of SC contain MeSH terms

MeSH terms from non-hierarchically related concepts

Vein + Neck

# Restrict to MeSH Example



# Restrict to MeSH Quantitative results

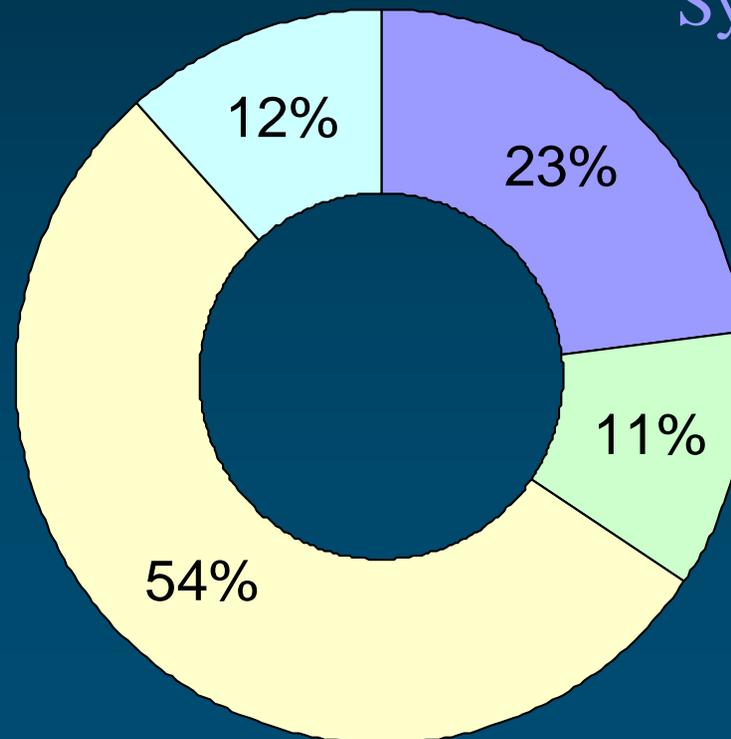
- ◆ 86% of UMLS concepts mapped to MeSH (2007)

Other related concepts

Synonymy

Graph of  
ancestors

Built-in  
mappings



# Restrict to MeSH Qualitative results

---

## ◆ Qualitative evaluation

- 1,036 concepts extracted from 200 MEDLINE citations
- manual review of every mapping or failure

## ◆ 61% Relevant

- Subtotal Gastrectomy → Gastrectomy
- Encephalopathy, NOS → Brain Diseases

## ◆ 28% More or less relevant

- Vitamin A measurement → Laboratory Procedure
- Swelling, NOS → Symptoms

## ◆ 11% Non relevant



## Part II

### How to use the UMLS?

#### *(5) Benefits and Limitations*

# Benefits

# UMLS compared to individual vocabularies

---

- ◆ Broader scope
- ◆ Extended coverage
- ◆ Finer granularity
- ◆ Unique identifier
- ◆ Synonymous terms clustered into concepts
- ◆ Additional synonyms
- ◆ Additional hierarchical relationships
- ◆ Semantic categorization

# Direct benefits

---

- ◆ Concept categorization
- ◆ Information retrieval
  - Synonyms
  - Cross-language features
- ◆ Information extraction
  - MetaMap
  - Normalization
- ◆ Information visualization
  - Knowledge Source Server
  - Semantic Navigator
  - RRF browser

# UMLS as an enabling resource

---

## ◆ Examples

- Mapping across vocabularies
- Semantics of statistical associations
- Redundancy in hierarchical relations

# Limitations

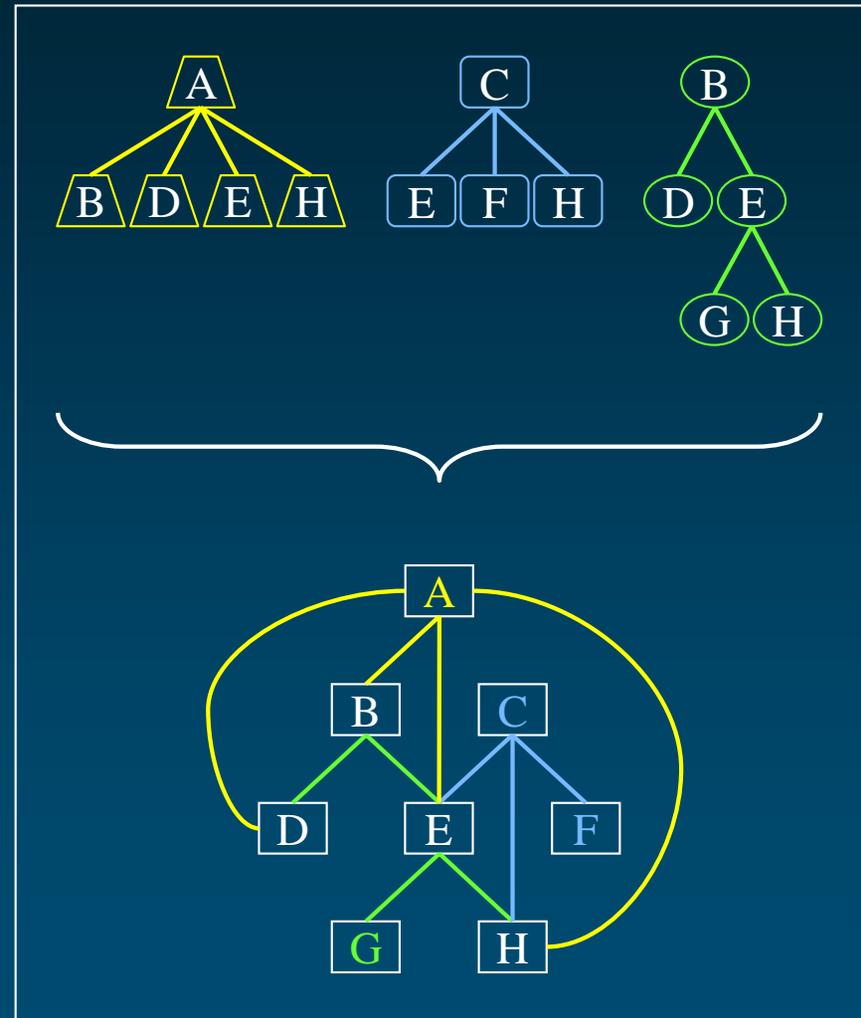
# Limitations

[Cimino, *JAMIA*, 1998]

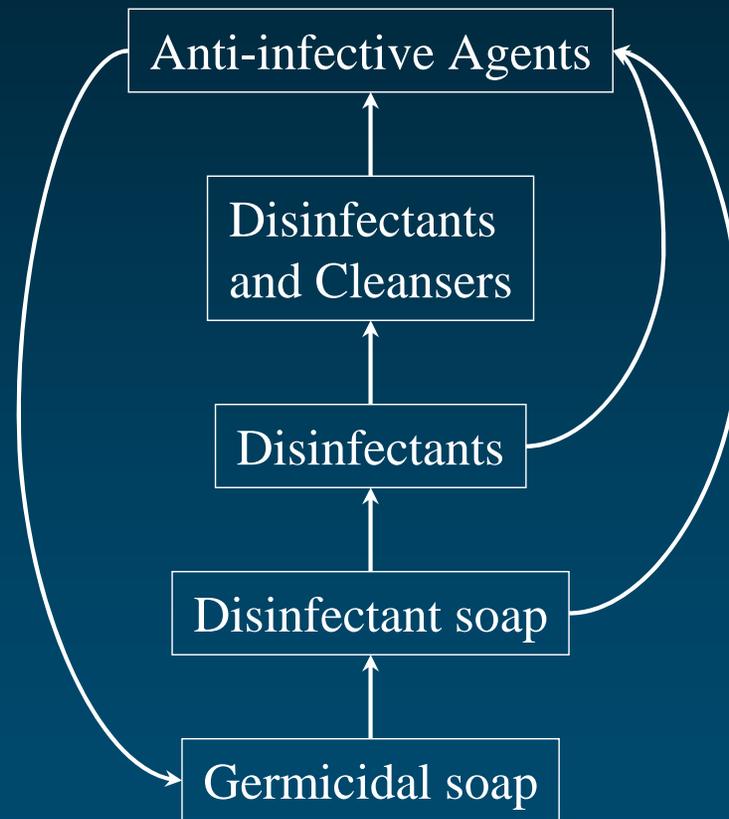
- ◆ Structural inconsistency
  - Cycles in the graph of hierarchical relations
- ◆ Semantic inconsistency
  - Between Metathesaurus and Semantic Network
- ◆ Underspecified relationships
- ◆ Missing relations
  - Synonymy
  - Hierarchical relations (missing or underspecified)

# Structural inconsistency From trees to graph

- ◆ Multiple tree structures combined into a graph structure
- ◆ Expected: Directed acyclic graph (DAG)



# Structural inconsistency Cycles in the UMLS graph



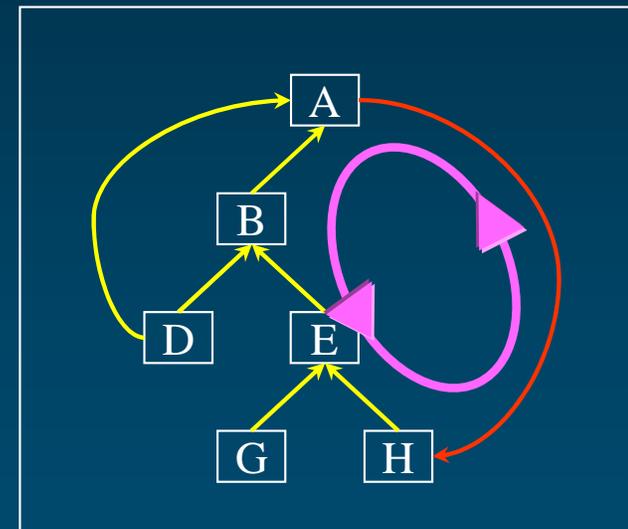
# Structural inconsistency Issues

## ◆ Theoretical

- Violate the antisymmetry property of partial ordering relations

## ◆ Practical

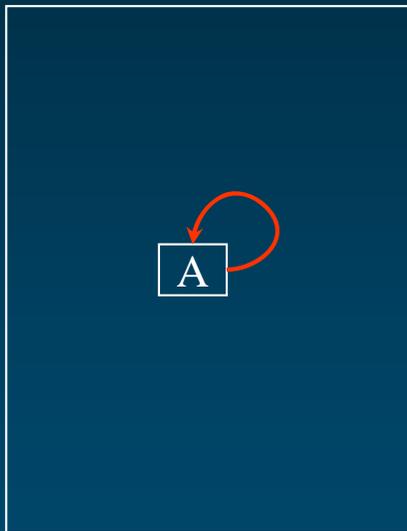
- Loops in graph traversal
- Impossible to perform transitive reduction



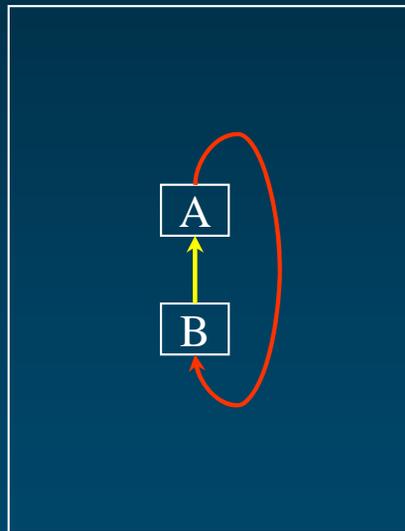
[Bodenreider, AMIA 2001]

# Acyclicity

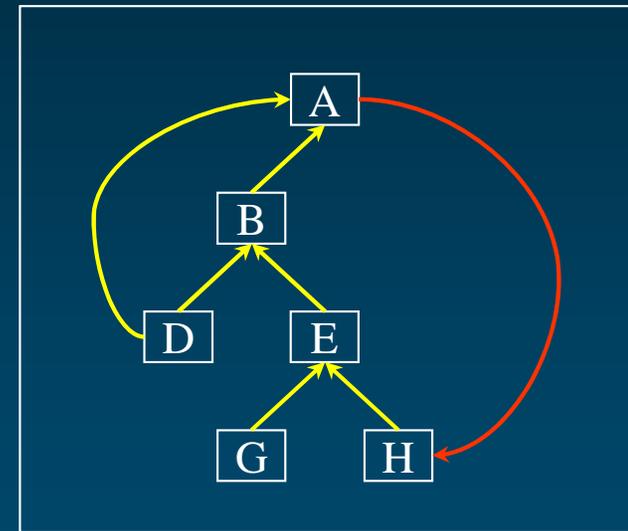
“back edge” from a child concept to a parent concept



Reflexive  
13,000

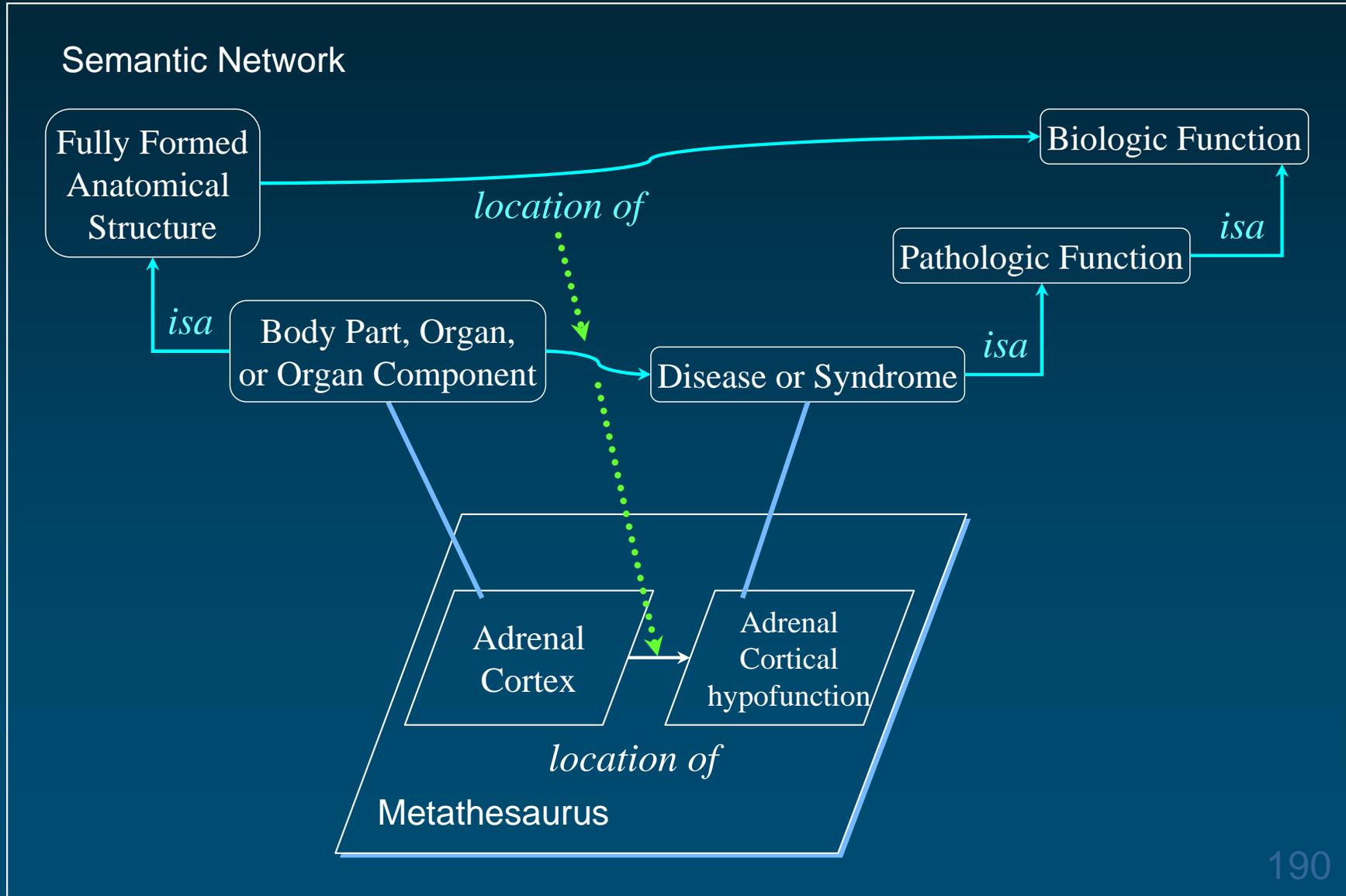


Direct  
1800



Indirect  
120

# Semantic inconsistency A two-level structure



# Semantic inconsistency A limited study

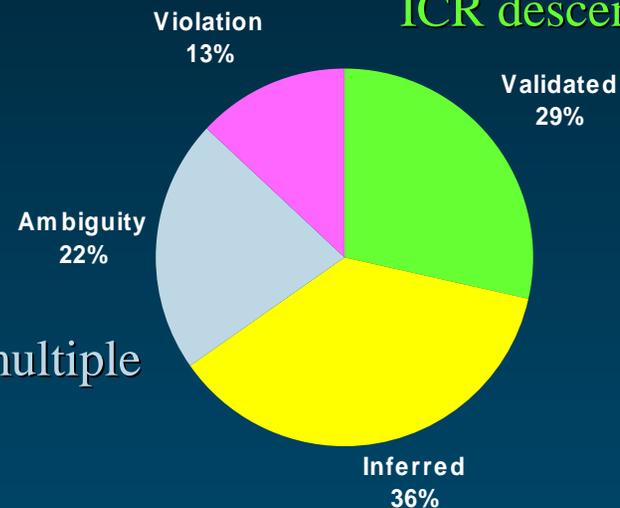
## ◆ 6894 interconcept relationships

- among the 3764 concepts in the semantic neighborhood of “Heart”

ICR not specified and SNR compatible and multiple

ICR and SNR not compatible

ICR = SNR or ICR descendant of SNR



ICR not specified and SNR compatible and unique

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

# Semantic inconsistency Issues

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- ◆ The UMLS integrates what terminologies represent
- ◆ Hierarchies in source vocabularies
  - Often task-driven rather than based on principles
  - Usually suitable for information retrieval
  - Not necessarily suitable for reasoning
- ◆ No automatic correction possible
  - Wrong categorization
  - Wrong inter-concept relationship
  - [Wrong semantic network relationship]

# Underspecified relationships

- ◆ Relationship “attribute” not always present
- ◆ Relations used to create hierarchies vs. hierarchical relations

[Environment and Public Health \[G03\]](#)

[Public Health \[G03.850\]](#)

▶ [Accidents \[G03.850.110\]](#)

[Accident Prevention \[G03.850.110.060\]](#) +

[Accidental Falls \[G03.850.110.085\]](#)

[Accidents, Aviation \[G03.850.110.185\]](#)

[Accidents, Home \[G03.850.110.205\]](#)

[Accidents, Occupational \[G03.850.110.250\]](#) +

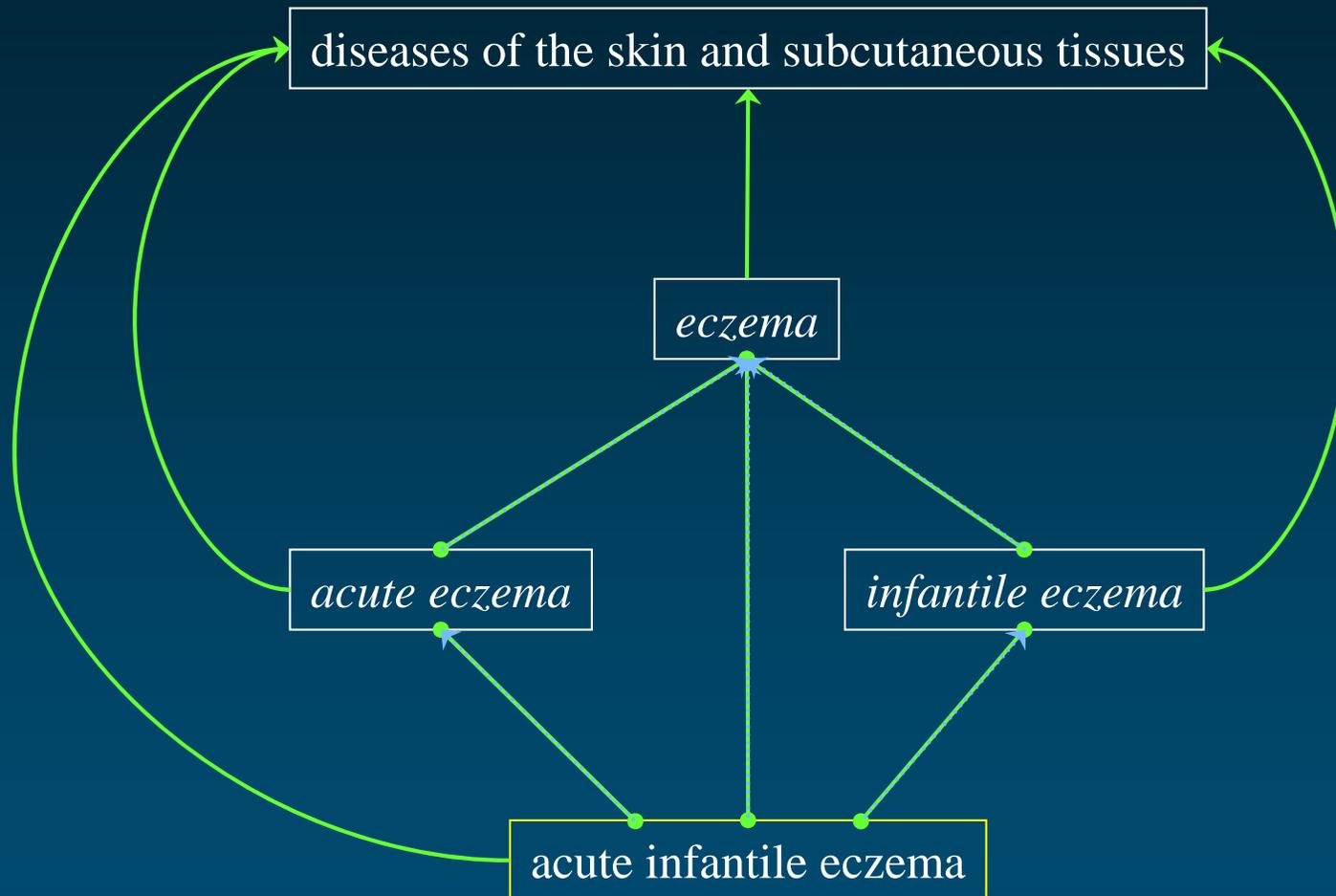
[Accidents, Radiation \[G03.850.110.285\]](#)

[Accidents, Traffic \[G03.850.110.320\]](#)

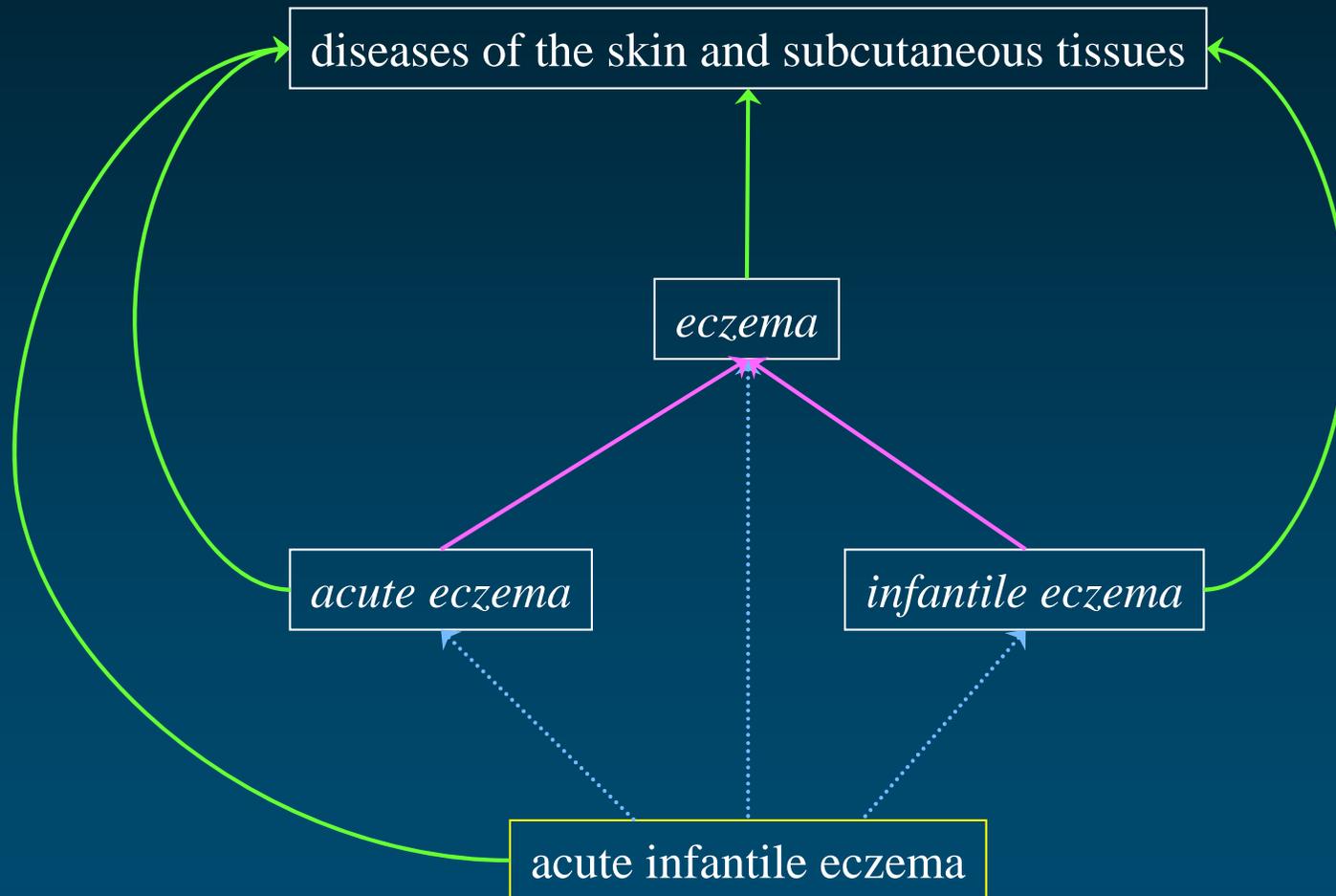
[Drowning \[G03.850.110.500\]](#) +



# Missing relations Example



# Missing relations Example



# Missing relations A limited study

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- ◆ 28,851 pairs of terms
  - Original SNOMED term
  - Demodified term (found in UMLS)
- ◆ Corresponding relationship in the Metathesaurus
  - Hierarchical in 50% of the cases
  - « Sibling » in 25% of the cases
  - Missing in 25% of the cases

[Bodenreider & al., *TIA*, 2001]

# Compensation mechanisms

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## ◆ Examples

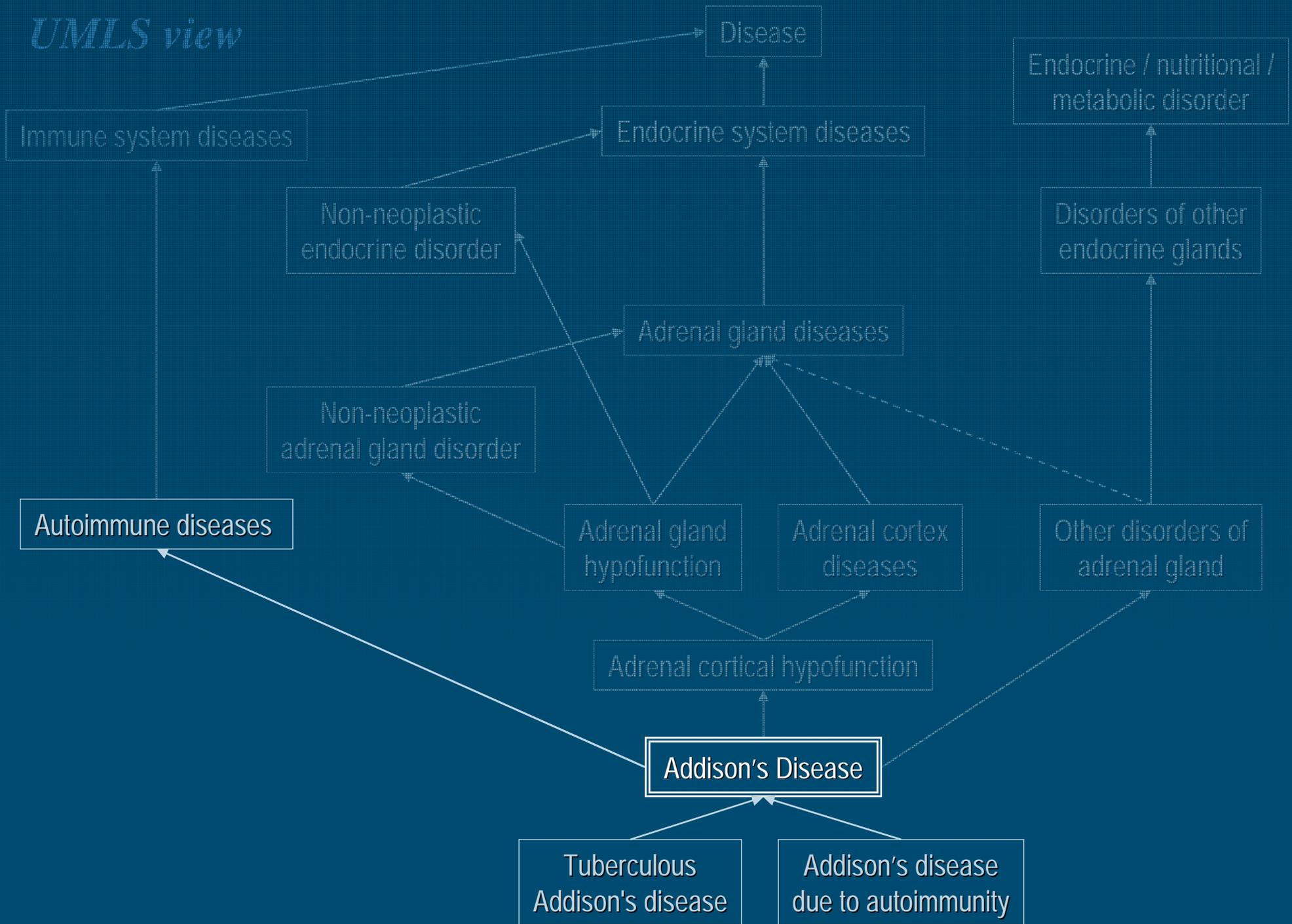
- Removing cycles from hierarchical relations
  - Using redundancy (number of sources asserting the relation)
  - Using terminological knowledge (e.g., NEC)
- Lexically-suggested hyponymic relations
  - Properties of adjectival modification

# More limitations

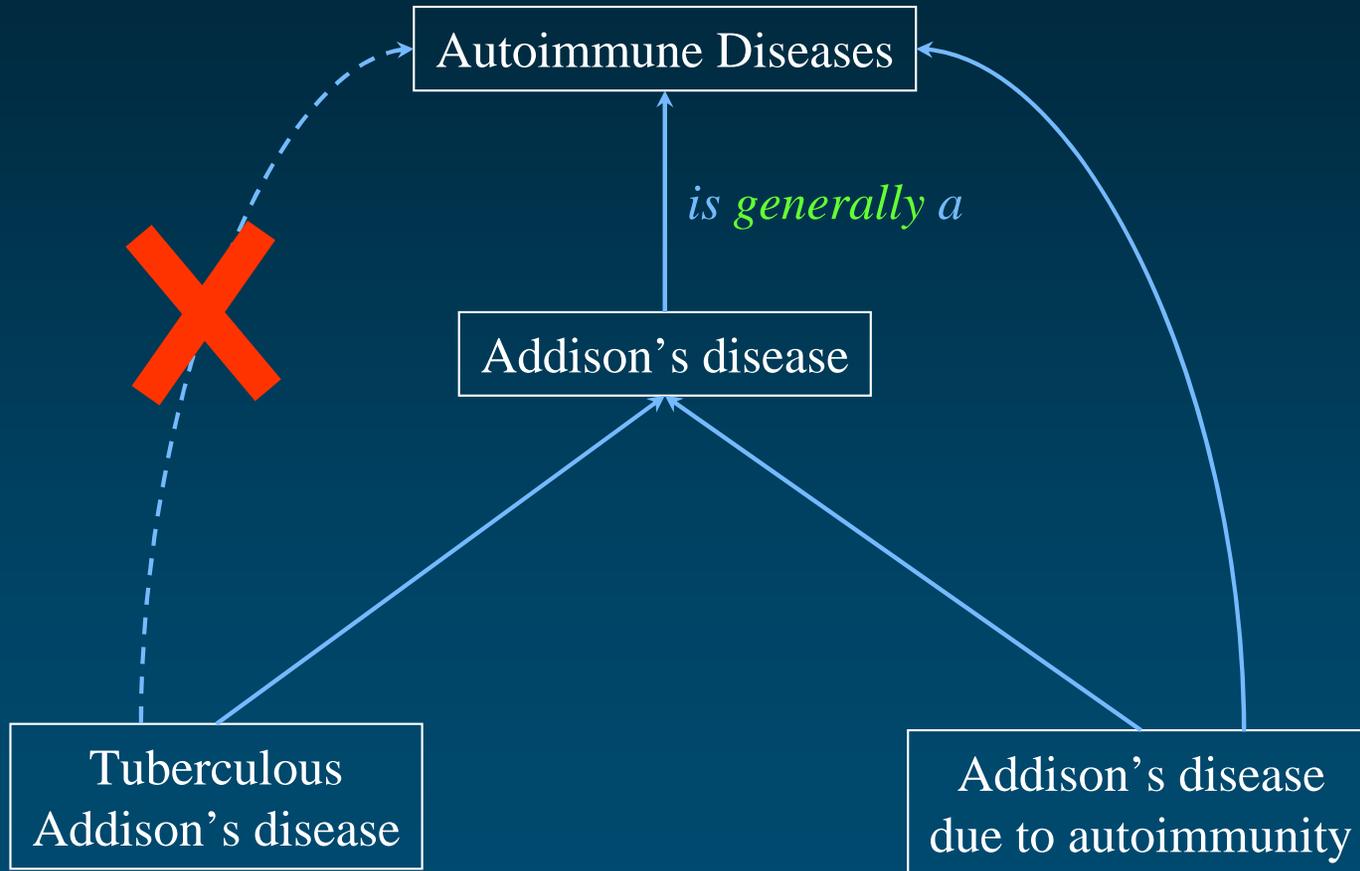
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- ◆ Semantics of hierarchical relations
- ◆ Some missing / wrong relations are hard to detect
- ◆ Some relations are present but hard to find

*UMLS view*

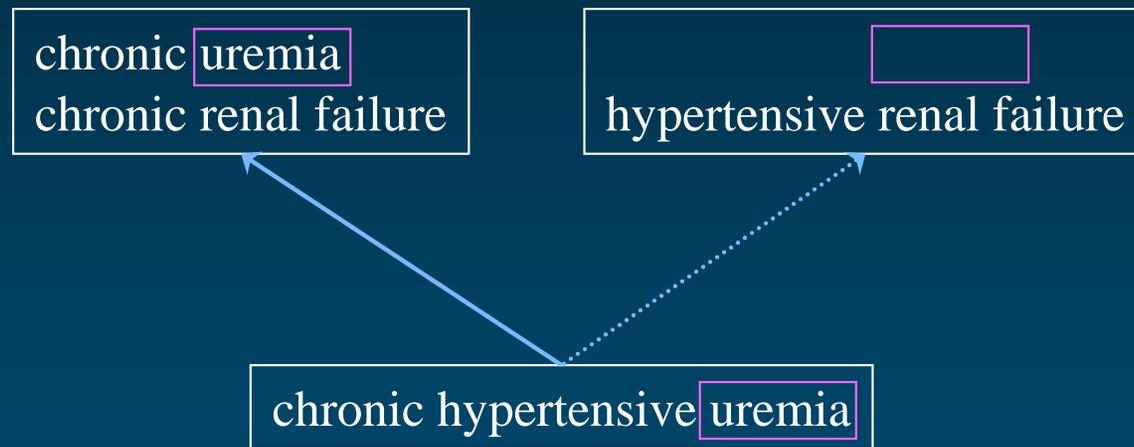


# Semantics of hierarchical links

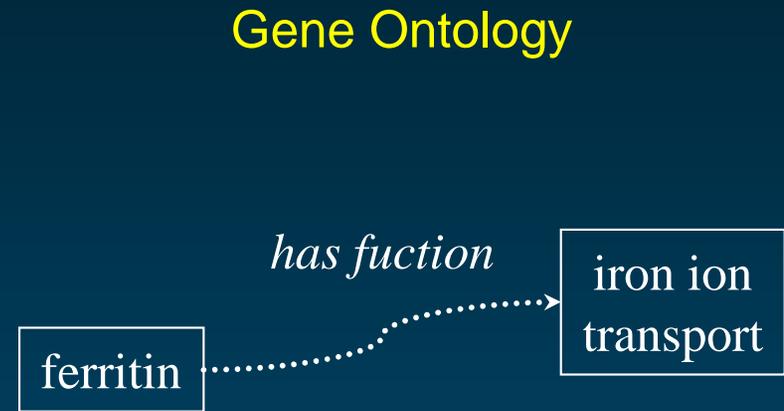
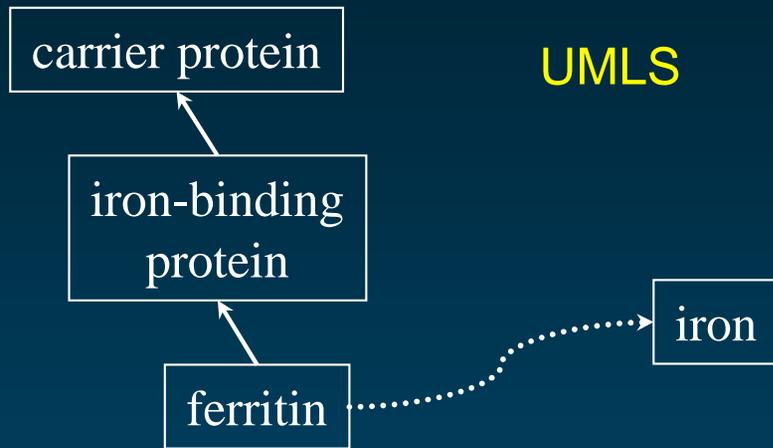


# Relations Missing and difficult to detect

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# Relations Existing but difficult to find



ferritin *is a* iron transporter

reified "transport" relationship

ferritin *transports* iron

"transport" relationship

# How to address these limitations?

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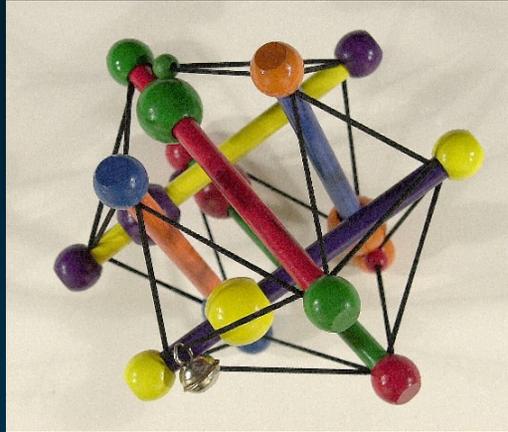
- ◆ Description logics
- ◆ Natural Language Processing  
(semantic interpretation of the terms)
- ◆ Comparing knowledge sources  
(alignment, inference)

# Summary

# UMLS Summary

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- ◆ UMLS = 3 Knowledge Sources
  - Metathesaurus
  - Semantic Network
  - SPECIALIST Lexicon and Lexical Tools
- ◆ MetamorphoSys
  - installs
  - customizes
- ◆ UMLSKS
  - remote access
  - resources and documentation



# Medical Ontology Research

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# Bibliography

# References: UMLS home page

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## ◆ UMLS home page

- [http:// www.nlm.nih.gov/research/umls/](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>

# References

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## ◆ Short presentation

- Bodenreider, O. (2004) The Unified Medical Language System (UMLS): integrating biomedical terminology. *Nucleic Acids Res*, 32(Database issue), D267-70.

## ◆ 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

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## ◆ Technical papers

- McCray, A. T., & Nelson, S. J. (1995). The representation of meaning in the UMLS. *Methods Inf Med*, 34(1-2), 193-201.

## ◆ Comprehensive bibliography 1986-96

<http://www.nlm.nih.gov/pubs/cbm/umlscbm.html>

# Documentation and Support

# UMLS documentation and support

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## ◆ UMLS homepage

- links to various UMLS resources
- <http://www.nlm.nih.gov/research/umls/>

## ◆ UMLSKS homepage

- links to the User's and Developer's guides
- <http://umlsks.nlm.nih.gov/>

## ◆ UMLS mailing list

- UMLSUSERS-L@LIST.NIH.GOV

## ◆ Email address for support

- [custserv@nlm.nih.gov](mailto:custserv@nlm.nih.gov)



# Appendix

UMLS files in  
Rich Release Format

# MRCONSO (sample rows 1..5)

(2004AB)

	1	2	3	4	5	6	7	8	9	10	11
	CUI	LAT	TS	LUI	STT	SUI	ISPRF	AUI	SAUI	SCUI	SDUI
1	C0001403	ENG	P	L0001403	PF	S0354372	Y	A4367951			
2	C0001403	ENG	P	L0001403	PF	S0354372	N	A2922421	485624014	363732003	
3	C0001403	ENG	P	L0001403	VC	S0010794	Y	A0019740		M0000346	D000224
4	C0001403	ENG	S	L0494851	PF	S2164152	N	A2018589			
5	C0001403	FRE	P	L3246333	PF	S3773545	Y	A3996251			D000224

	12	13	14	15	16	17	18
	SAB	TTY	CODE	STR	SRL	SUPPRESS	CVF
1	MTH	PN	NOCODE	Addison's disease	0	N	
2	SNOMEDCT	PT	363732003	Addison's disease	4	N	
3	MSH	MH	D000224	Addison's Disease	0	N	
4	MDR	LT	10052381	Primary adrenal insufficiency	3	N	
5	MSHFRE	MH	D000224	Addison, maladie	3	N	



# MRCONSO (sample rows 6..10)

(2004AB)

	1	2	3	4	5	6	7	8	9	10	11
	CUI	LAT	TS	LUI	STT	SUI	ISPPREF	AUI	SAUI	SCUI	SDUI
6	C0001403	FRE	S	L1272481	PF	S1514427	Y	A1464383			
7	C0001403	GER	P	L1229627	PF	S1471573	Y	A4030156			D000224
8	C0001403	GER	S	L1239271	PF	S1481217	Y	A4034094			D000224
9	C0001403	JPN	P	L3437833	PF	S3965327	Y	A4264008			D000224
10	C0001403	JPN	S	L3465347	PF	S3992841	Y	A4291522			D000224

	12	13	14	15	16	17	18
	SAB	TTY	CODE	STR	SRL	SUPPRESS	CVF
6	WHOFRE	IT	0410	MALADIE D'ADDISON	2	N	
7	MSHGER	MH	D000224	Addison-Krankheit	3	N	
8	MSHGER	SY	D000224	Bronzehautkrankheit	3	N	
9	MSHJPN	MH	D000224	Addison病	3	N	
10	MSHPJN	SY	D000224	副腎性黒皮症	3	N	



# MRCONSO (sample rows 11-13)

(2004AB)

	1	2	3	4	5	6	7	8	9	10	11
	CUI	LAT	TS	LUI	STT	SUI	ISPREF	AUI	SAUI	SCUI	SDUI
11	C0001403	POR	P	L3302998	PF	S3831123	N	A6382080			
12	C0001403	RUS	P	L3336992	PF	S3864473	Y	A4157629			
13	C0001403	SPA	P	L1226877	PF	S1468823	Y	A1419475			



	12	13	14	15	16	17	18
	SAB	TTY	CODE	STR	SRL	SUPPRESS	CVF
11	MDRPOR	LT	1001130	Doença de Addison	3	N	
12	MSHRUS	MH	D000224	АДДИСОНОВА БОЛЕЗНЬ	3	N	
13	WHOSPA	IT	0410	ADDISON, ENFERMEDAD	3	N	



# MRHIER (sample rows)

(2004AB)

	1	2	3	4	5	6
	CUI	AUI	CXN	PAUI	SAB	RELA
1	C0001403	A0019740	1	A0020270	MSH	
2	C0001403	A0019740	2	A0028022	MSH	
3	C0001403	A0019743	3	A1988358	PSY	member_of_cluster
4	C0001403	A2922421	1	A3307650	SNOMEDCT	isa
5	C0001403	A2922421	2	A3307650	SNOMEDCT	isa

	7	8	9
	PTR	HCD	CVF
1	A0434168.A2367943.A2366890.A0135391.A0054194.A0020267.A0020270	C19.053.264.263	
2	A0434168.A2367943.A2366890.A0135391.A0072566.A0028022	C20.111.163	
3	A0449751.A1988279.A1988358		
4	A3684559.A3886745.A2880798.A3398606.A3399335.A3398961.A2872359. A2872360.A3307650		
5	A3684559.A3886745.A2880798.A3398606.A3399335.A3398961.A2872359. A2933400.A2989549.A3307650		



# MRREL (sample rows)

(2004AB)

	1	2	3	4	5	6	
	CUI1	AUI1	STYPE1	REL	CUI2	AUI2	STYPE2
1	C0001403		CUI	RB	C0001621		CUI
2	C0001403	A0019738	AUI	SY	C0001403	A0049628	AUI
3	C0001403	A2922421	SCUI	CHD	C0085859	A2977940	SCUI
4	C0001403	A6326321	SCUI	RO	C0688490	A6339383	SCUI
5	C0001403	A0019743	AUI	PAR	C0935495	A1988358	AUI

	7	8	9	10	11	12	13	14	15
	RELA	RUI	SRUI	SAB	SL	RG	DIR	SUPPRESS	CVF
1		R02837989		MTH			N	N	
2		R18849683		MSH	MSH			N	
3	isa	R19859511	1658795027	SNOMEDCT	SNOMEDCT	0	Y	N	
4	may_treat	R27600039		NDFRT	NDFRT			N	
5	has_member	R08110401		PSY	PSY			N	



# MRDEF

(2004AB)

CUI	AUI	ATUI	SATUI	SAB	DEF	SUPPRESS	CVF
C0001403	A0019740	AT15061584		MSH	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.	N	



# MRSAT (sample rows)

(2004AB)

	1	2	3	4	5	
	CUI	LUI	SUI	METAUI	STYPE	CODE
1	C0001403	L0001403	S0010792	A0019738	AUI	D000224
2	C0001403	L0001403	S0010794	A6326321	SCUI	C712
3	C0001403	L0001403	S0354372	A2922421	SAUI	363732003
4	C0001403			R15742591	SRUI	
5	C0001403				CUI	

	7	8	9	10	11	14	15
	ATUI	SATUI	ATN	SAB	ATV	SUPPRESS	CVF
1	AT15321482		DID	MSH	D000224	N	
2	AT33411754		MESH_UI	NDFRT	D000224	N	
3	AT24166602		DESCRIPTION STATUS	SNOMEDCT	0	N	
4	AT27438950		REFINABILITY	SNOMEDCT	0	N	
5	AT02925340		ST	MTH	R	N	



# MRSTY

(2004AB)

---

CUI	TUI	STN	STY	ATUI	CVF
C0001403	T047	B2.2.1.2.1	Disease or Syndrome	AT17683850	



# MRHIST (sample rows)

(2004AB)

	1	2	3	4	5
	CUI	SOURCEUI	SAB	SVER	CHANGETYPE
1	C0001403	1198962018	SNOMEDCT	20020731	0
2	C0001403	1212124016	SNOMEDCT	20020731	0
3	C0001403	1490869013	SNOMEDCT	20030131	0
4	C0001403	363732003	SNOMEDCT	20020129	0
5	C0001403	373662000	SNOMEDCT	20020731	0



	6	7	8	9
	CHANGEKEY	CHANGEVAL	REASON	CVF
1	DESCRIPTIONSTATUS	0		
2	DESCRIPTIONSTATUS	0		
3	DESCRIPTIONSTATUS	0		
4	CONCEPTSTATUS	0		
5	CONCEPTSTATUS	0		

