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# The Unified Medical Language System
## *What is it and how to use it?*


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# Outline

- 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

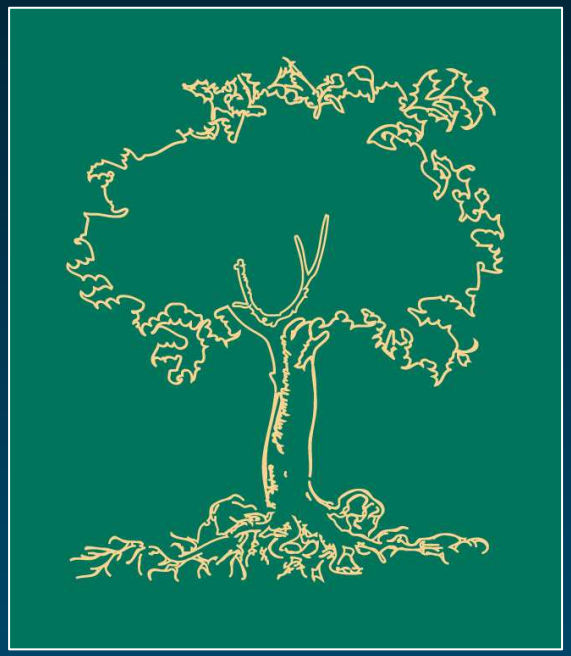
- Part I: *What is the UMLS?*
  - Introduction
  - Overview through an example
  - The three UMLS Knowledge Sources
    - UMLS Metathesaurus
    - UMLS Semantic Network
    - SPECIALIST Lexicon and lexical tools

# Part I
# What is the UMLS?

## *(1) Introduction*

# What does UMLS stand for?

- Unified
- Medical
- Language
- System



UMLS®
Unified Medical Language System®
UMLS Metathesaurus®

# Motivation

- Started in 1986
- National Library of Medicine
- "Long-term R&D project"
- Complementary to IAIMS

(Integrated Academic Information Management Systems)

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

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

NLM

# The UMLS in practice

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

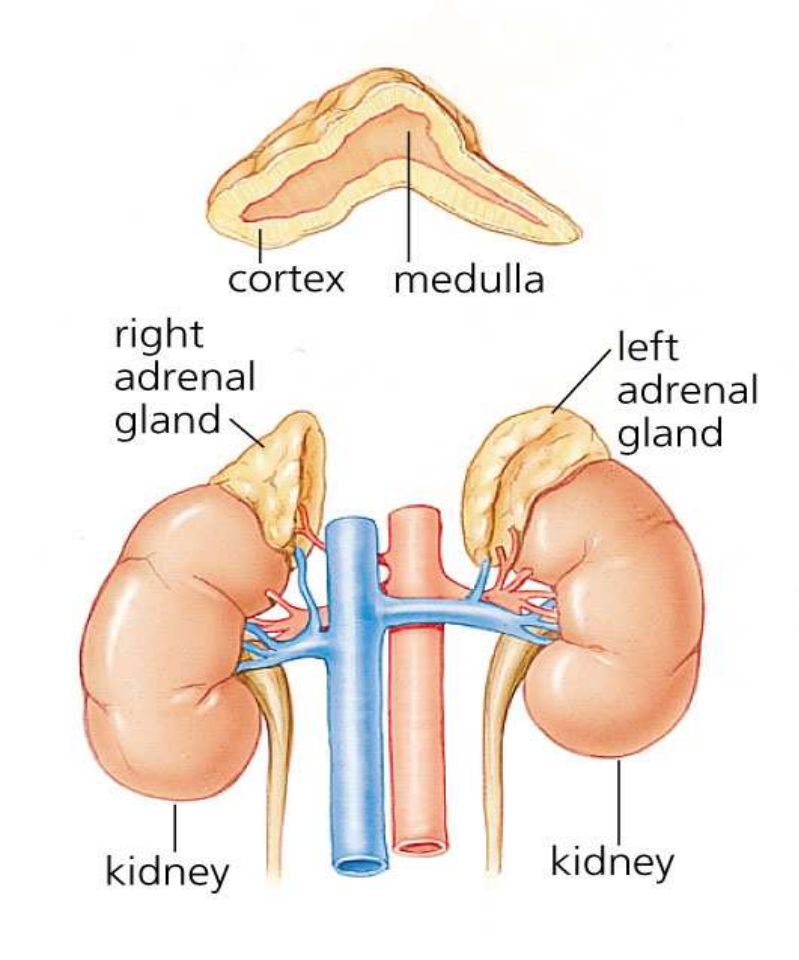
The UMLS is *not* an end-user application

# Part I
# What is the UMLS?

## *(2) Overview through an example*

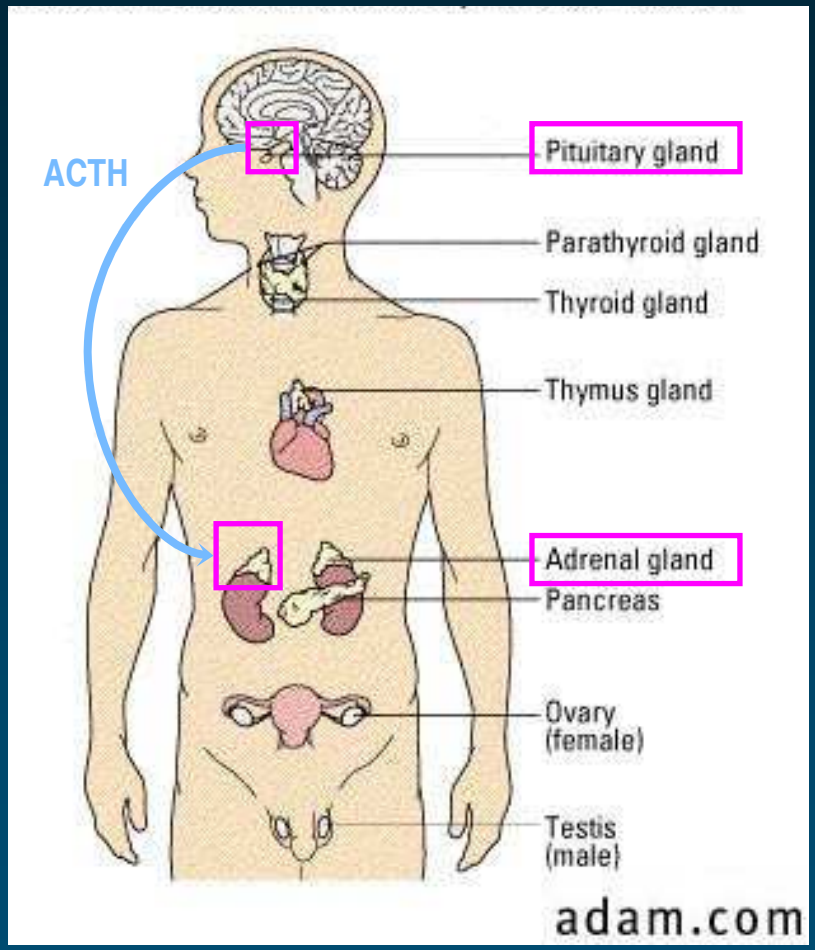
# Addison's disease

- 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

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

# AD in medical vocabularies

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

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# Organize terms

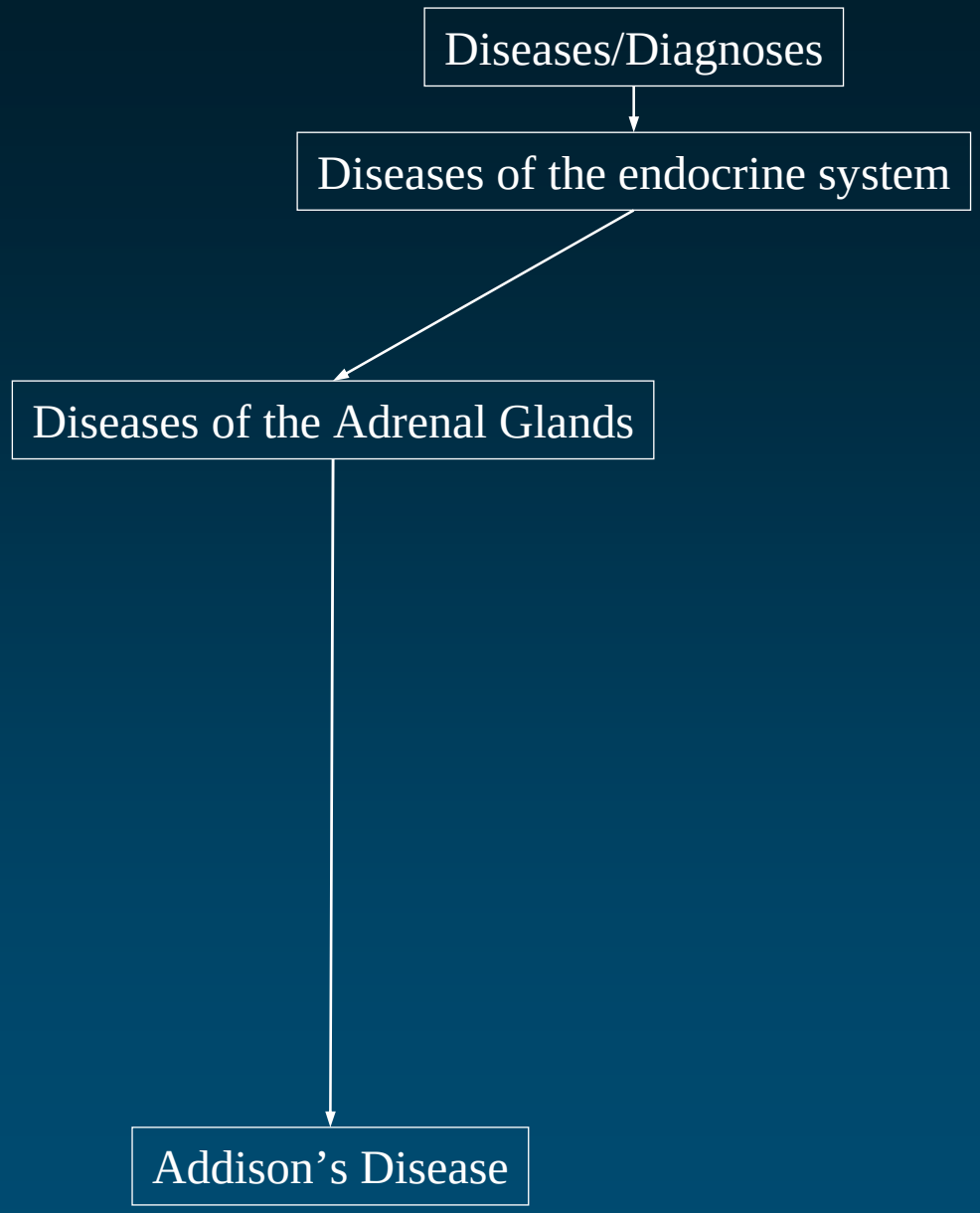
- Synonymous terms clustered into a concept
- Preferred term
- Unique identifier (CUI)

| | | |
|---|---|---|
| Adrenal gland diseases | MeSH | D000307 |
| Adrenal disorder | AOD | 0000005418 |
| Disorder of adrenal gland | Read | C15z. |
| Diseases of the adrenal glands | SNOMED | DB-70000 |

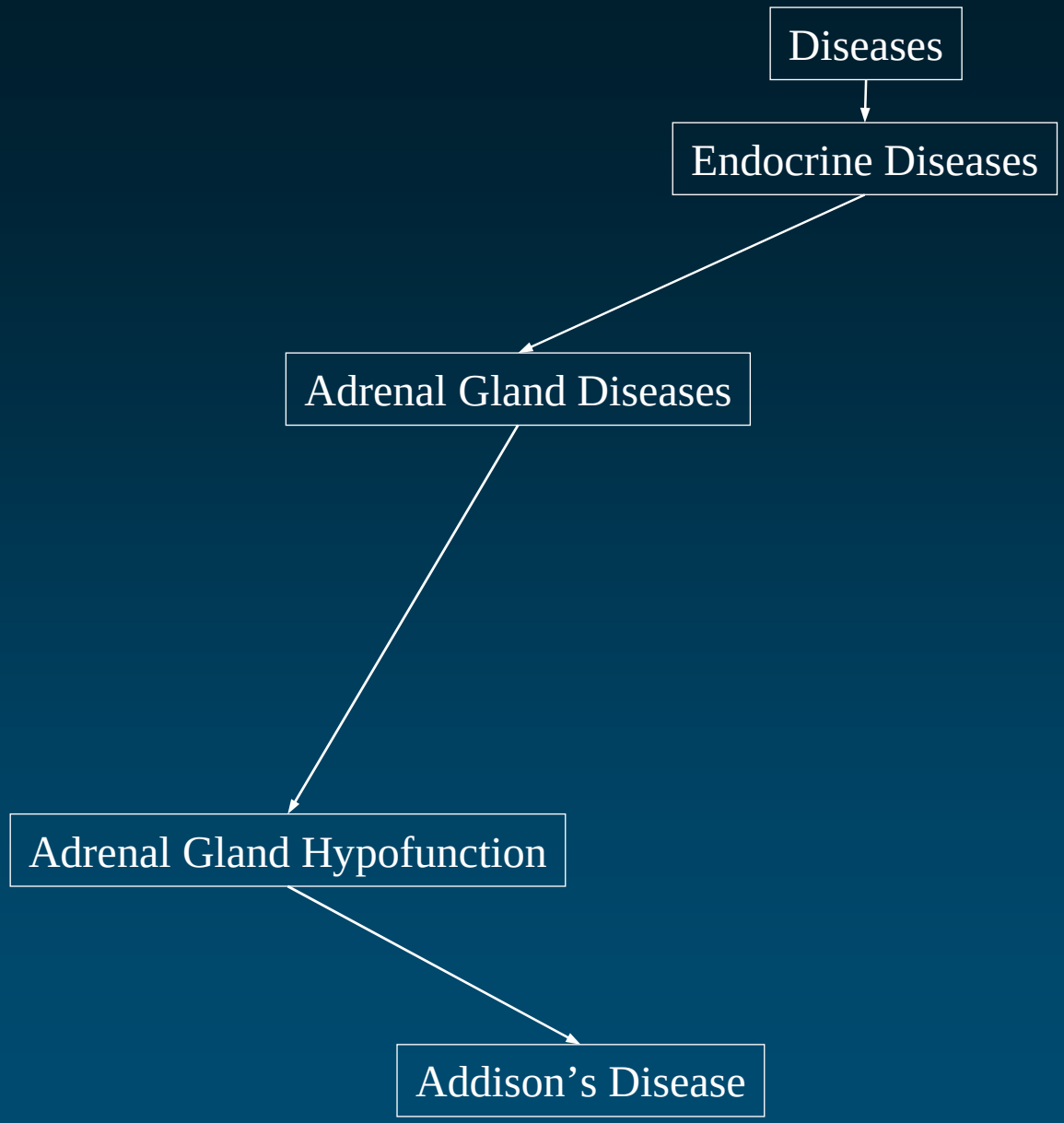
C0001621

Adrenal Gland Diseases

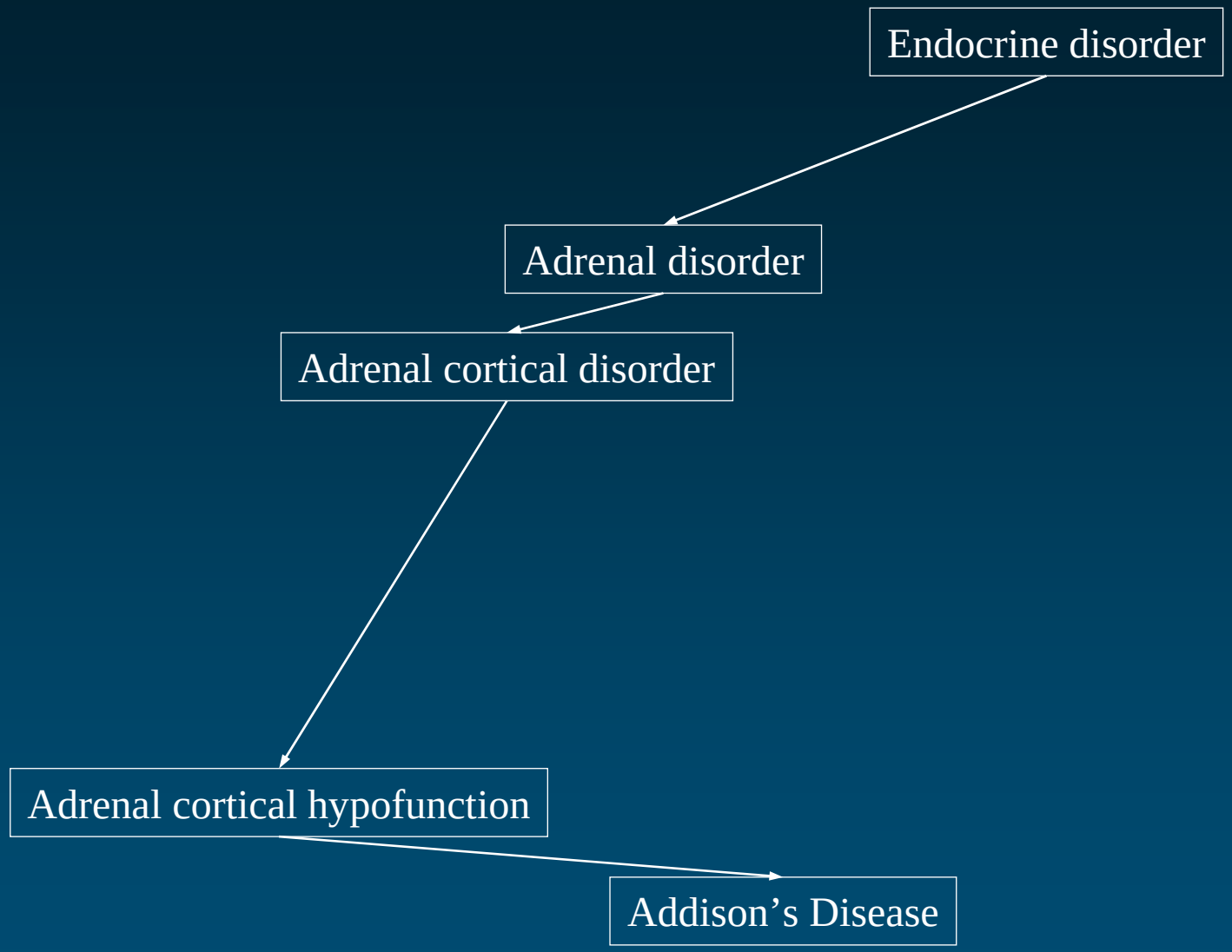
# SNOMED International



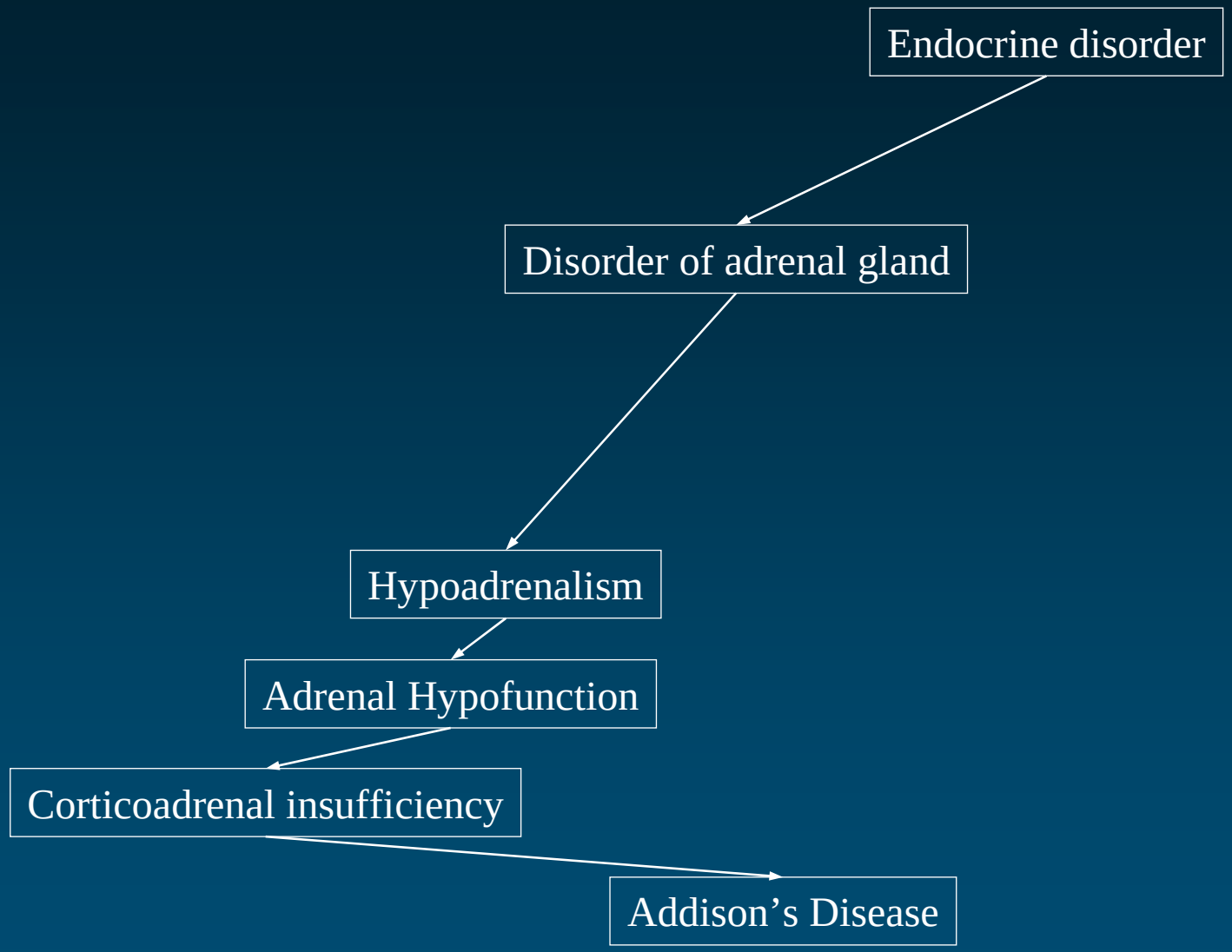
# MeSH


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

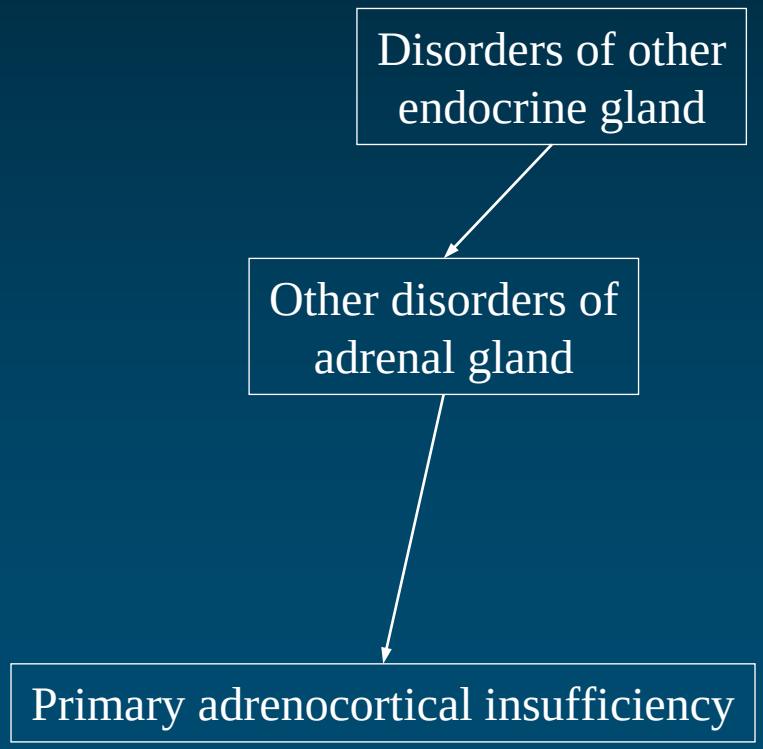
# AOD


Endocrine disorder
Adrenal disorder
Adrenal cortical disorder
Adrenal cortical hypofunction
Addison’s Disease

# Read Codes

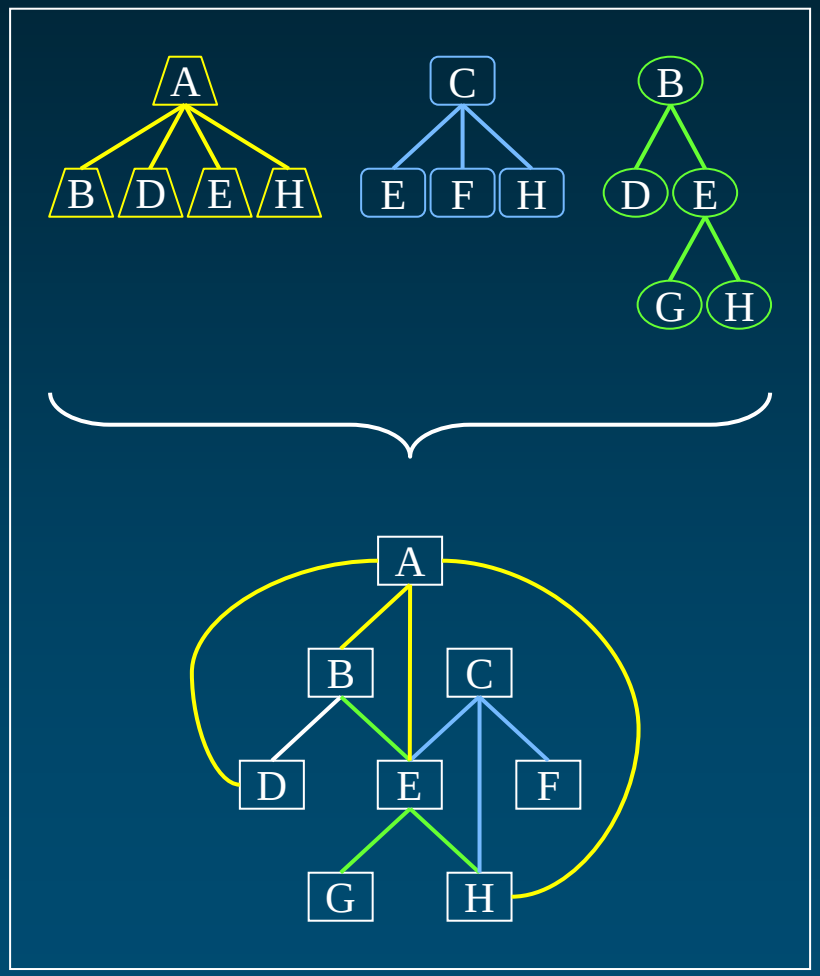

Endocrine disorder
Disorder of adrenal gland
Hypoadrenalism
Adrenal Hypofunction
Corticoadrenal insufficiency
Addison's Disease

# ICD-10

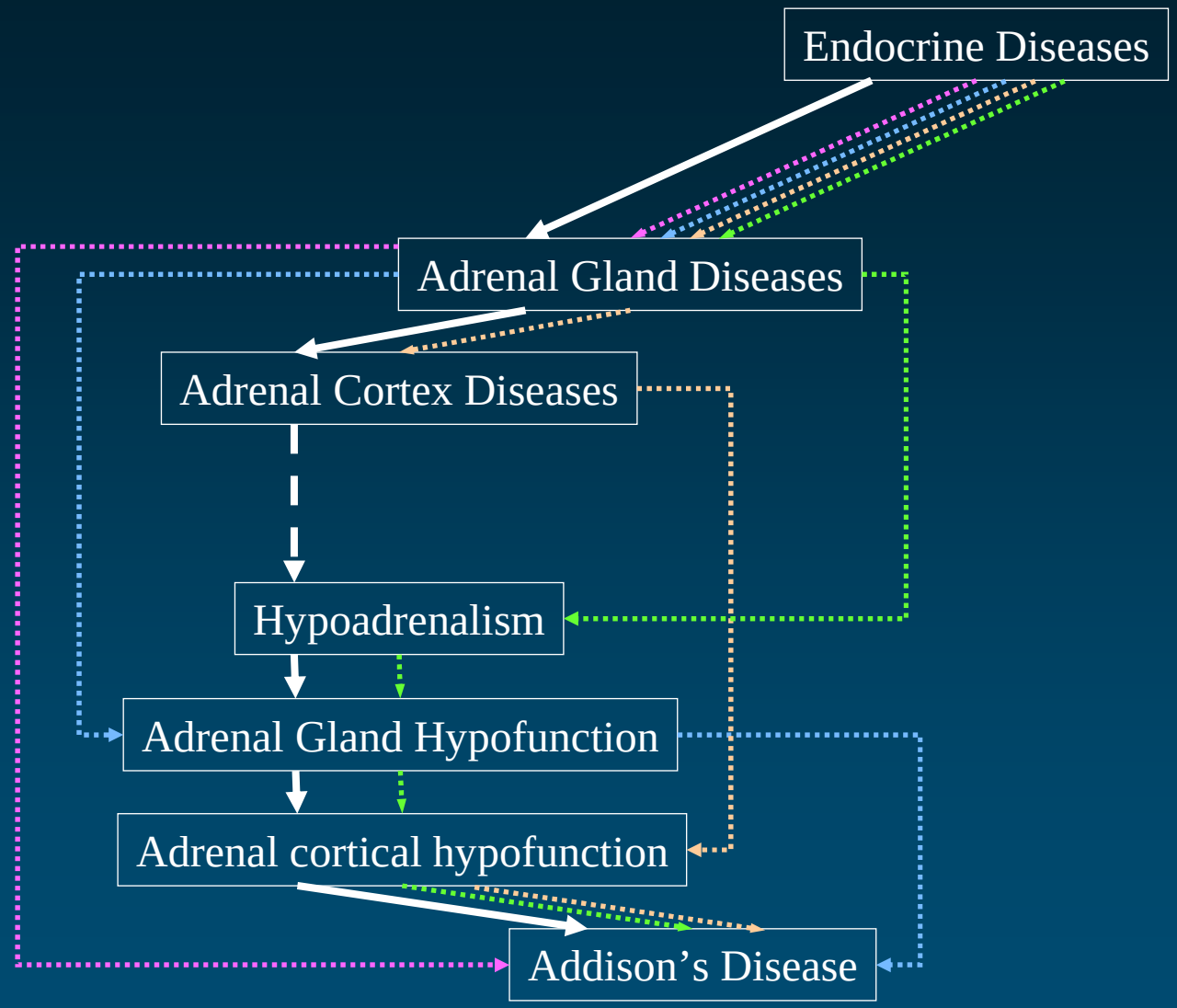

Disorders of other endocrine gland
Other disorders of adrenal gland
Primary adrenocortical insufficiency

# Organize concepts

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



## *organize concepts*

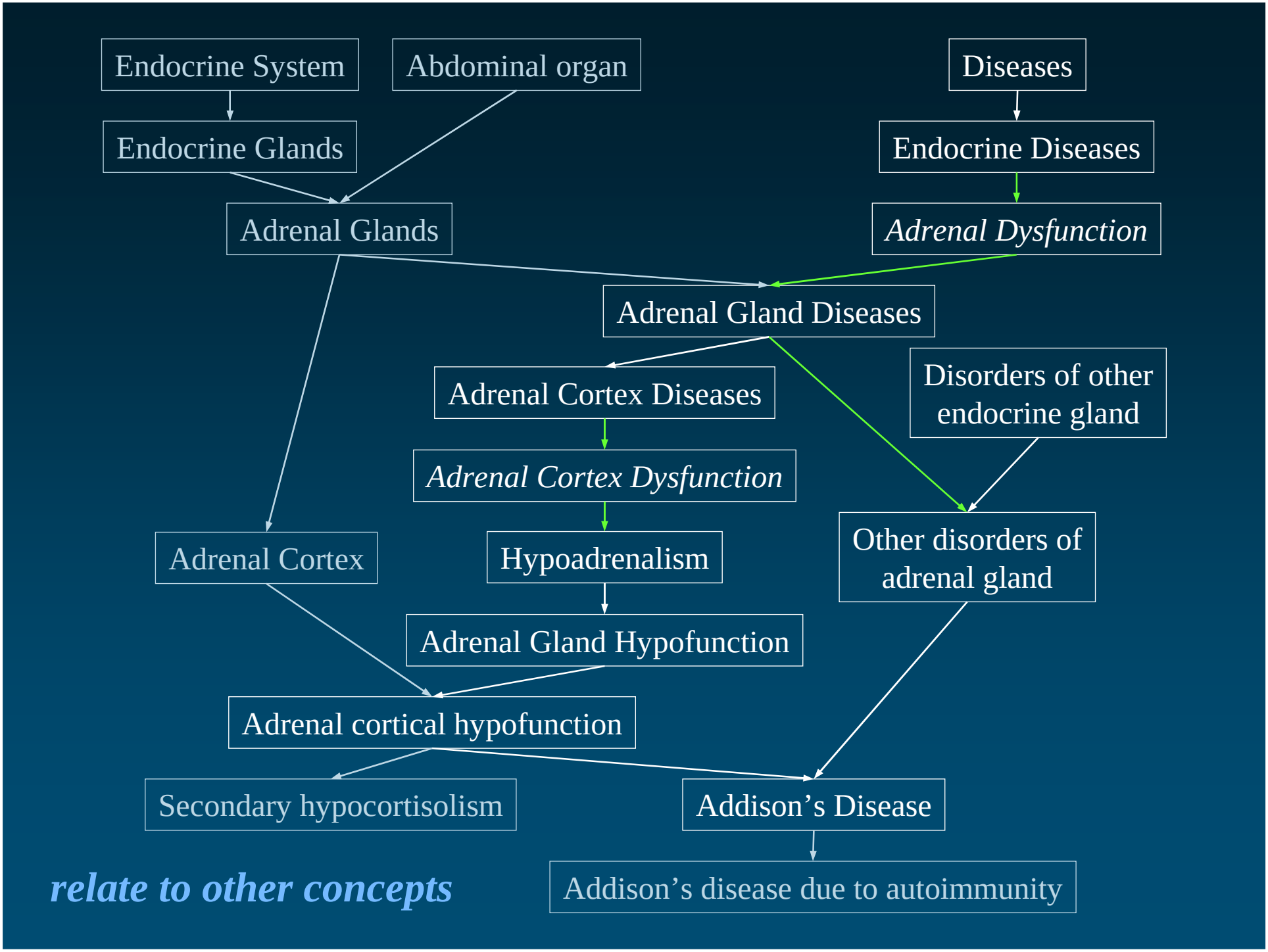



**SNOMED**
**MeSH**
**AOD**
**Read Codes**

**UMLS**

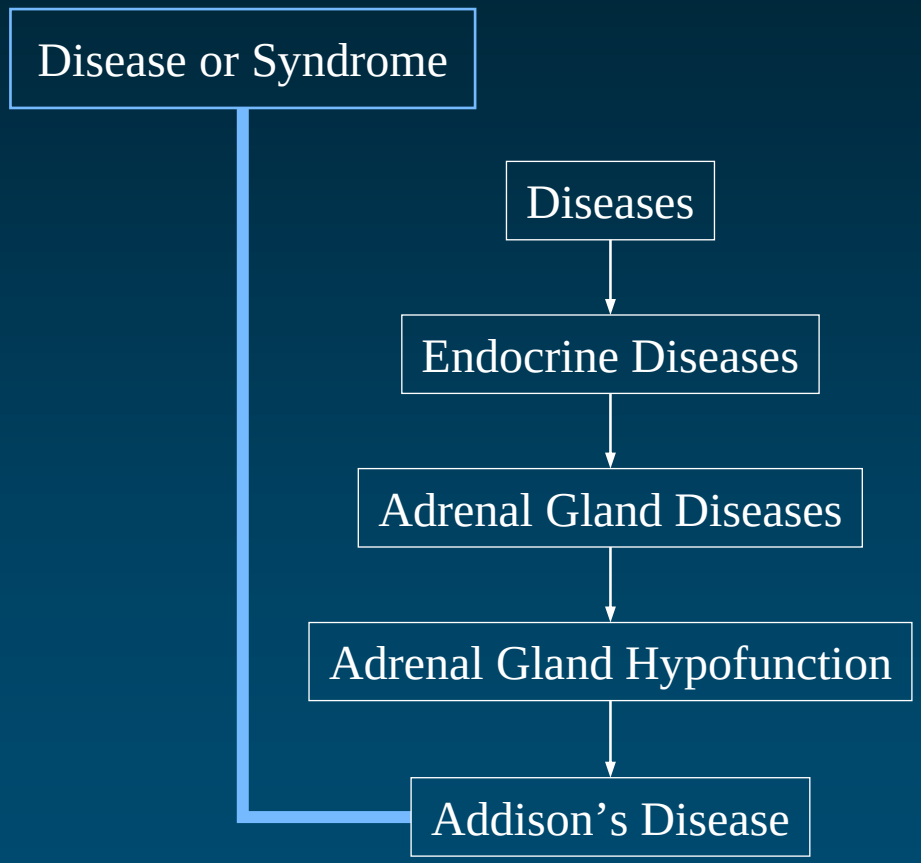
# Relate to other concepts

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


Endocrine System
Abdominal organ
Diseases
Endocrine Glands
Endocrine Diseases
Adrenal Glands
Adrenal Dysfunction
Adrenal Gland Diseases
Adrenal Cortex Diseases
Disorders of other endocrine gland
Adrenal Cortex Dysfunction
Adrenal Cortex
Hypoadrenalism
Other disorders of adrenal gland
Adrenal Gland Hypofunction
Adrenal cortical hypofunction
Secondary hypocortisolism
Addison's Disease
relate to other concepts
Addison's disease due to autoimmunity

# Categorize concepts

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



# How do they do that?

- Lexical knowledge
- Semantic pre-processing
- UMLS editors

# Lexical knowledge

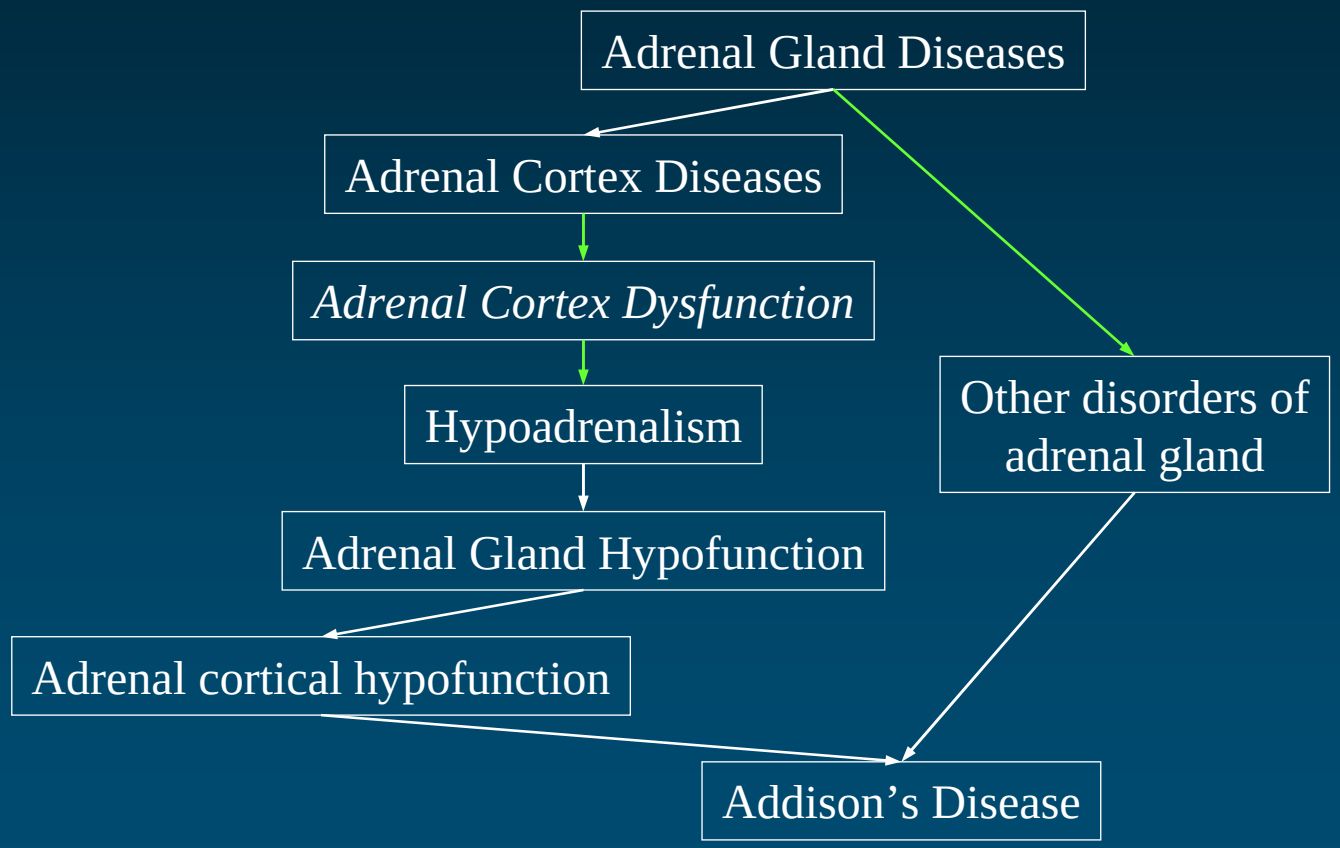
Adrenal gland diseases
Adrenal disorder
Disorder of adrenal gland
Diseases of the adrenal glands
C0001621

# Semantic pre-processing

- Metadata in the source vocabularies

- Tentative categorization
- Positive (or negative) evidence for tentative synonymy relations based on lexical features

# Additional knowledge: UMLS editors



# UMLS Summary

- Synonymous terms clustered into concepts
- Unique identifier

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

# Part I
# What is the UMLS?

## *(3) UMLS Knowledge Sources*

# UMLS 3 components

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

# UMLS Metathesaurus

# Metathesaurus Basic organization

- Concepts
  - Synonymous terms are clustered into a concept
  - Properties are attached to concepts, e.g.,
    - Unique identifier
    - Definition
- Relations
  - Concepts are related to other concepts
  - Properties are attached to relations, e.g.,
    - Type of relationship
    - Source

# Source Vocabularies

(2005AA)

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

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# Biomedical terminologies

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

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# Biomedical terminologies (cont'd)

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

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

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# Addison's Disease: Concept

Disease or Syndrome

Addison's Disease

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

MALADIE D'ADDISON - French
Addison-Krankheit - German
Morbo di Addison - Italian
DOENCA DE ADDISON - Portuguese
ADDISONOVA BOLEZN' - Russian
ENFERMEDAD DE ADDISON - Spanish

**SNOMED**
**MeSH**
**AOD**
**Read Codes**
**…**

C0001403

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

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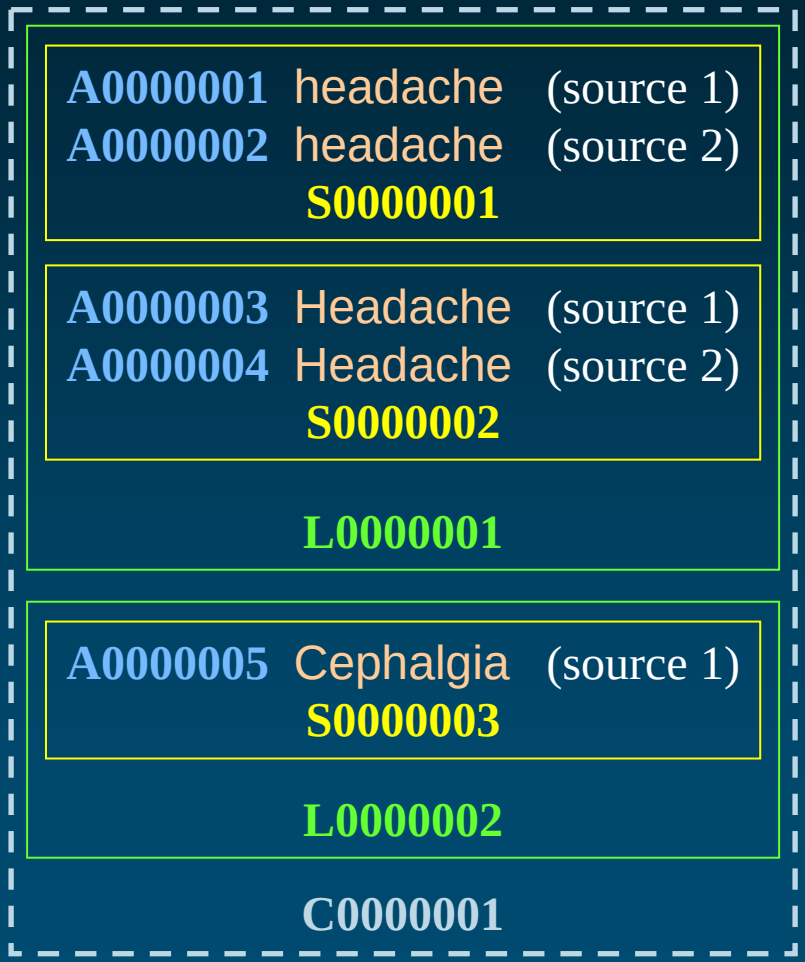
# Metathesaurus Concepts

(2005AA)

- Concept (~ 1.2M) CUI
  - Set of synonymous concept names
- Term (~ 4.2 M) LUI
  - Set of normalized names
- String (~ 4.7M) SUI
  - Distinct concept name
- Atom (~ 5.5M) AUI
  - Concept name in a given source



# Cluster of synonymous terms

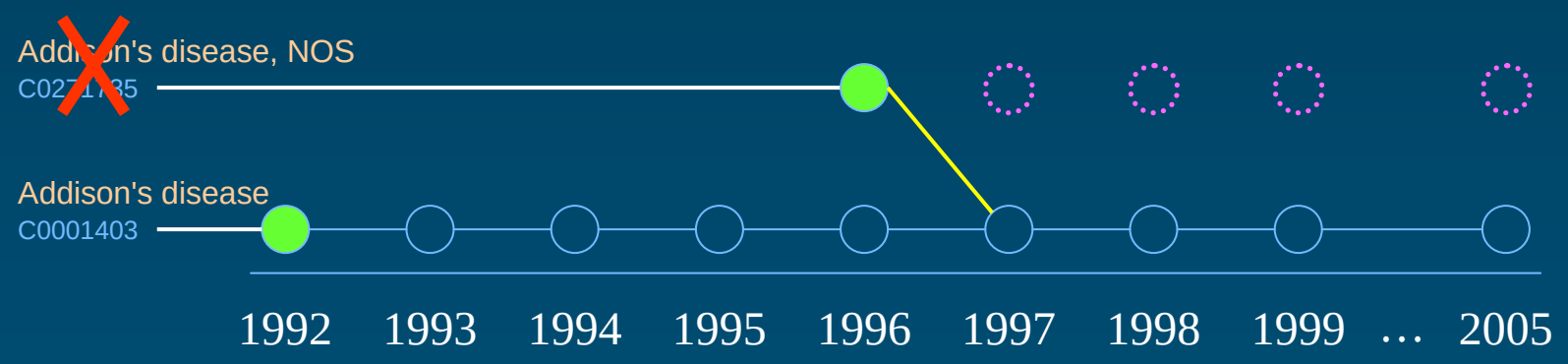
Concept
C0001621

| | | |
|---|---|---|
| Term L0001621 | **S0011232** *Adrenal Gland Diseases*<br>**S0011231** Adrenal Gland Disease<br>**S0000441** Disease of adrenal gland<br>**S0481705** Disease of adrenal gland, NOS<br>**S0220090** Disease, adrenal gland<br>**S0044801** Gland Disease, Adrenal | […] |
| Term L0041793 | **S0860744** *Disorder of adrenal gland, unspecified*<br>**S0217833** Unspecified disorder of adrenal glands | |
| Term L0161347 | **S0225481** *ADRENAL DISORDER*<br>**S0627685** DISORDER ADRENAL (NOS) | […] |
| Term L0181041 | **S0632950** *Disorder of adrenal gland*<br>**S0354509** Adrenal Gland Disorders | […] |
| Term L0368399 | **S0586222** *Adrenal disease*<br>**S0466921** ADRENAL DISEASE, NOS | […] |
| Term L1279026 | **S1520972** *Nebennierenkrankheiten* | GER |
| Term L0162317 | **S0226798** *SURRENALE, MALADIES* | FRE |

[…]

# Metathesaurus Evolution over time

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



# Metathesaurus Relationships

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

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

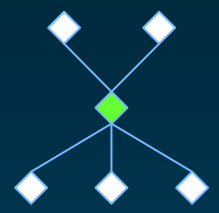
# Symbolic relations

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

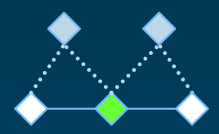
Source transparency: the information is recorded at the “atom” level

# Symbolic relationships Type

- Hierarchical
  - Parent / Child **PAR/CHD**
  - Broader / Narrower than **RB/RN**



- Derived from hierarchies
  - Siblings (children of parents) **SIB**

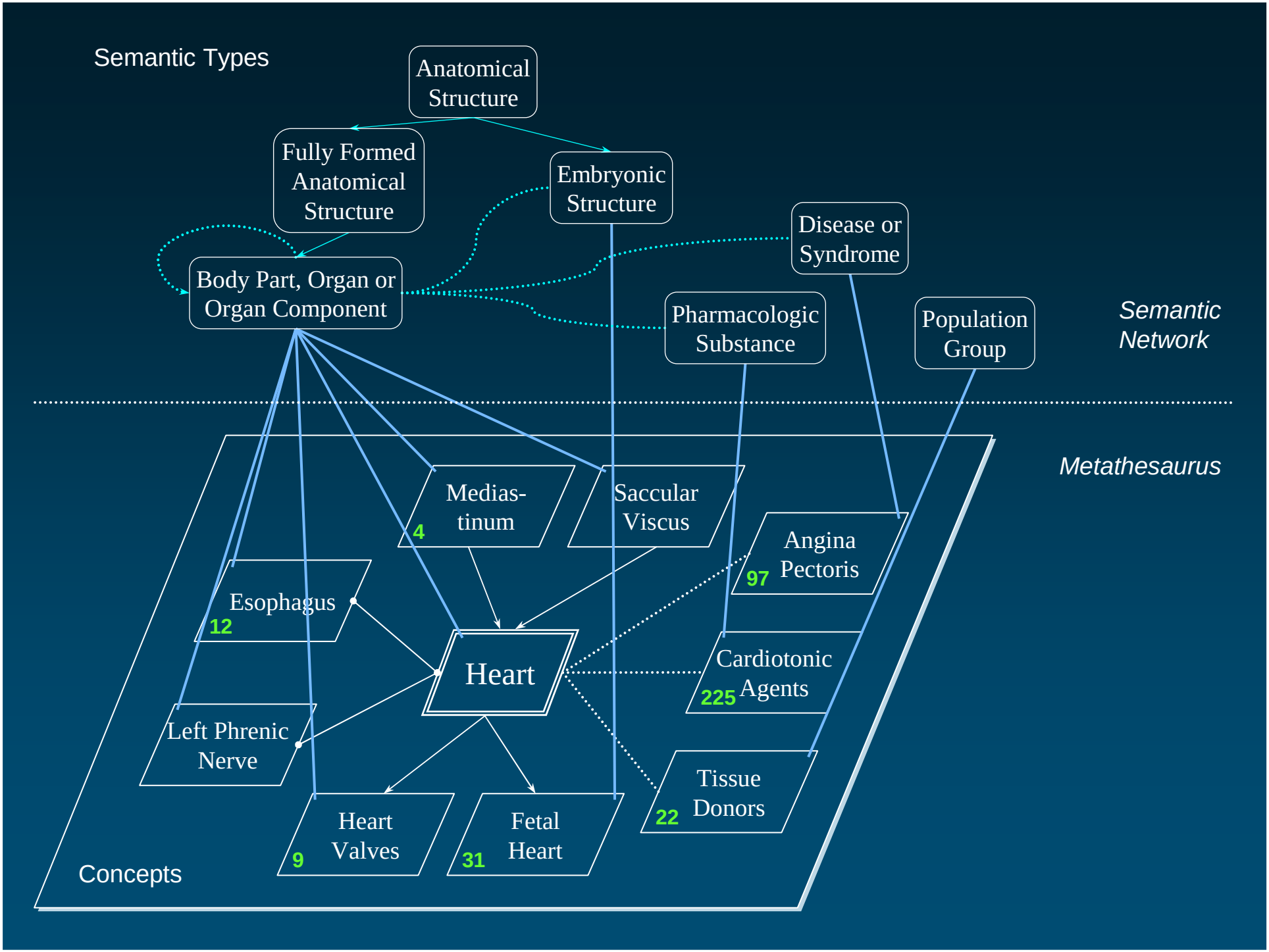


- Associative
  - Other **RO**

- Various flavors of near-synonymy
  - Similar **RL**
  - Source asserted synonymy **SY**
  - Possible synonymy **RQ**

# Symbolic relationships Attribute

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


Semantic Types
Anatomical Structure
Fully Formed Anatomical Structure
Embryonic Structure
Disease or Syndrome
Body Part, Organ or Organ Component
Pharmacologic Substance
Population Group
Semantic Network
Metathesaurus
Medias-tinum
4
Saccular Viscus
Angina Pectoris
97
Esophagus
12
Heart
Cardiotonic Agents
225
Left Phrenic Nerve
Tissue Donors
22
Heart Valves
9
Fetal Heart
31
Concepts

# UMLS Semantic Network

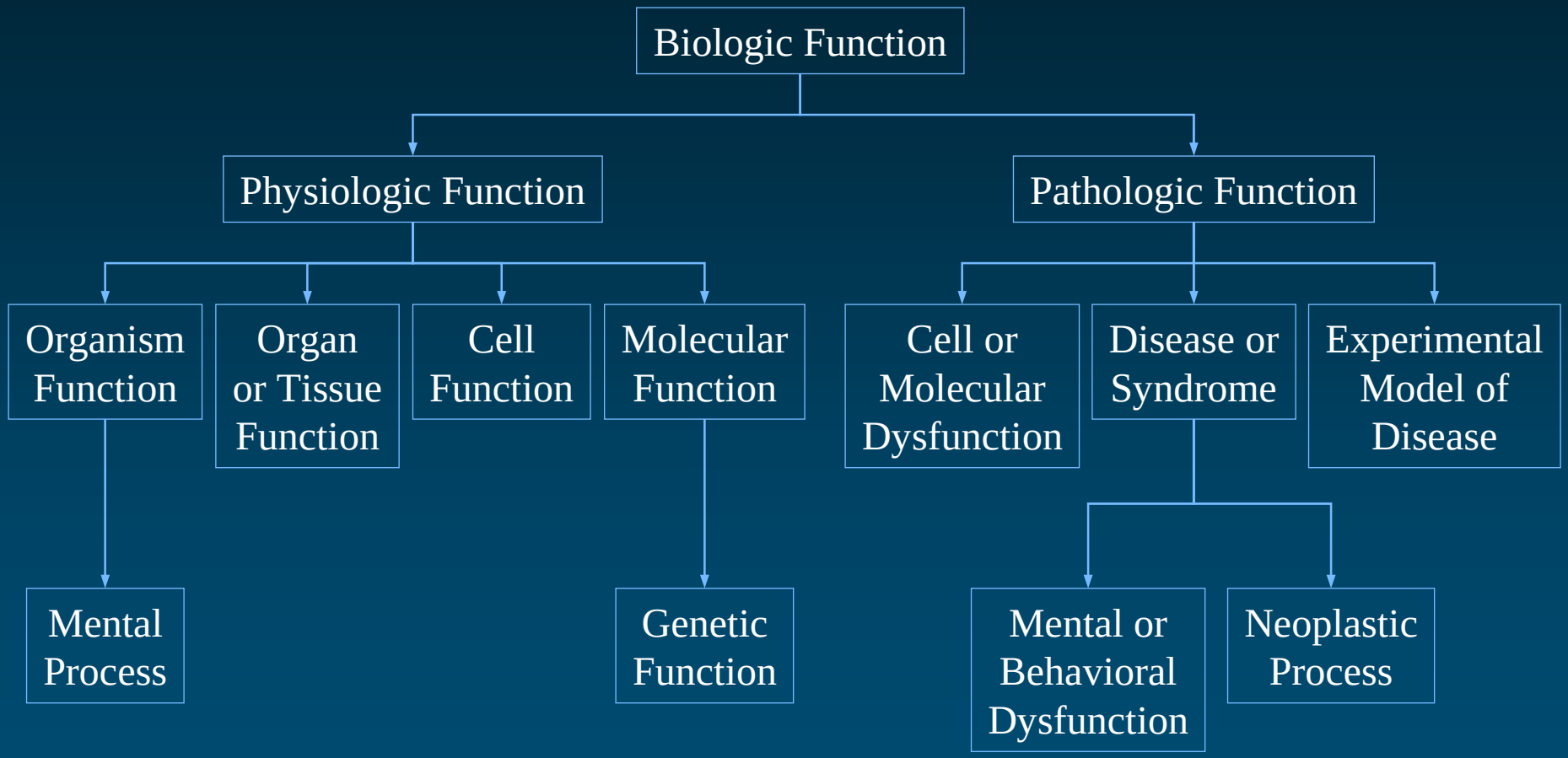
# Semantic Network

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

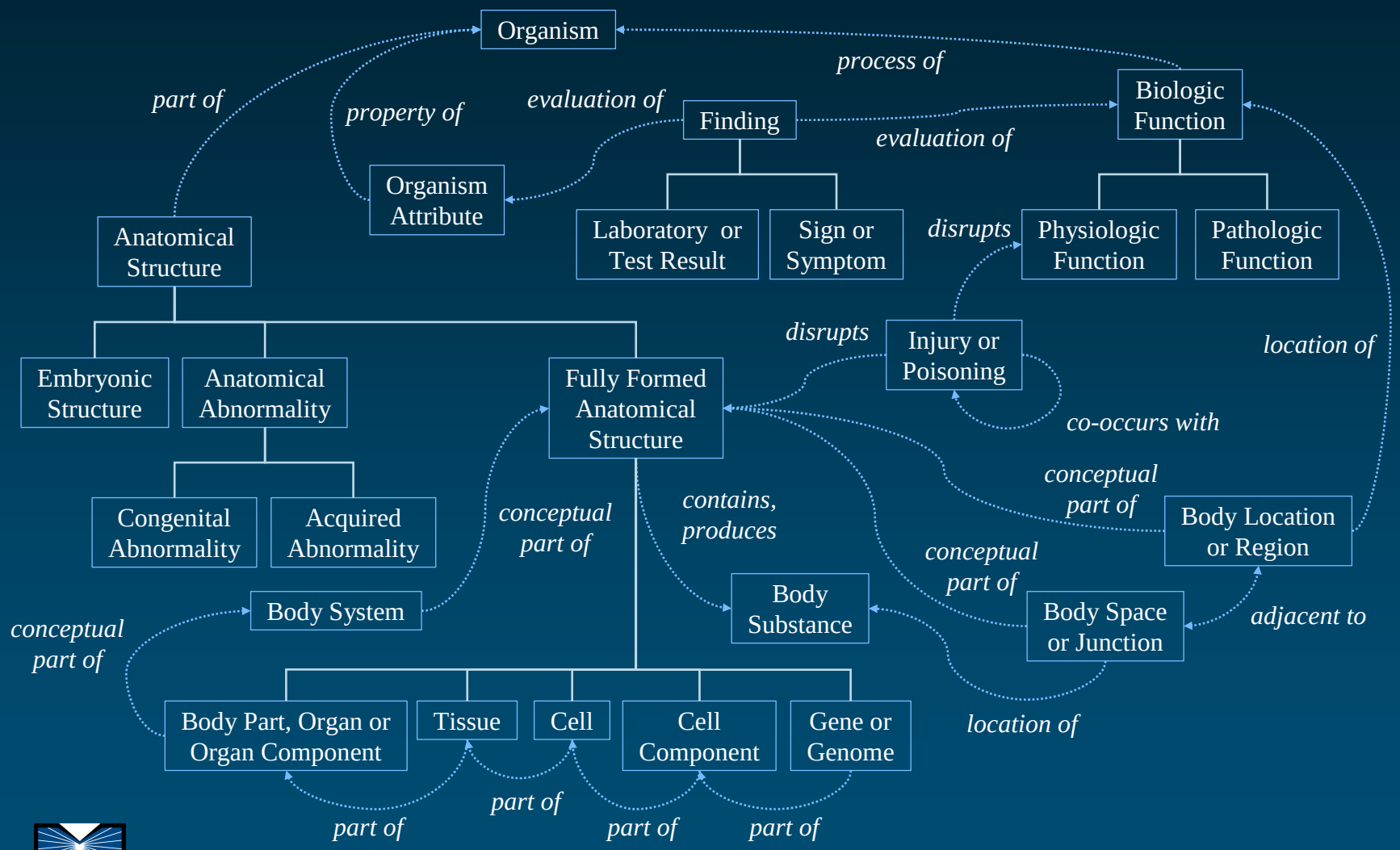
# Semantic Network

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

# “Biologic Function” hierarchy (isa)



# Associative (non-isa) relationships

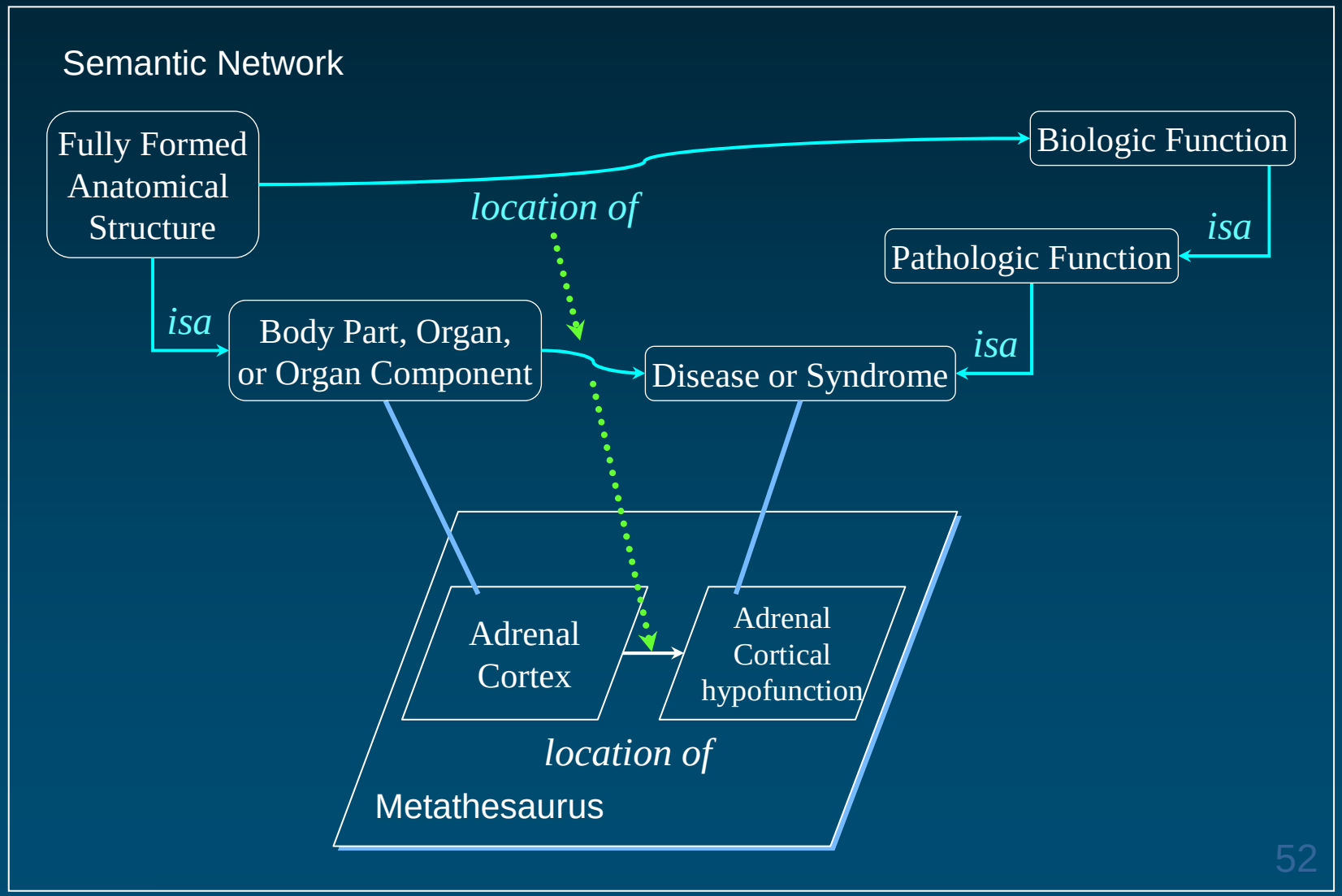


# Why a semantic network?

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

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

# Relationships can inherit semantics



# SPECIALIST Lexicon and lexical tools

# SPECIALIST Lexicon

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

# Morphology

- Inflection
  - noun — nucleus, nuclei
  - verb — cauterize, cauterizes, cauterized, cauterizing
  - adjective — red, redder, reddest
- Derivation
  - verb ⟺ noun — cauterize -- cauterization
  - adjective ⟺ noun — red -- redness

# Orthography

- Spelling variants
  - oe/e — oesophagus - esophagus
  - ae/e — anaemia - anemia
  - ise/ize — cauterise - cauterize
  - genitive mark — Addison's disease<br>Addison disease<br>Addisons disease

# Syntax

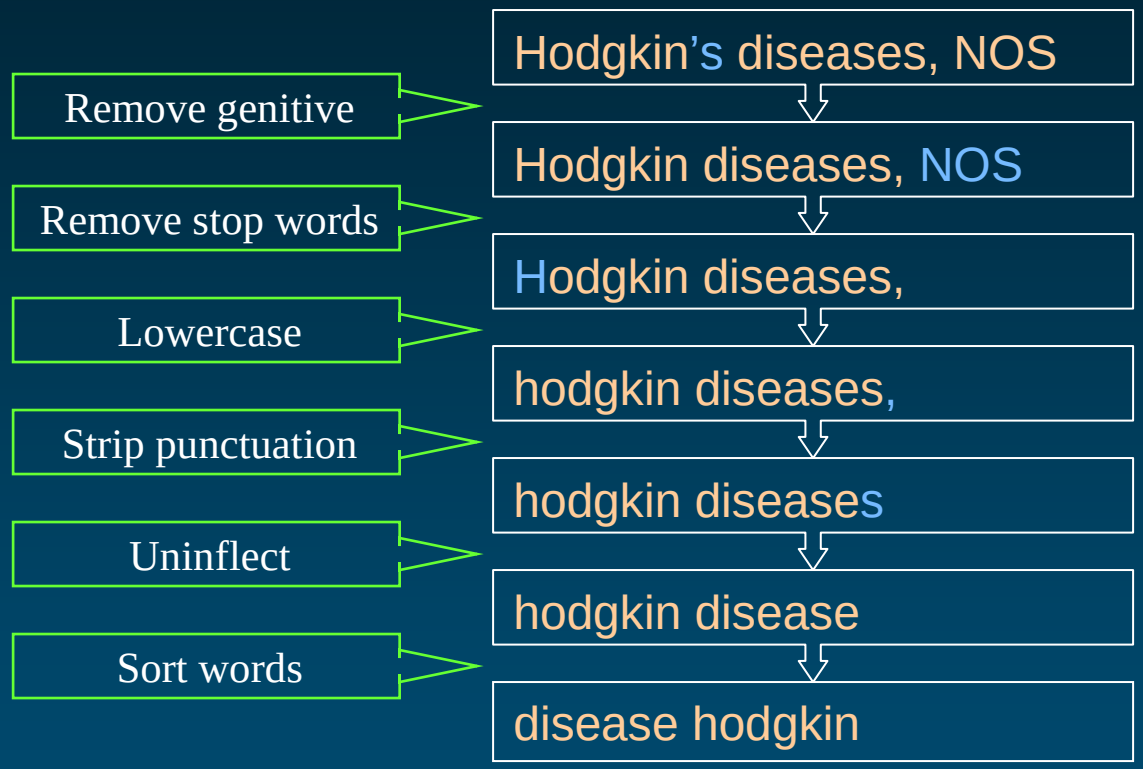
- Complementation
  - verbs
    - intransitive — I'll treat.
    - transitive — He treated the patient.
    - ditransitive — He treated the patient with a drug.
  - nouns
    - prepositional phrase

      Valve of coronary sinus
- Position for adjectives

# Lexical tools

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

# Normalization



# Normalization: Example

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

normalize → disease hodgkin

# Normalization Applications

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

# Indexes

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

# Lexical Variant Generation program

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

# Part II

# How to use the UMLS?

# Outline

- 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

- Online Web-based license:

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

  - Read license — Accept and continue
  - Read appendix — Accept
  - Print a copy for your records — Printer-friendly Version
  - Complete the Web form — Submit

  - 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 2nd 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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annual report

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12. 2. **Category 2:**

All category 1 restrictions AND

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Accept & continue

Accept and continue | Not accept

**Last updated:** 26 March 2004
**First published:** 01 January 1997
**Permanence level:** Permanence Not Guaranteed
**Previous version**

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## APPENDIX A.1

### Appendix to the License Agreement for Use of the UMLS® Metathesaurus

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

Contact: May Cheh, Lister Hill Center, National Library of Medicine, Bethesda MD; e-mail: cheh@nlm.nih.gov

---

**ALT2003** Alternative Billing Concepts (AltLink). Albuquerque (NM): Alternative Link LLC, 2003.

CATEGORY 3 RESTRICTIONS APPLY

Contact: Alternative Link LLC; 6121 Indian School Road NE, Suite 131; Albuquerque, NM 87110; phone: 877-621-5465; http://www.alternativelink.com; e-mail: mail@alternativelink.com

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**VANDF03** U.S. Department of Veterans Affairs, Veterans Health Administration National Drug File. Department of Veterans Affairs, Washington, DC. Release Date: March 13, 2003.

*NOTE: Now a CATEGORY 0

Contact: Steven Brown; CPEP Office; 1310 24th Avenue S; Nashville, TN 37215; e-mail: Steven.Brown@msd.va.gov

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**WHO97** WHO Adverse Drug Reaction Terminology (WHOART). Uppsala (Sweden): WHO Collaborating Centre for International Drug Monitoring, 1997.

CATEGORY 2 RESTRICTIONS APPLY

The Metathesaurus includes translations of WHO97 in French (WHOFRE_1997), German (WHOGER_1997), Portuguese (WHOPOR_1997), and Spanish (WHOSPA_1997).

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

Accept Not accept

**Last updated:** 20 July 2004
**First published:** 26 March 2004
**Permanence level:** Permanence Not Guaranteed

# License Restriction Levels 0-4

(2004AB)

- Level 0 (28.2%)
  - *unrestricted*
- Level 1 (1.6%)
  - *negotiate to translate*
- Level 2 (0.4%)
  - *negotiate to use in health data creation*
- Level 3 (30.6%)
  - *negotiate to use in production*
  - *explicitly prohibited to provide Internet access*
- Level 4 (39.2%)
  - *unrestricted for U.S. use and distribution*

**67%**

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NLM

# 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)

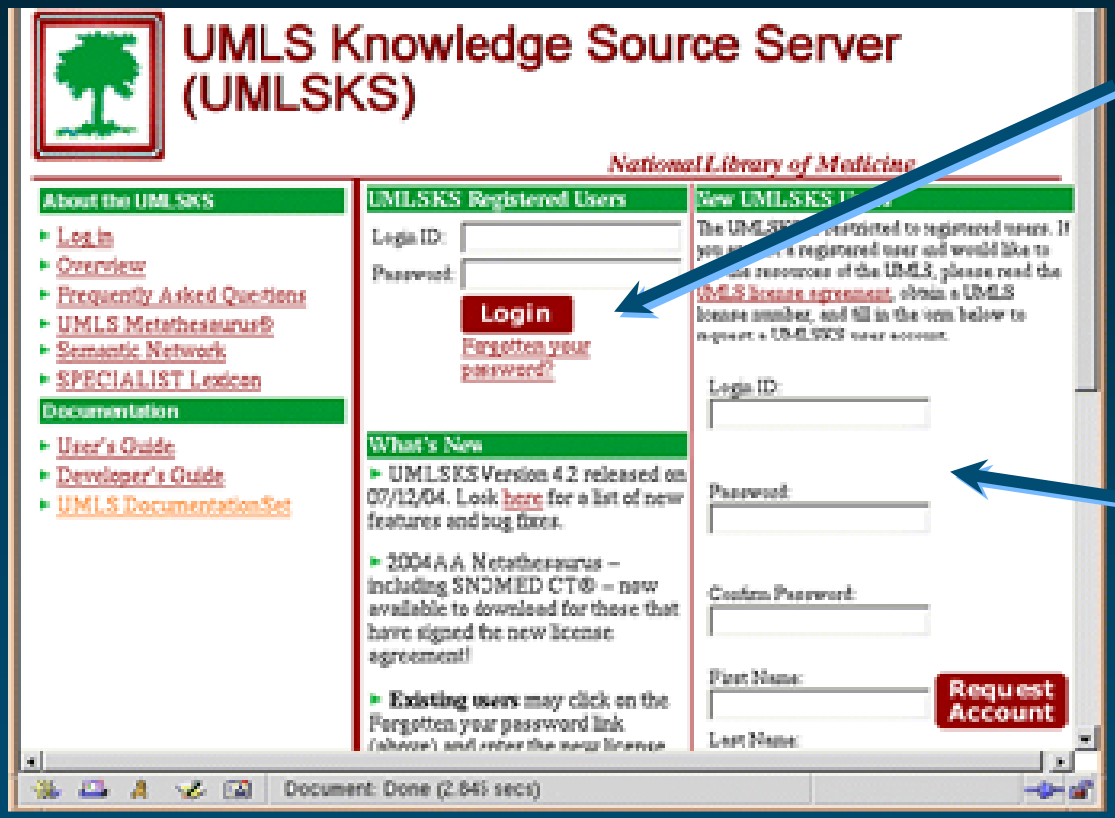
# Knowledge Source Server

*Web search interface*

# UMLSKS Web search interface

- Logging in
- Basic searching
- Advanced searching

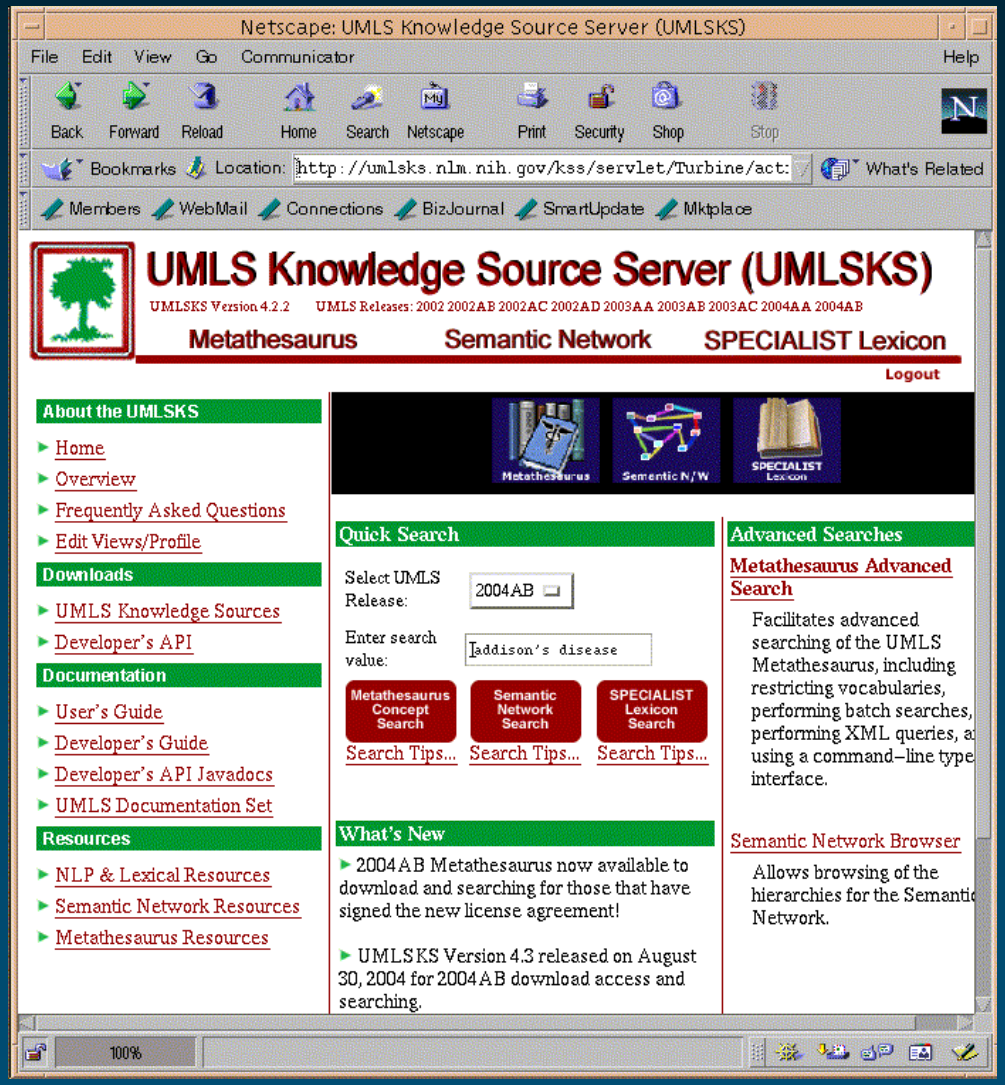
# UMLSKS Web search interface log in



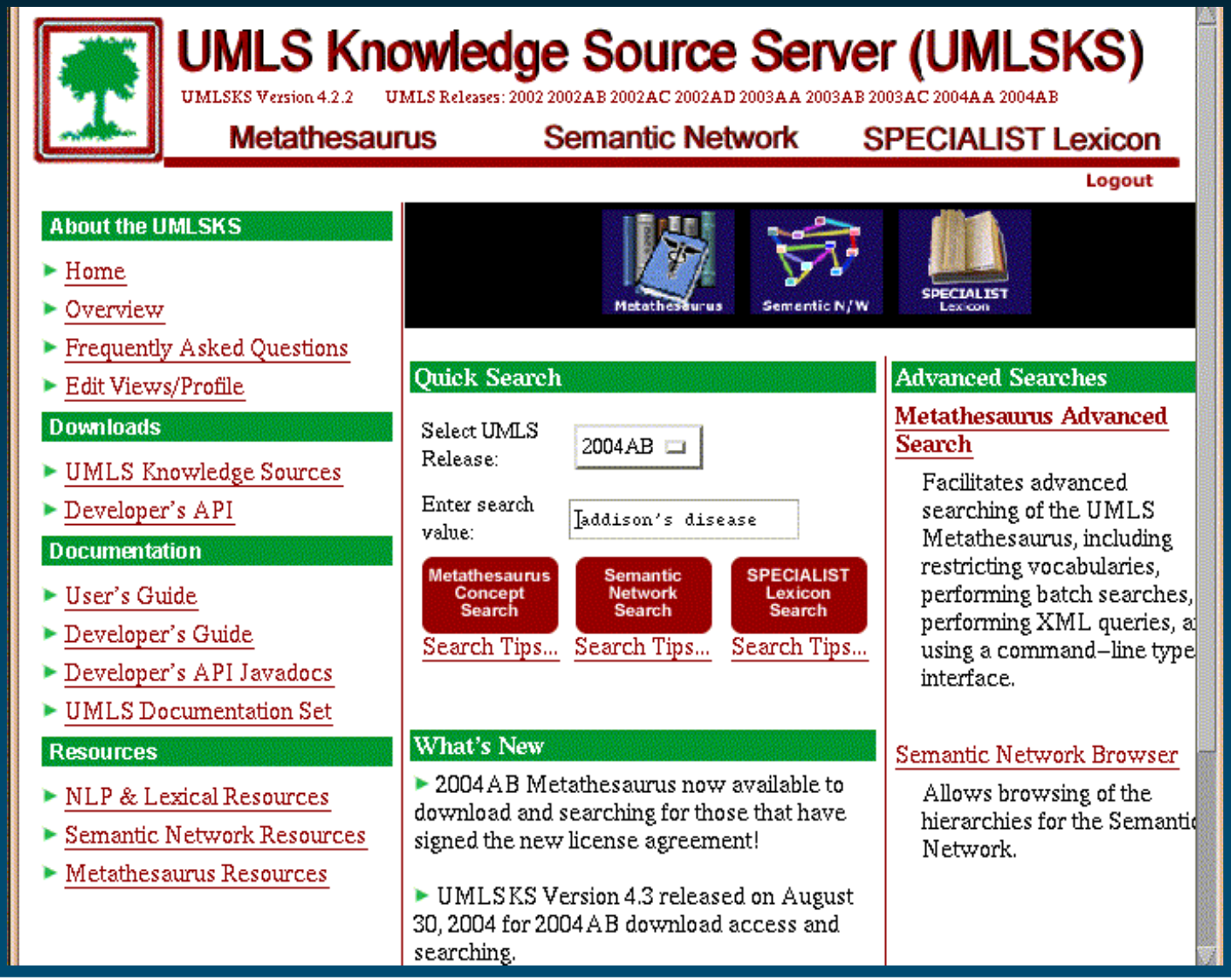

- Returning users log in
- New users create account

# UMLS Knowledge Source Server Home Page

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

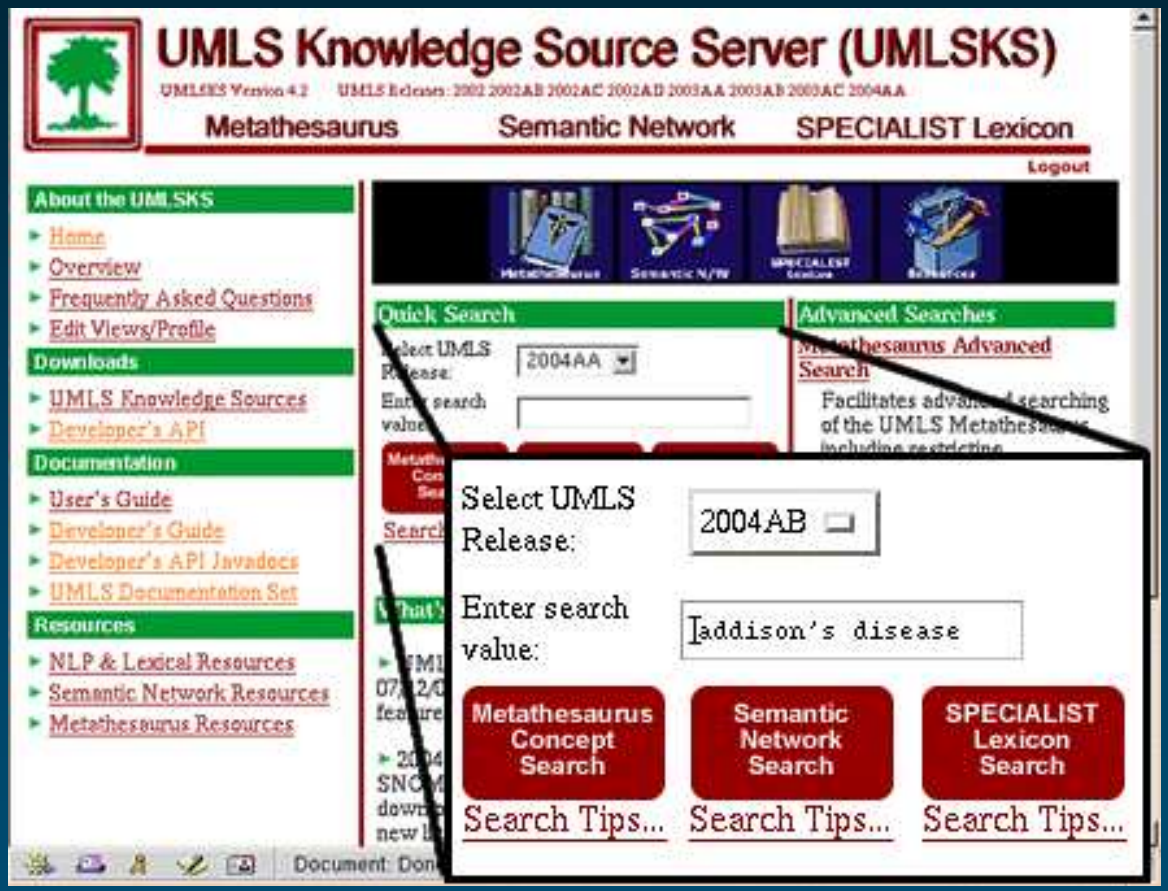


# UMLS Knowledge Source Server Home Page



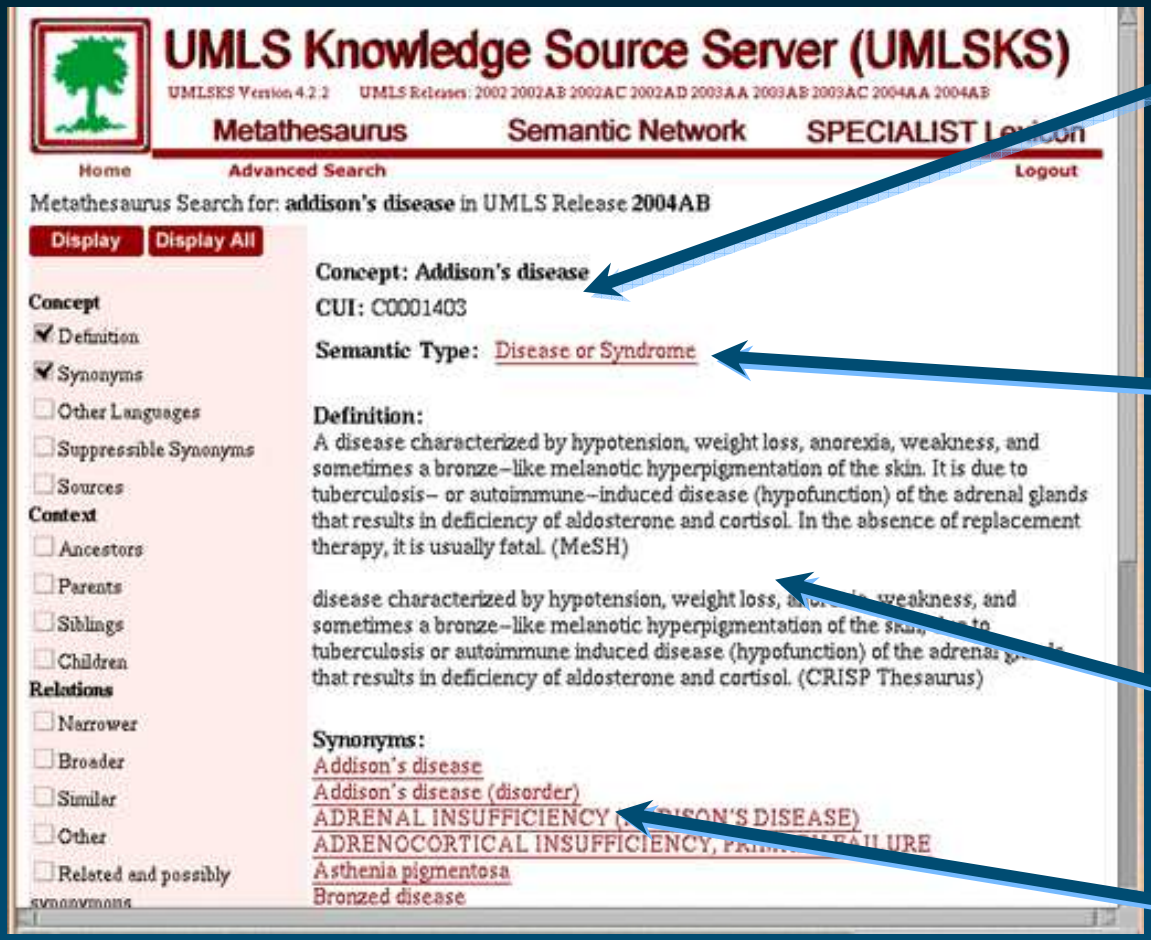
# Metathesaurus Basic Search

## *Addison's disease*



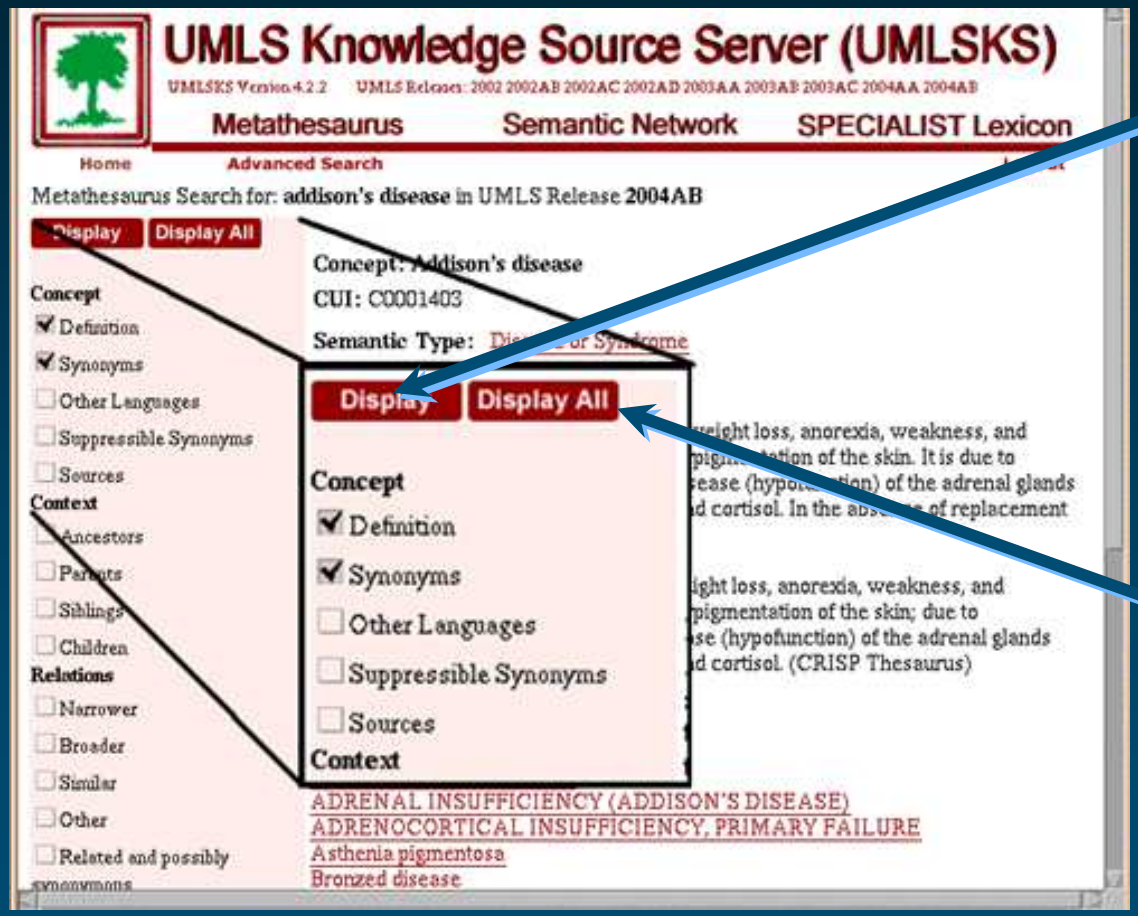

- UMLS Release
- Search Term
- UMLS Knowledge Source

# Concept Report *Addison's disease*




- Concept Name /CUI
- Semantic Type(s)
- Definition(s)
- Synonyms

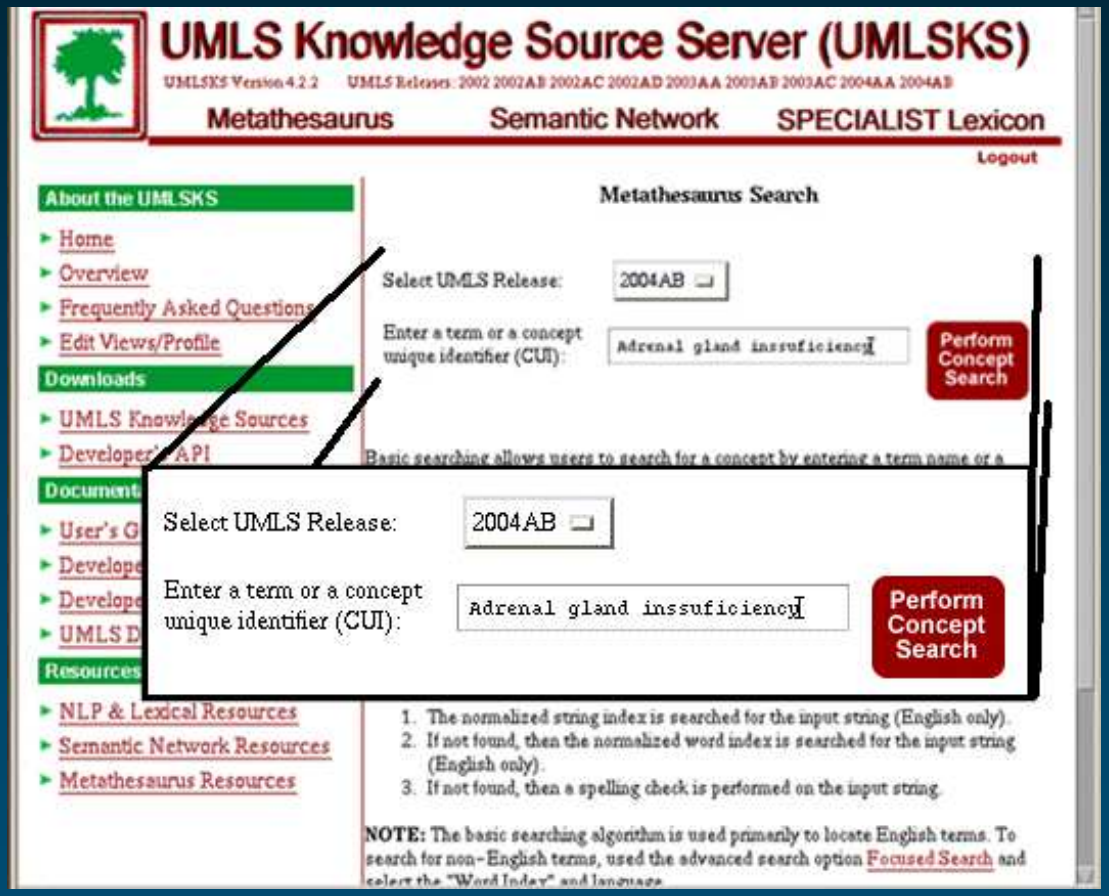
# Display All




- "Display" shows results for selected options
- "Display All" shows results for all available options

# Metathesaurus Basic Search

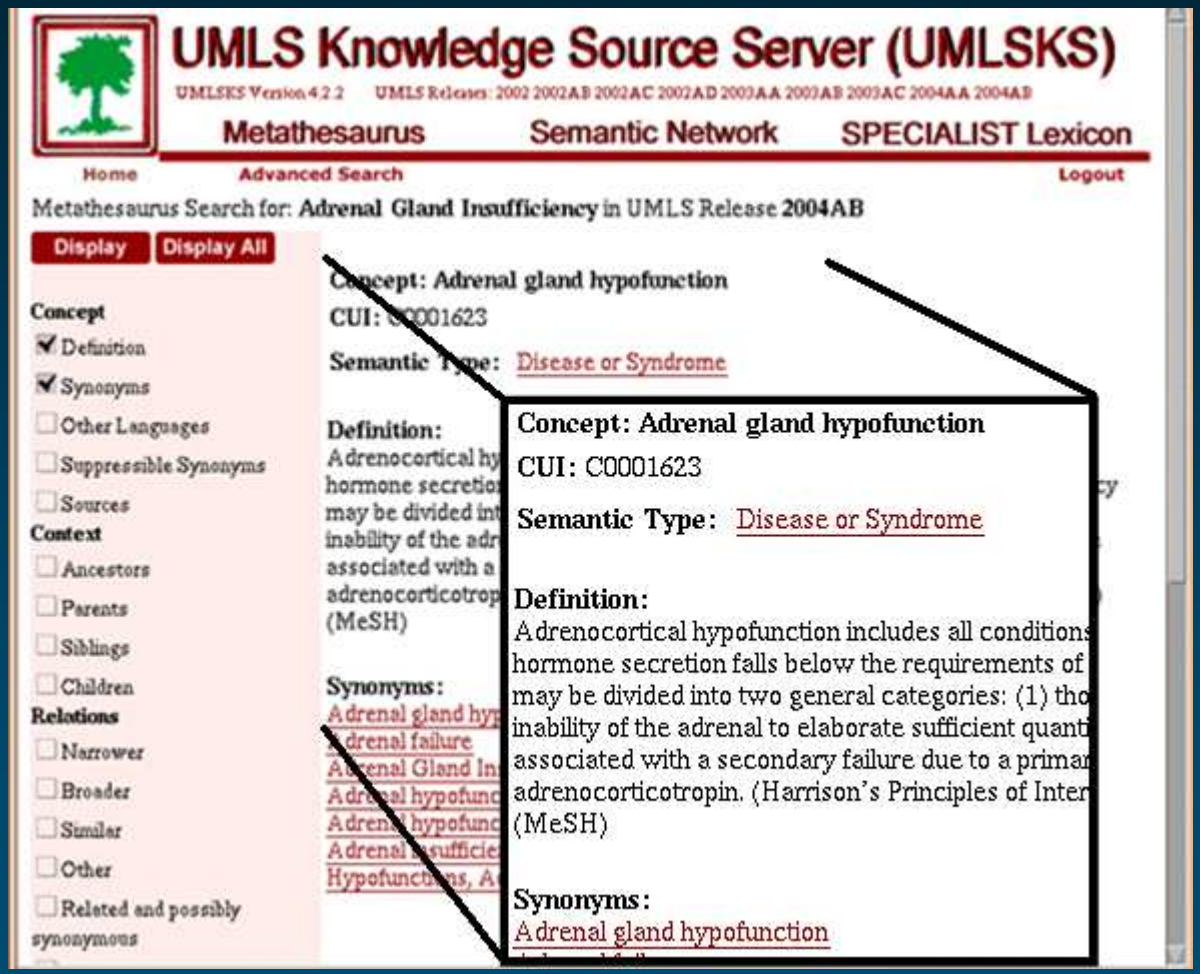
## *Adrenal gland insufficiency*



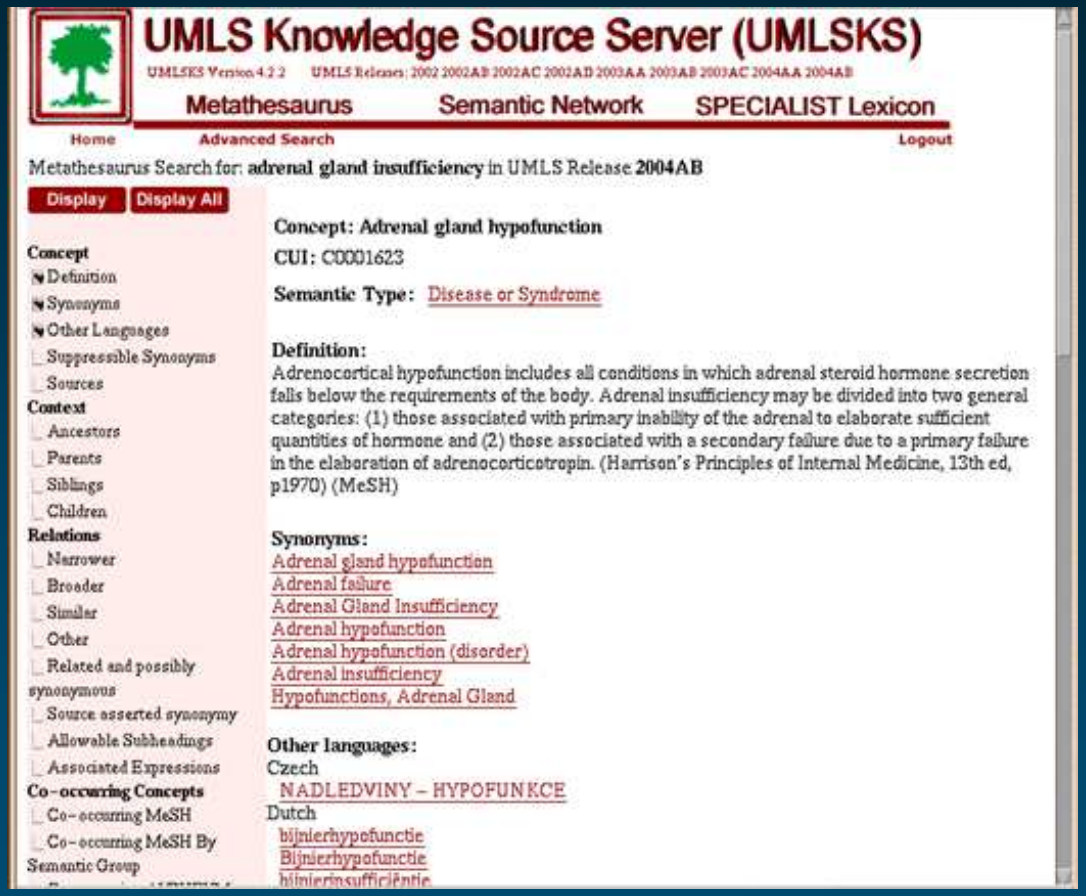

- Specify:
  - UMLS Release
  - Search term
- Algorithm:
  - Search Normalized String
  - Search Normalized Word
  - Suggest Spelling

# Basic Concept Report

## *Adrenal gland insufficiency*



# Concept Report Display All
## *Adrenal Gland Insufficiency*




- 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
HIPOFUNCAO SUPRA–RENAL
Hipofunção supra–renal
INSUFICIENCIA SUPRA–RENAL
Insuficiência supra–renal
Insuficiência supra–ren
Russian
НАДПОЧЕЧН
Spanish
hipofuncion adrenal
hipofunción adrenal
Hipofunción de las Glán
hipofuncion suprarrenal
Hipofuncion suprarrenal
hipofunción suprarrenal
Hipofuncion suprarrenal
hipofuncion suprarrenal
hipofunción suprarrenal
Insuficiencia adrenal
Insuficiencia suprarrena
INSUFICIENCIA SUP
Insuficiencia suprarren
SUPRARRENAL HIPOFUNCION
Swedish
Binjurehypofunktion

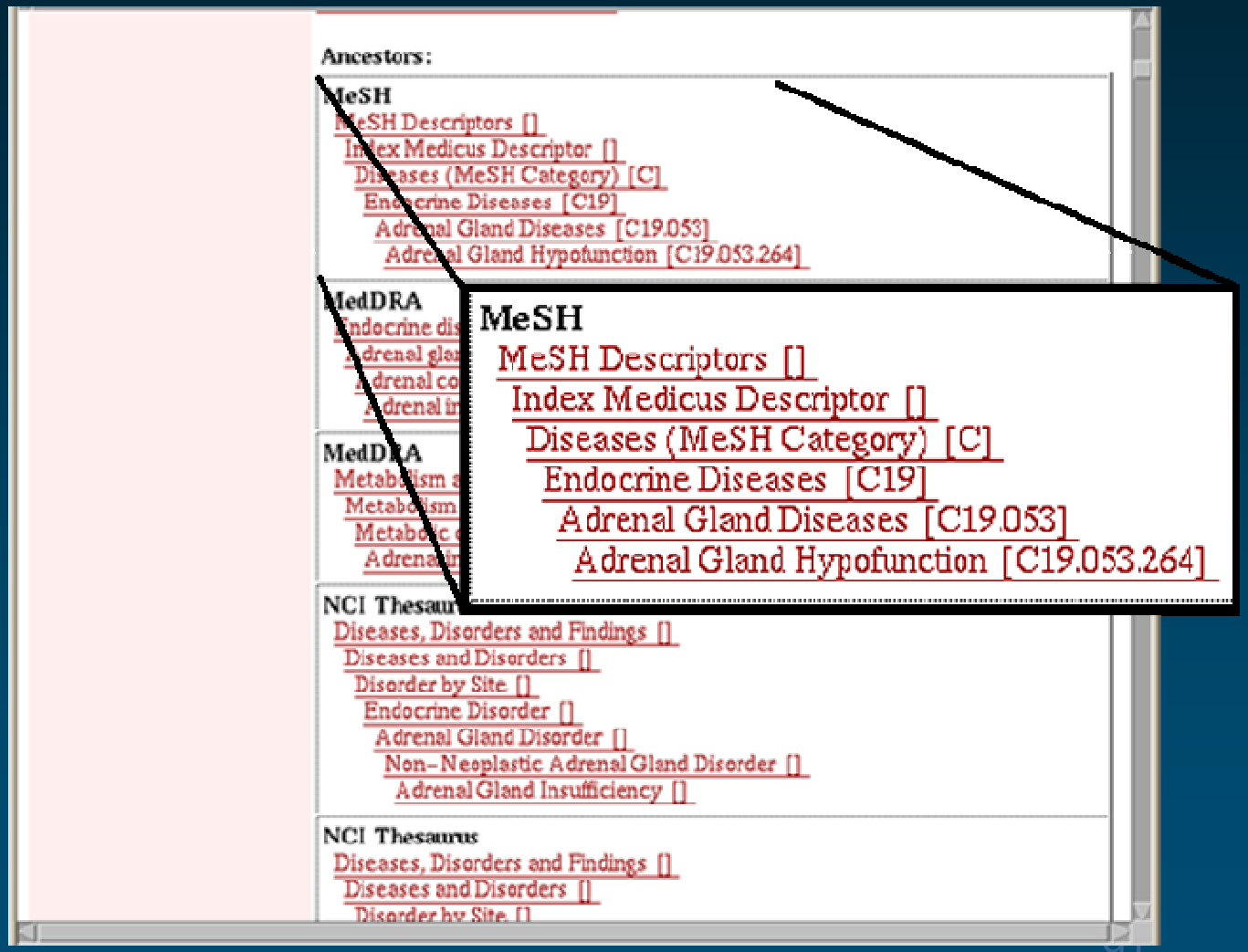
**Suppressible Synonyms:**
adrenal insufficiency <2>

**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
HIPOFUNCAO SUPRA–RENAL
Hipofunção supra–renal
INSUFICIENCIA SUPRA–RENAL
Insuficiência supra–renal
Insuficiência supra–renal NE
Russian
НАДПОЧЕЧНИКОВ ГИПОФУНКЦИЯ
Spanish
hipofuncion adrenal

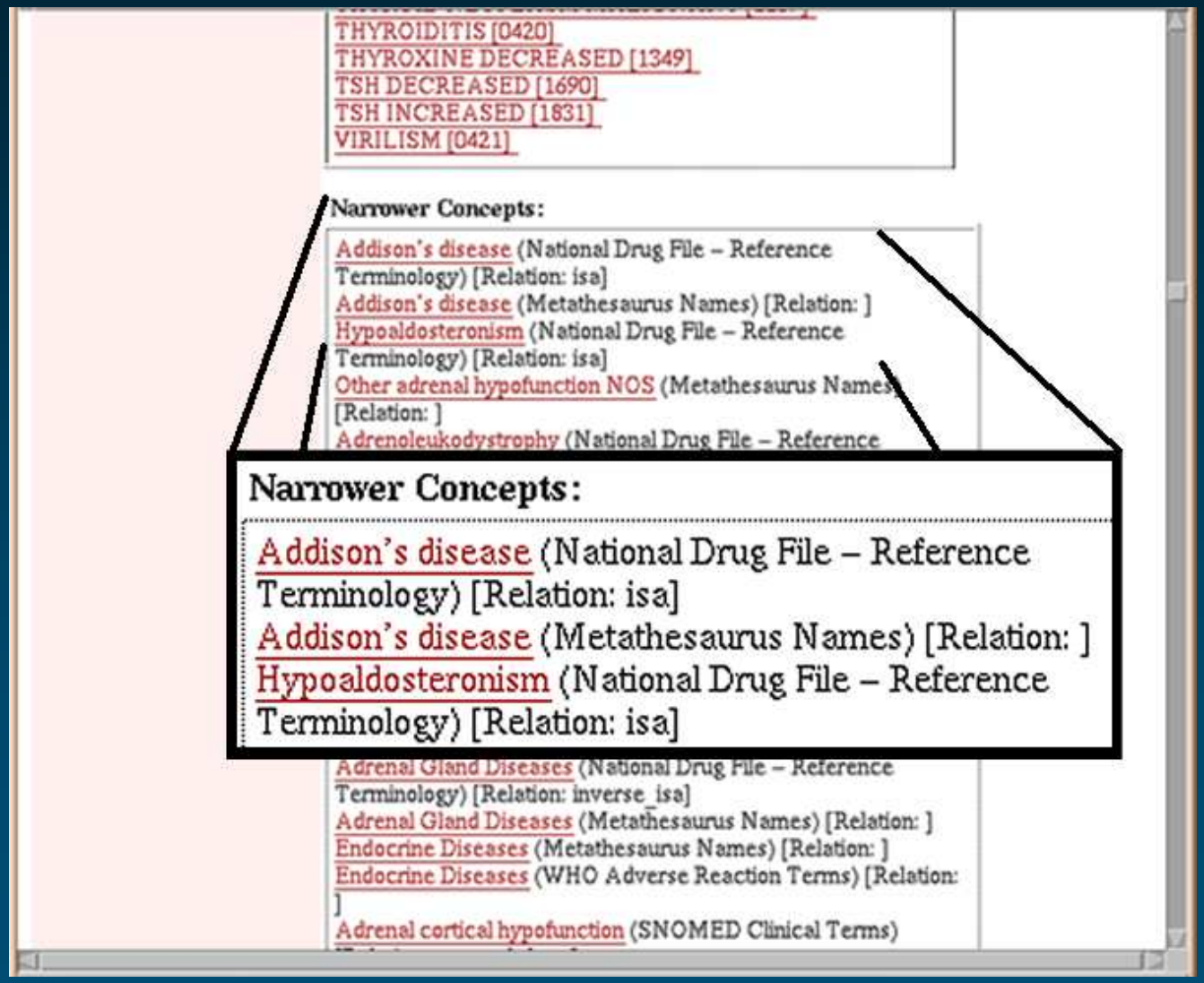
# Concept Report Display All (continued)

Hierarchies



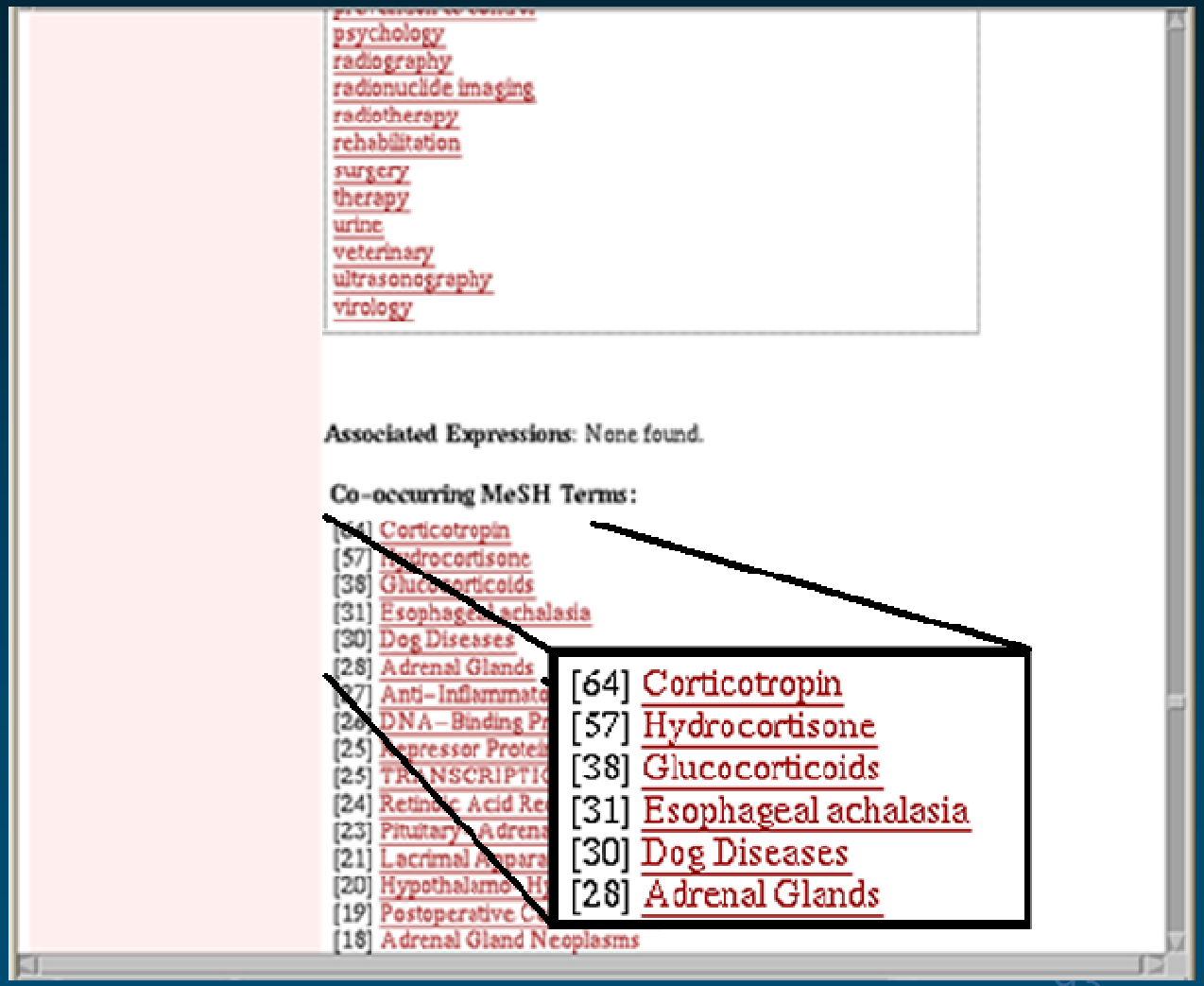
# Concept Report Display All (continued)

Relations



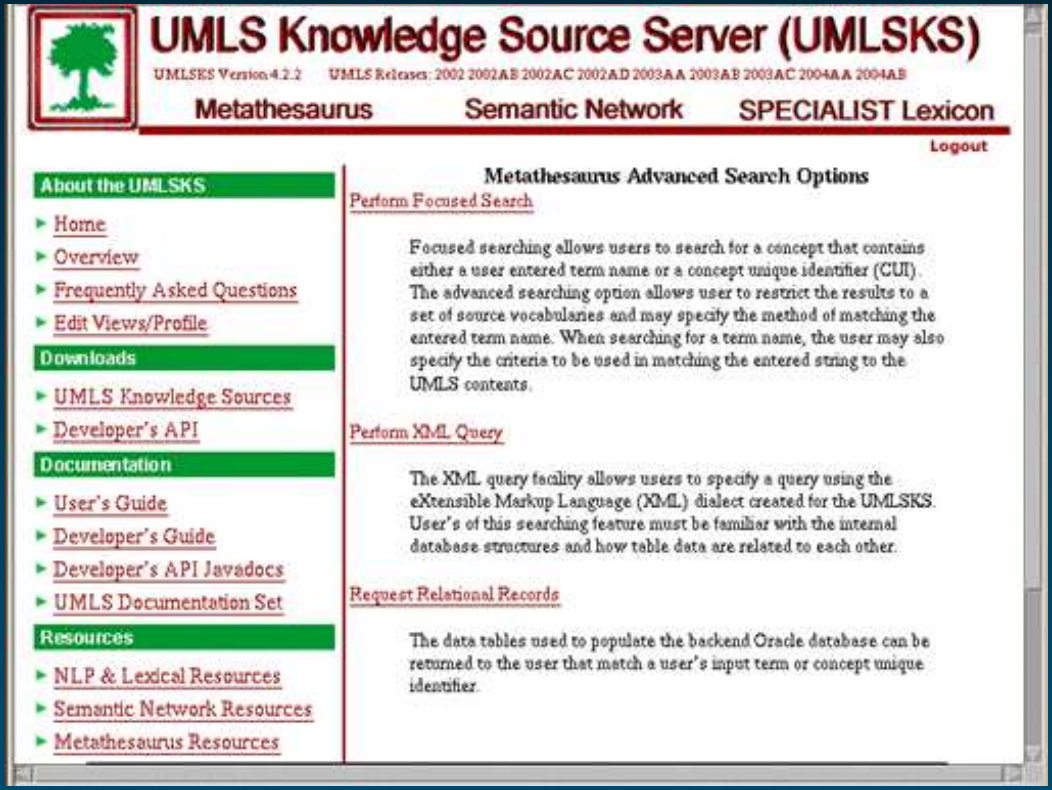
# Concept Report Display All (continued)

Co-occurrence data



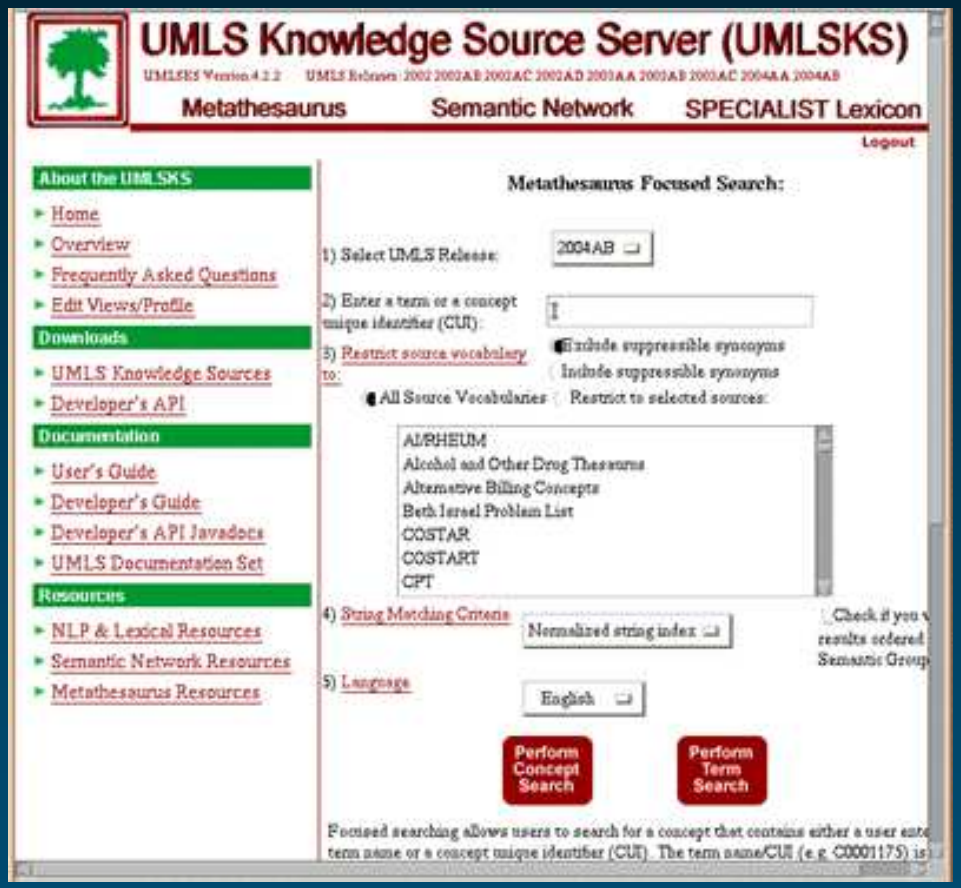
# Metathesaurus Advanced Search Options

- Focused Search
- Raw Relational Records



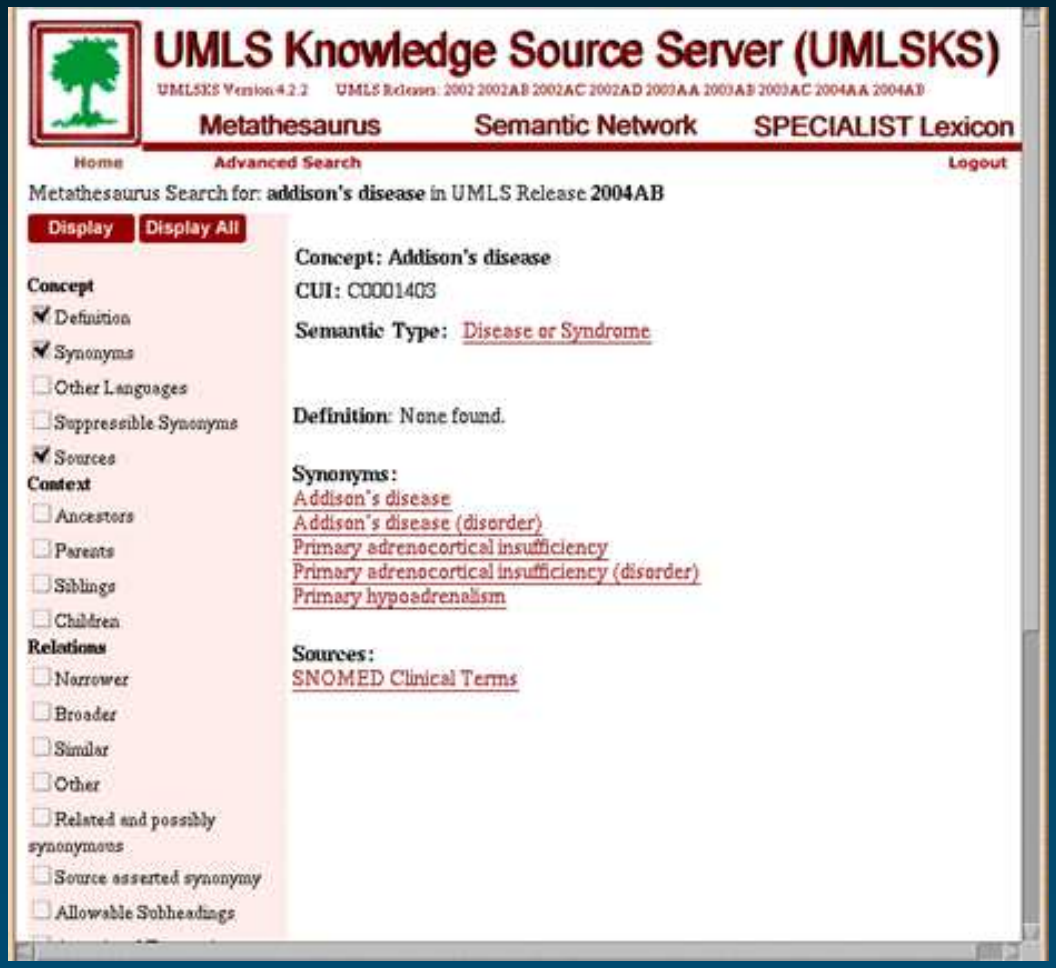
# Metathesaurus Advanced Search Feature

## Focused Search




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

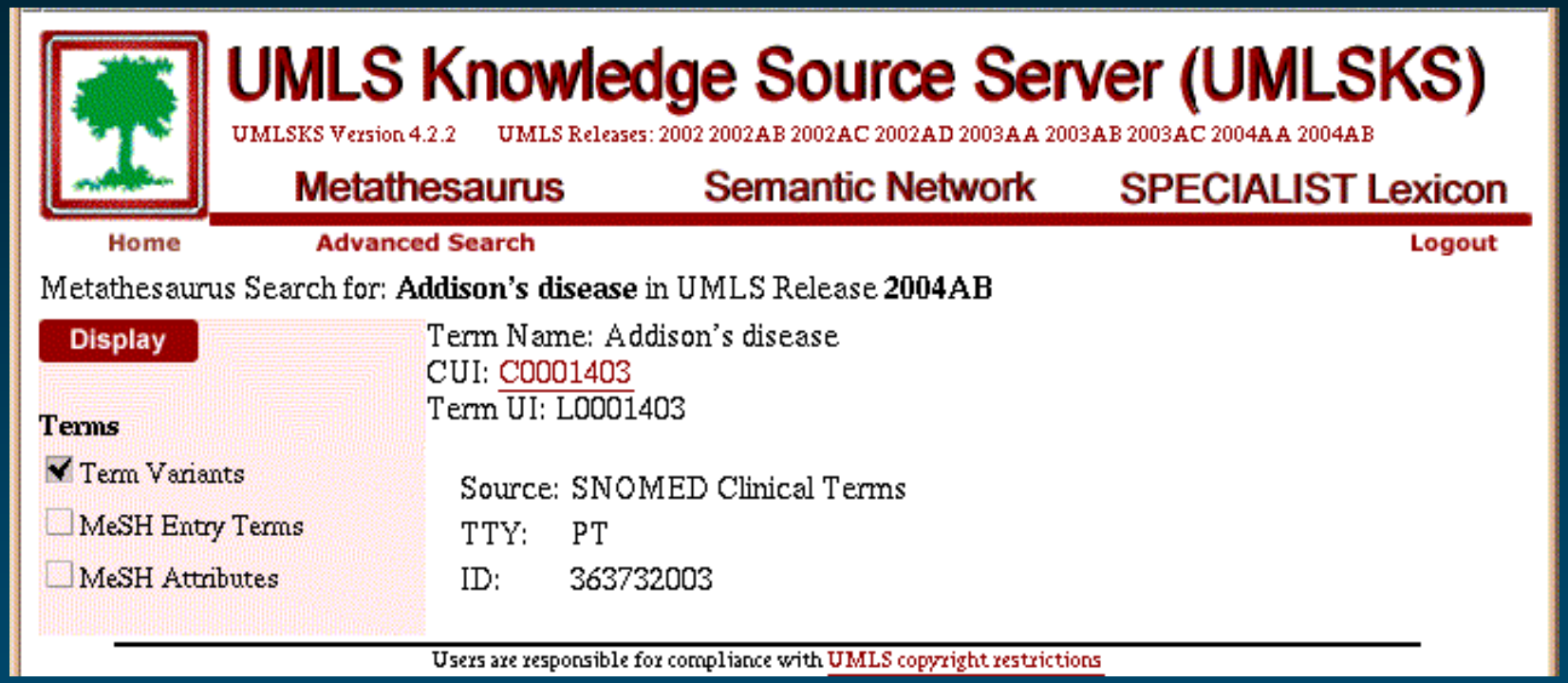
# Restricted Source Concept Report
## *Addison's Disease*




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

NLM

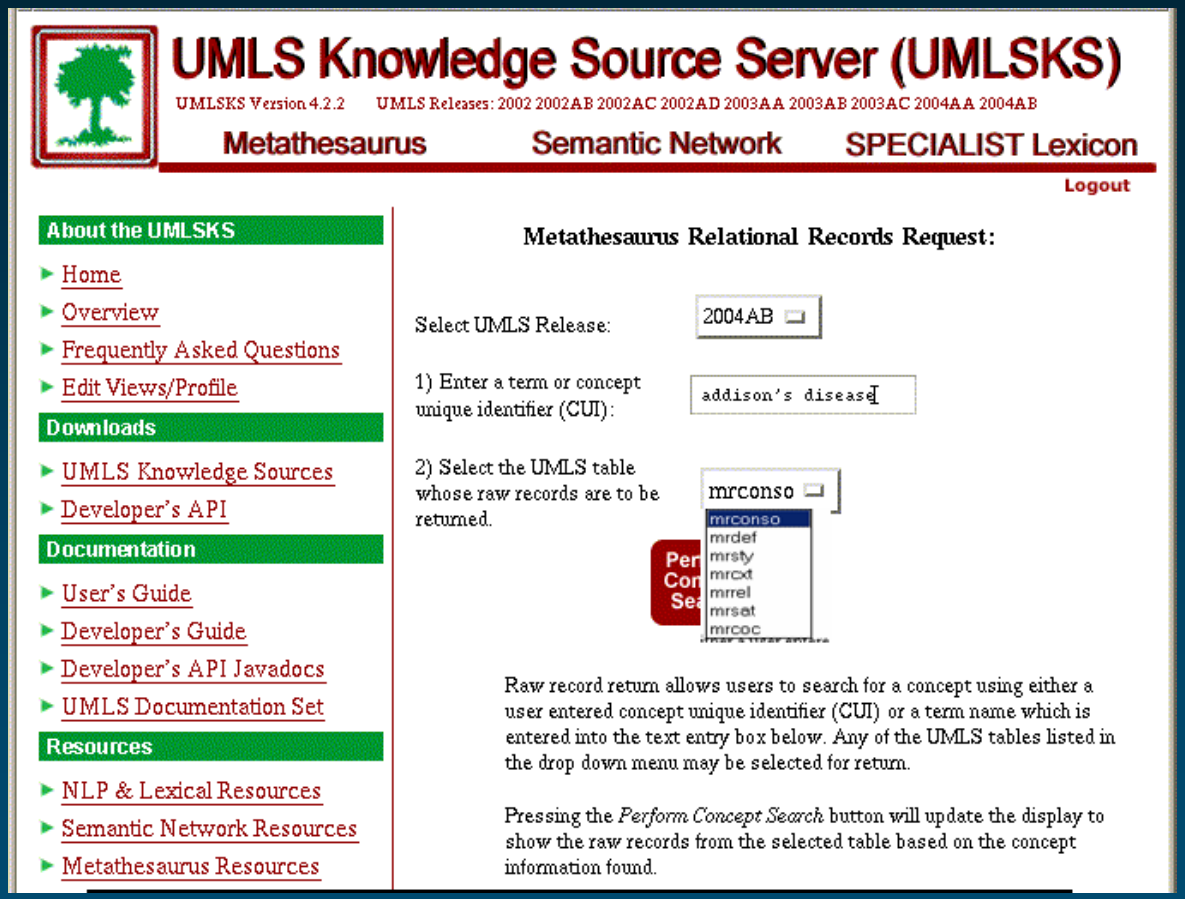
# Addison's disease in SNOMED CT
## Preferred Term and Code




- TTY: Term Type
- ID: Source Code Descriptor

# Metathesaurus Advanced Search Feature
## Relational Record Request

- UMLS Release
- Search Term
- UMLS Relational Table

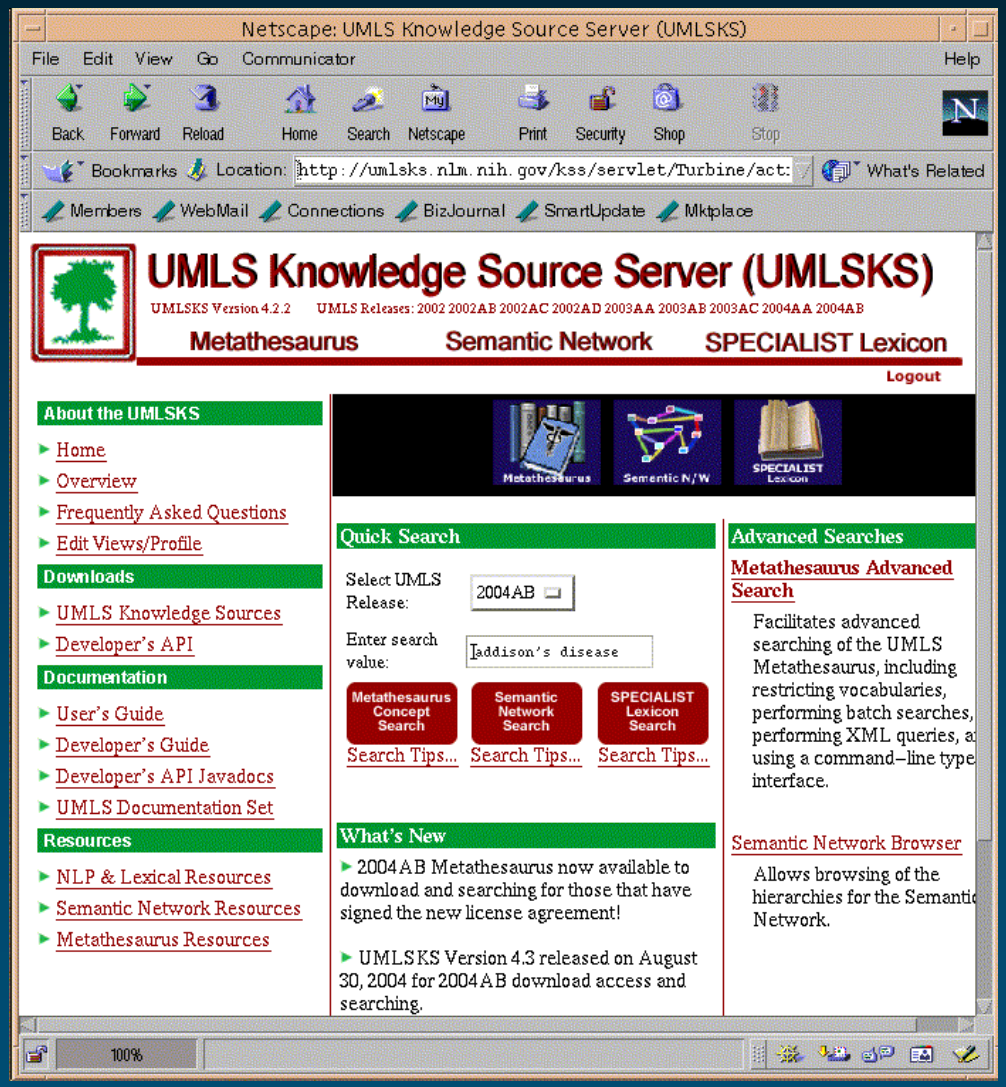


# Relational Records MRCONSO.RRF

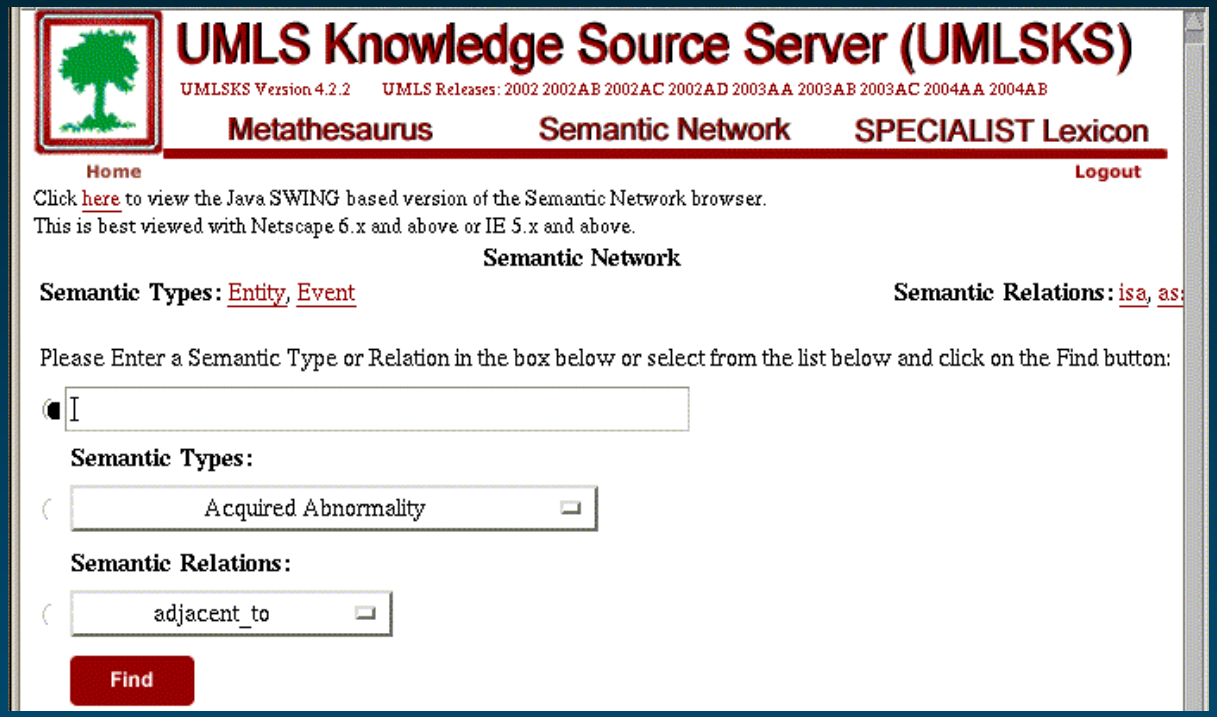
```
CUI|LAT|TS|LUI|STT|SUI|ISPREF|AUI|SAUI|SCUI|SDUI|SAB|TTY|CODE|STR|SUPPRESS|CVF|
C0001403|CZE|P|L3180742|PF|S3708232|Y|A3910108|||D000224|MSHCZE|MH|D000224|ADDISONOVA NEMOC|
C0001403|DUT|P|L2048638|PF|S2386860|N|A6566810|||10001130|MDRDUT|LT|10001130|Addison, ziekte
C0001403|DUT|P|L2048638|PF|S2386860|Y|A3931189|||D000224|MSHDUT|MH|D000224|Addison, 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 Addiso
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||||AOD|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



# Semantic Network Search




- Enter search string

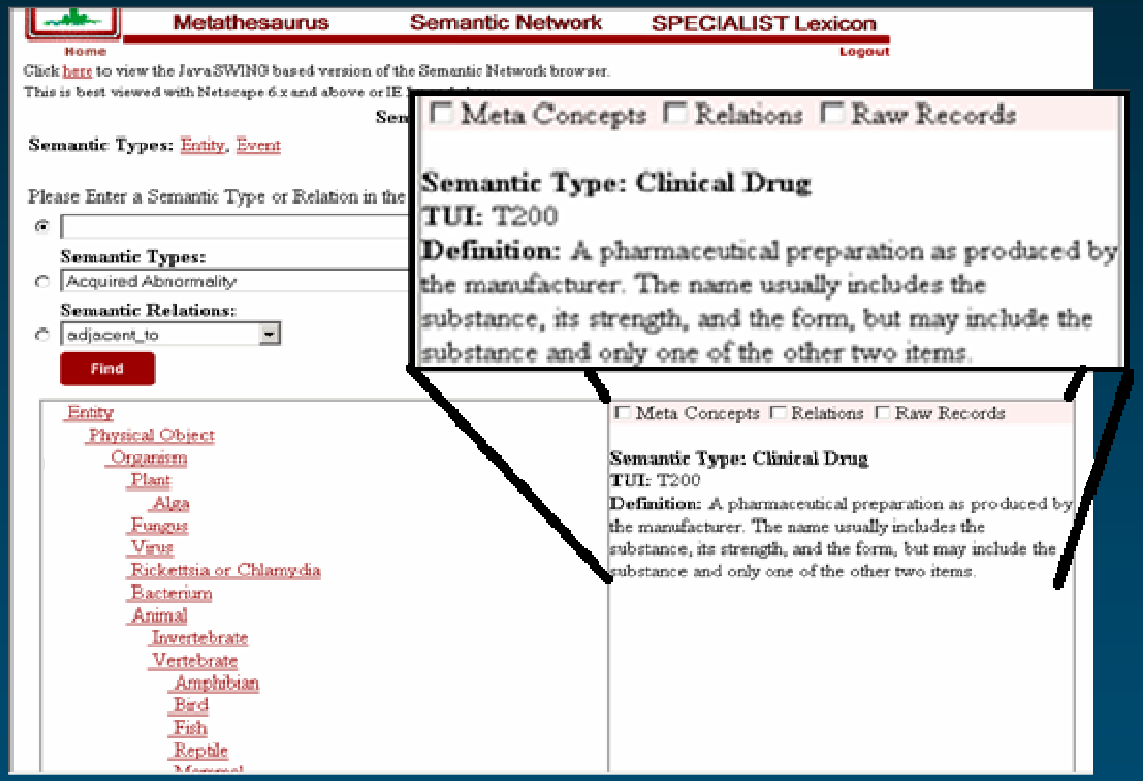
  *-or-*

- Select semantic type

  *-or-*

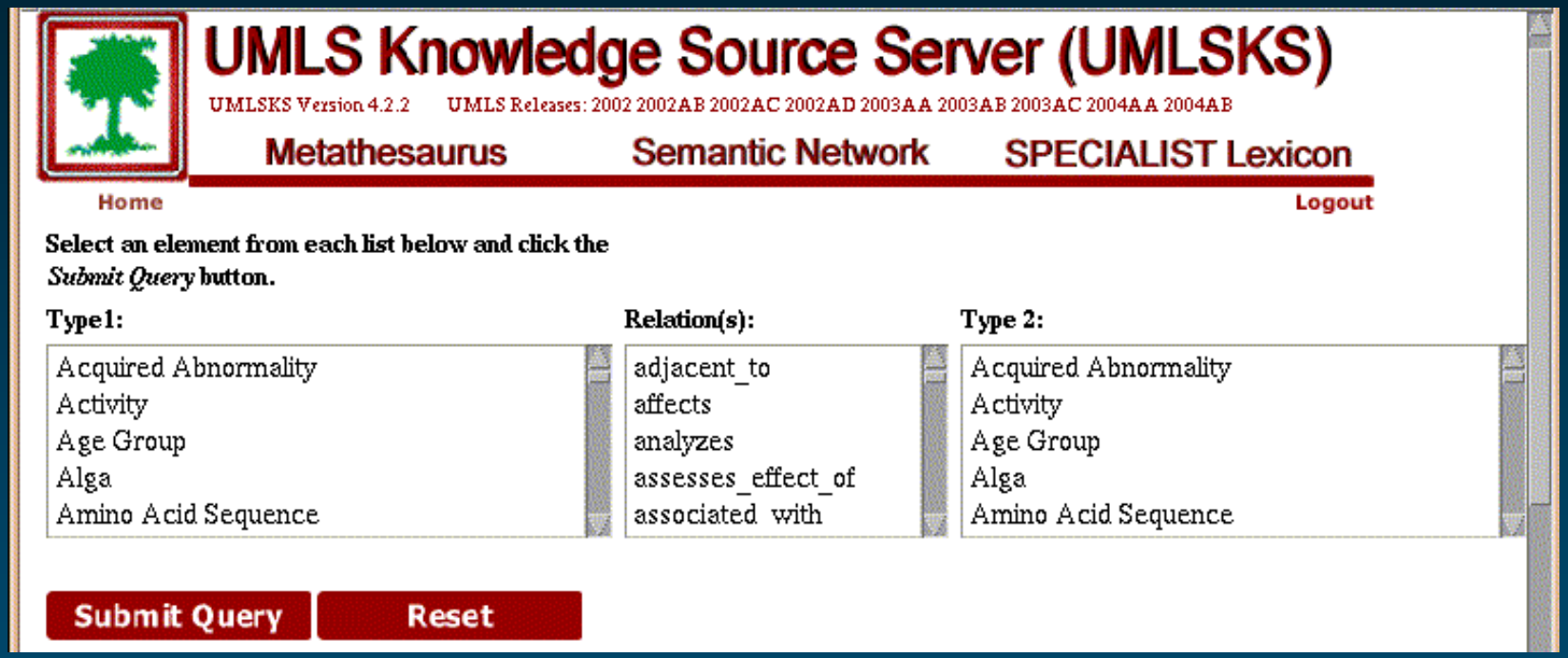
- Select semantic relation

# Semantic Type Clinical Drug




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

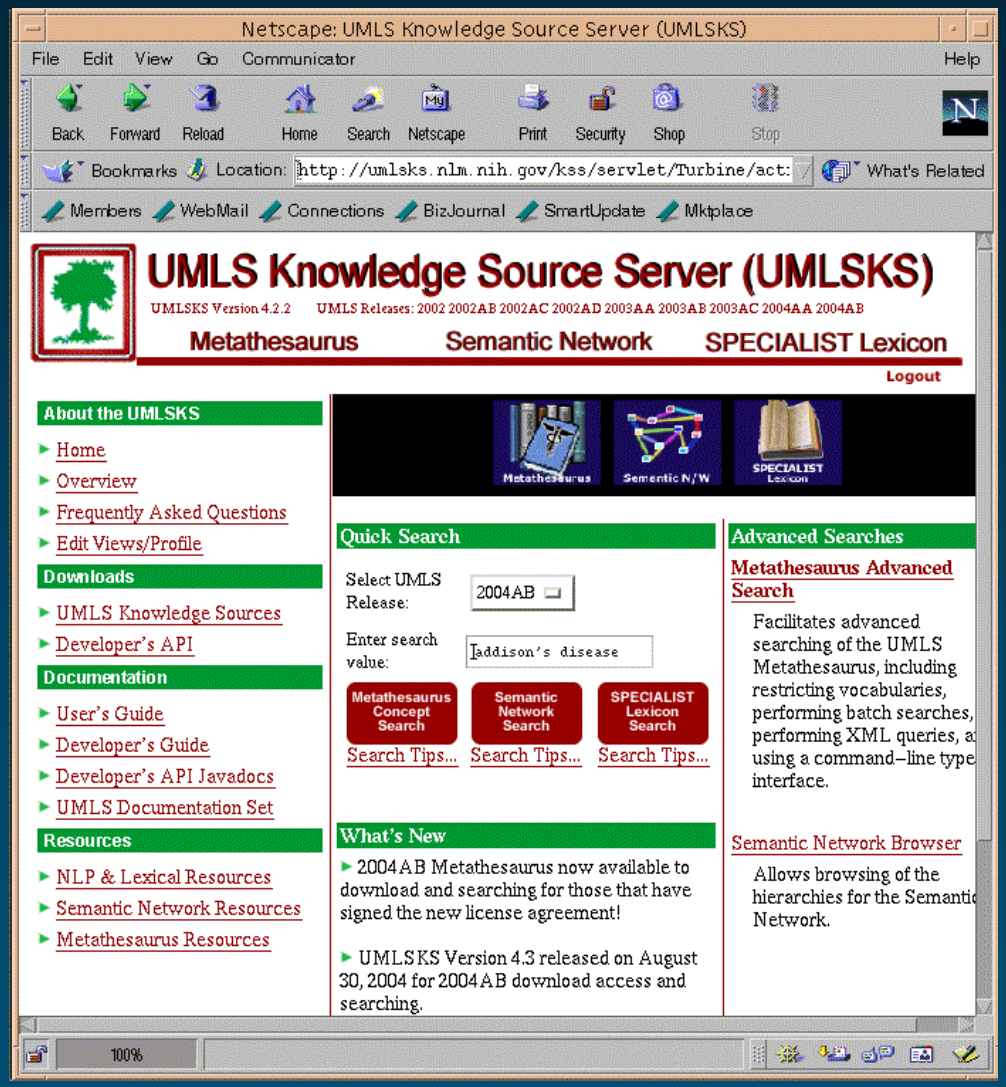
# Show Relations Between Types



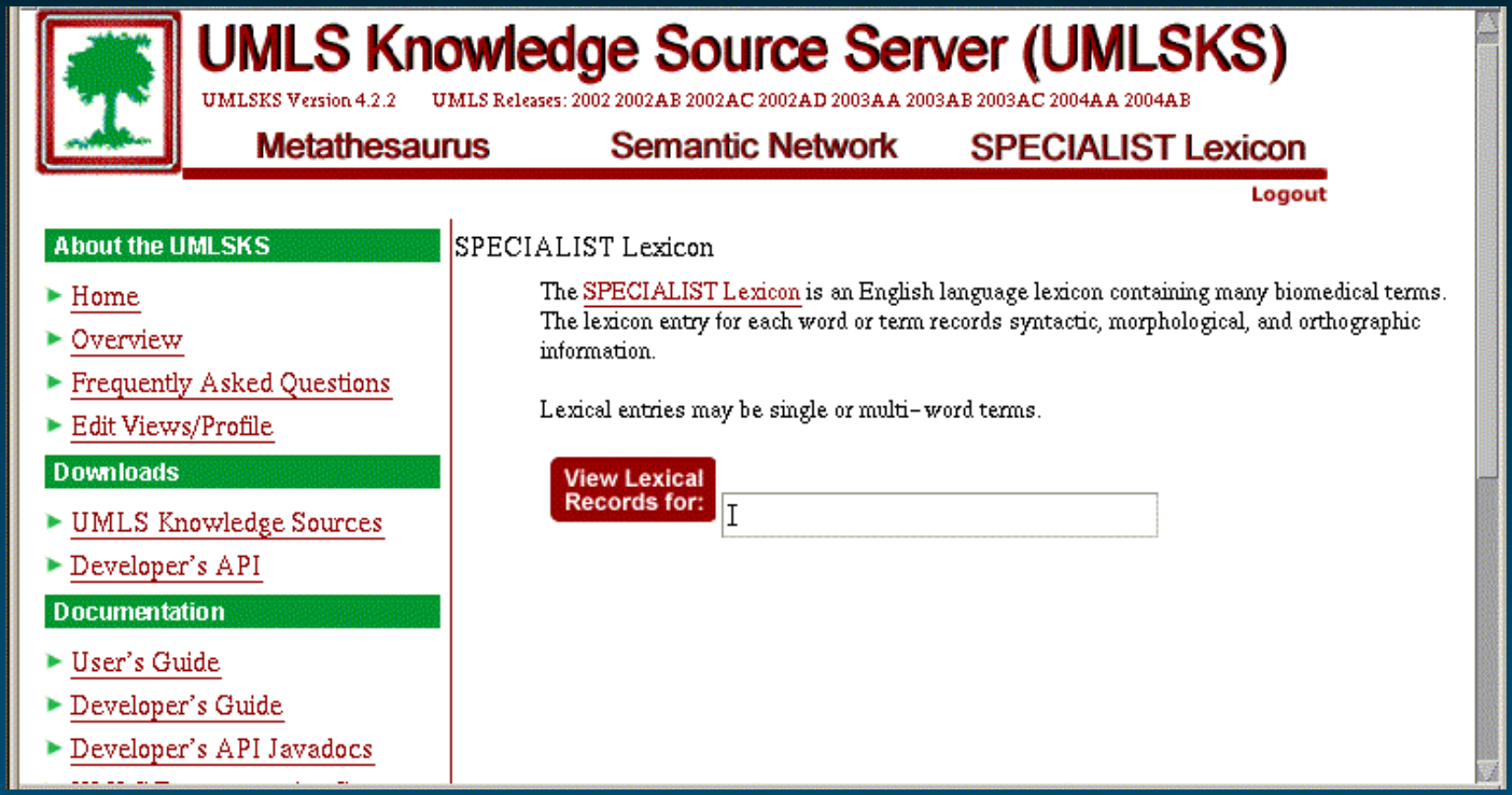

- Validates whether a selected Semantic Relationship (SR) holds between two selected Semantic Types (ST)

# SPECIALIST Lexicon Searching

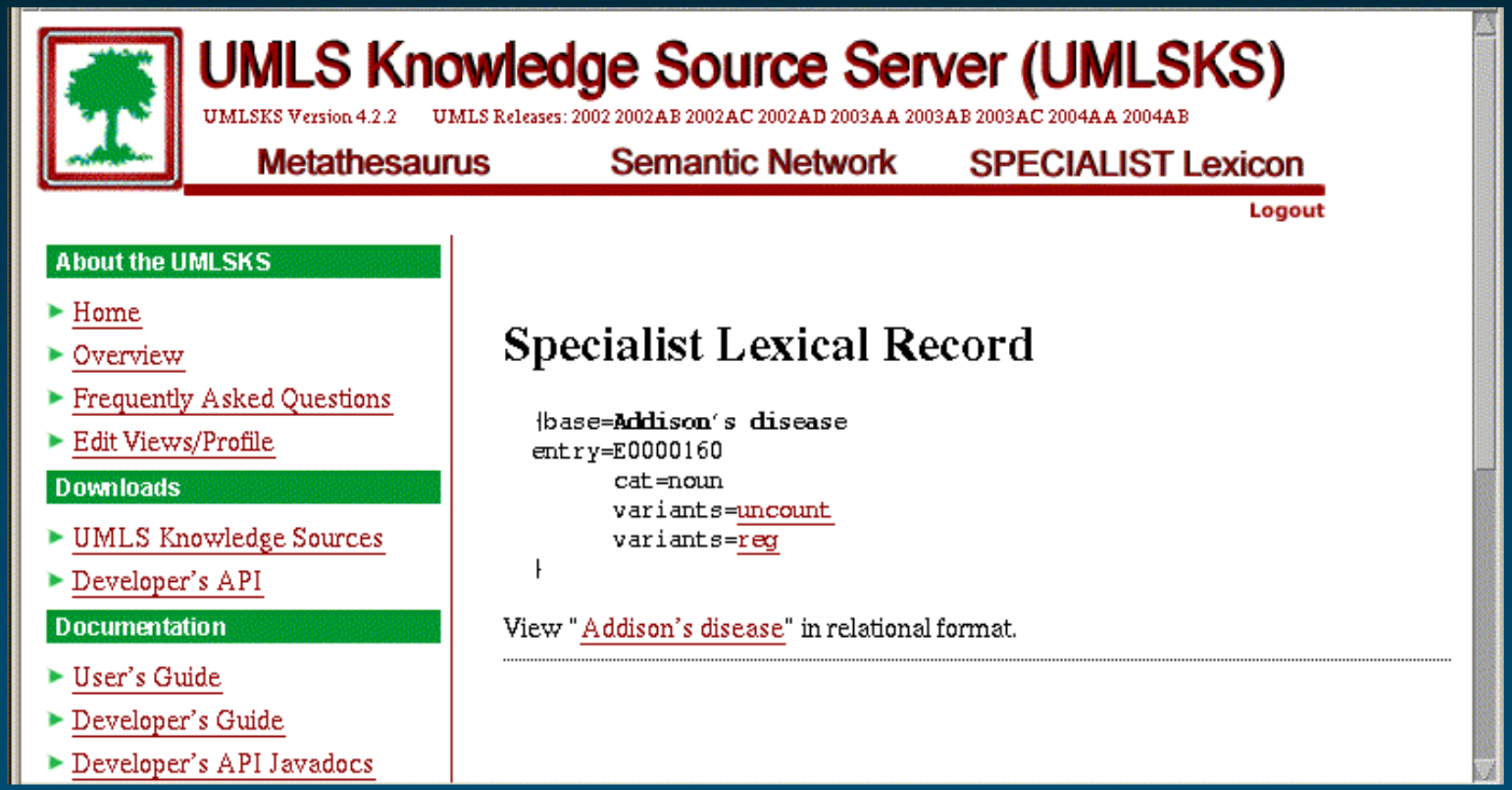
- Select Tab along top
- Quick search



# SPECIALIST Lexicon Search

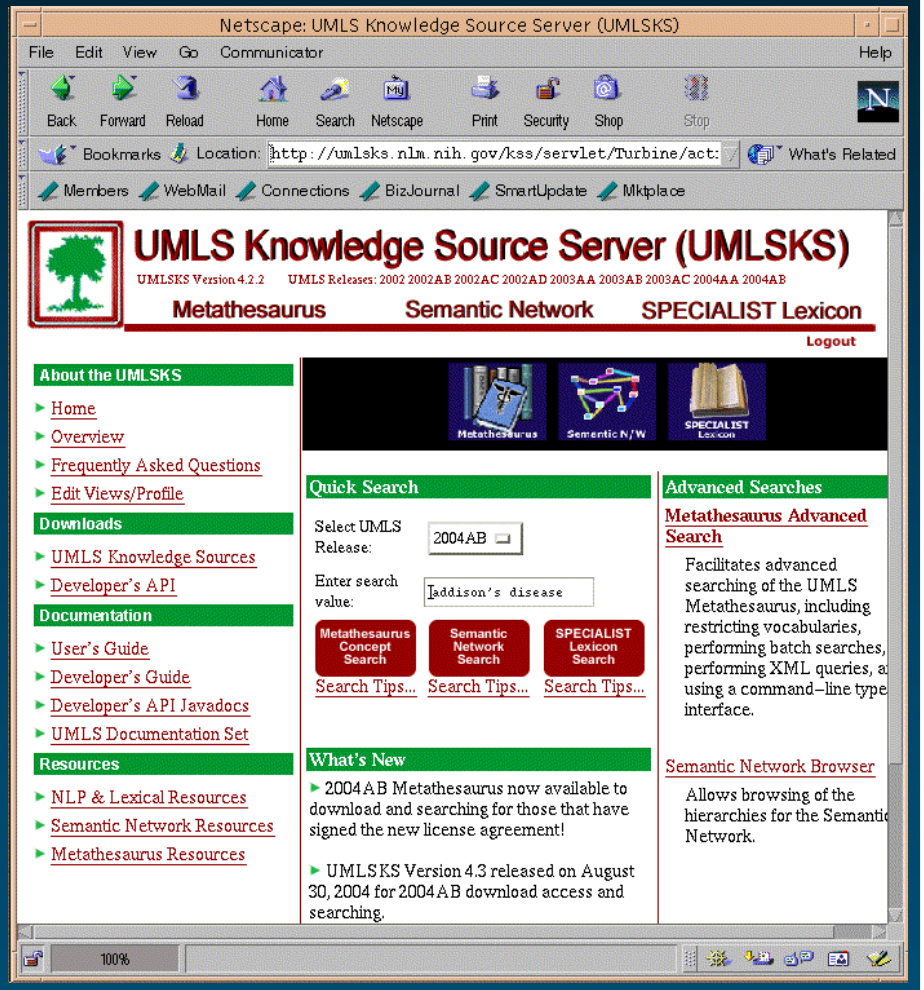


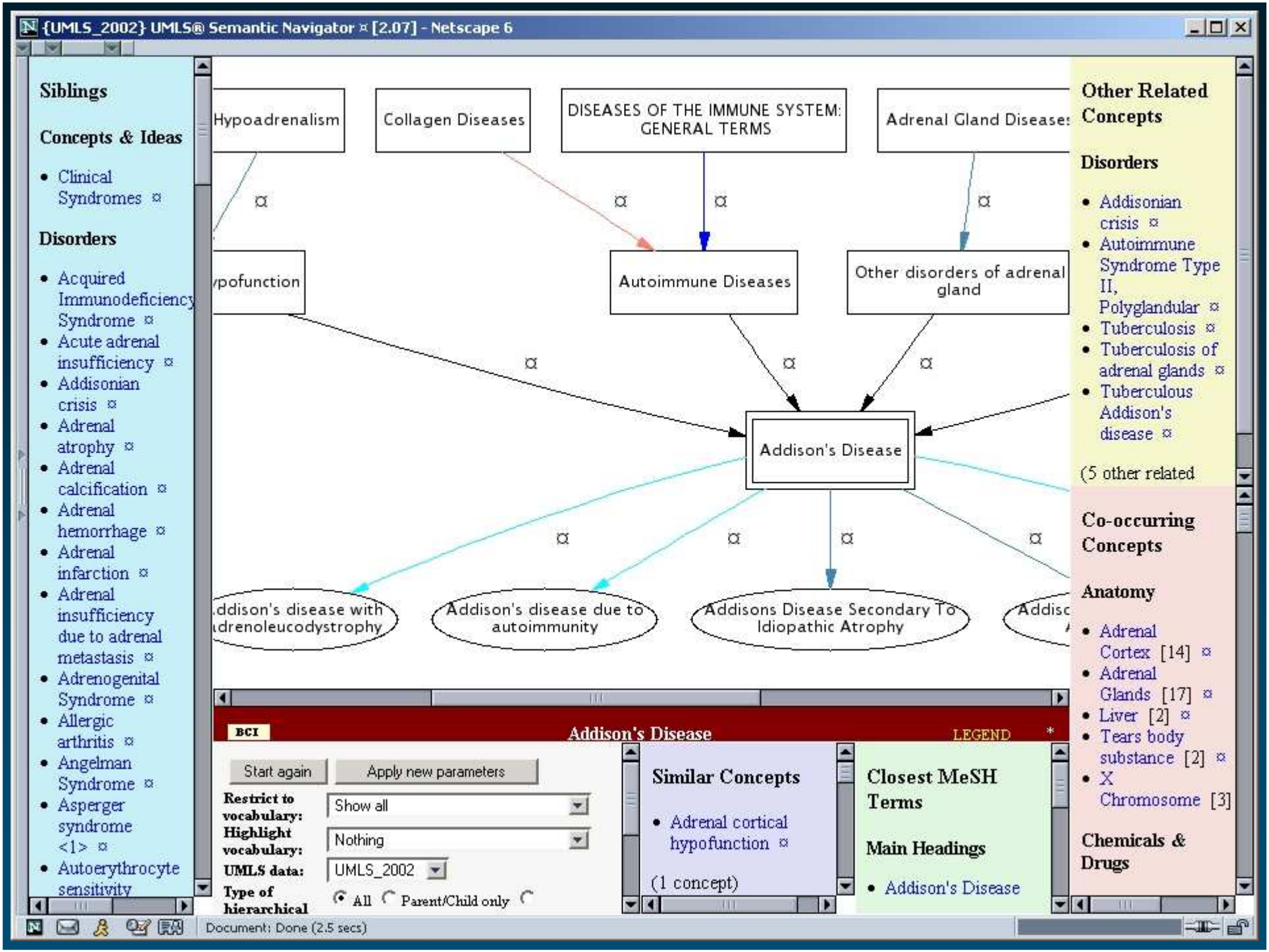
# SPECIALIST Lexical Record



# UMLS Resources

- NLP & Lexical Resources
  - MetaMap Transfer (MMTx)
  - Word Sense Disambiguation (WSD) Test Collection
- Semantic Network
  - Semantic Navigator
  - Semantic Groups
- Metathesaurus
  - String Properties





{UMLS_2002} UMLS® Semantic Navigator ¤ [2.07] - Netscape 6
Siblings
Concepts & Ideas
Clinical Syndromes ¤
Disorders
Acquired Immunodeficiency Syndrome ¤
Acute adrenal insufficiency ¤
Addisonian crisis ¤
Adrenal atrophy ¤
Adrenal calcification ¤
Adrenal hemorrhage ¤
Adrenal infarction ¤
Adrenal insufficiency due to adrenal metastasis ¤
Adrenogenital Syndrome ¤
Allergic arthritis ¤
Angelman Syndrome ¤
Asperger syndrome <1> ¤
Autoerythrocyte sensitivity
Hypoadrenalism
Collagen Diseases
DISEASES OF THE IMMUNE SYSTEM: GENERAL TERMS
Adrenal Gland Diseases
ypofunction
Autoimmune Diseases
Other disorders of adrenal gland
Addison's Disease
ddison's disease with drenoleucodystrophy
Addison's disease due to autoimmunity
Addisons Disease Secondary To Idiopathic Atrophy
Addisc
Other Related Concepts
Disorders
Addisonian crisis ¤
Autoimmune Syndrome Type II, Polyglandular ¤
Tuberculosis ¤
Tuberculosis of adrenal glands ¤
Tuberculous Addison's disease ¤
(5 other related
Co-occurring Concepts
Anatomy
Adrenal Cortex [14] ¤
Adrenal Glands [17] ¤
Liver [2] ¤
Tears body substance [2] ¤
X Chromosome [3]
Chemicals & Drugs
BCI
Addison's Disease
LEGEND
Start again
Apply new parameters
Restrict to vocabulary:
Show all
Highlight vocabulary:
Nothing
UMLS data:
UMLS_2002
Type of hierarchical
All
Parent/Child only
Similar Concepts
Adrenal cortical hypofunction ¤
(1 concept)
Closest MeSH Terms
Main Headings
Addison's Disease
Document: Done (2.5 secs)

# 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
  - Port: 8042

# Developer's Guide

## Documentation

- User's Guide
- Developer's Guide
  1. Introduction
  2. Installing the UMLSKS
  3. Building UMLSKS Software Applications
  4. Using the XML Query Facility
  5. Using the UMLSKS Socket Server
- UMLS Documentation Set

UMLS Knowledge Sourc

UMLSKS Version 4.2.2 UMLS Releases: 2002 2002AB 2002AC 2002

Lister

**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
  1. Introduction
  2. Installing the UMLSKS
  3. Building UMLSKS Software Applications
  4. Using the XML Query Facility
  5. Using the UMLSKS Socket Server
- UMLS Documentation Set

**Resources**

- NLP & Lexical Resources
- Semantic Network Resources

Print/Text Version Table o

This guide describes the installati
Knowledge Source Server (UMLSI

***Audience***

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

***Release Notes***

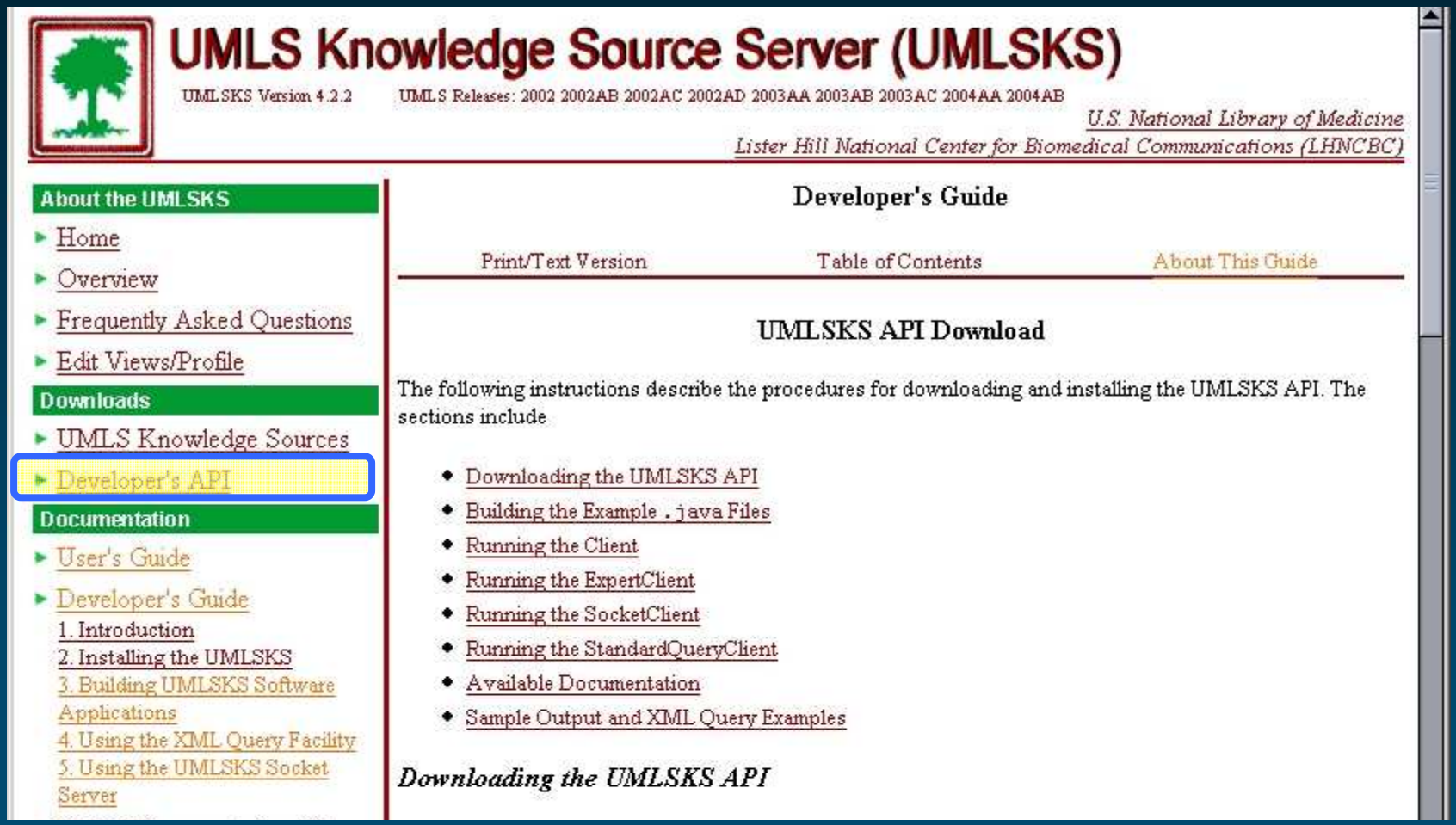
Please refer to the Release Bulletin for a detailed list of features, bug fixes, and known problems with this version of the UMLSKS.

***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

# Documentation Java API



UMLS Knowledge Source Server (UMLSKS)

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

U.S. National Library of Medicine

Lister Hill National Center for Biomedical Communications (LHNCBC)

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  5. Using the UMLSKS Socket Server

Developer's Guide

Print/Text Version | Table of Contents | About This Guide

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*

# Documentation Javadocs

All Classes

Packages

gov.nih.nlm.kss.api
gov.nih.nlm.kss.example
gov.nih.nlm.kss.models
gov.nih.nlm.kss.models.lex
gov.nih.nlm.kss.models.meta
gov.nih.nlm.kss.models.meta.ass
gov.nih.nlm.kss.models.meta.attr
gov.nih.nlm.kss.models.meta.cor
gov.nih.nlm.kss.models.meta.cor
gov.nih.nlm.kss.models.meta.coo
gov.nih.nlm.kss.models.meta.del

All Classes

AdjEntry
AdvEntry
AssocExprsExecutor
AssociatedExp
AssociatedExpVector
AssociativeRelation
AssociativeRelationVector
AssociativeRelExistence
Attr
AttributeContext
AttributeValue
AttrVector
AuxEntry
BasicConceptPropsExecutor
CatEntry
Client
ClientV2_1
ClientV3_0

Overview Package Class Use Tree Deprecated Index Hel

PREV NEXT FRAMES NO FRAMES

Packages

| | |
|---|---|
| gov.nih.nlm.kss.api | |
| gov.nih.nlm.kss.example | |
| gov.nih.nlm.kss.models | |
| gov.nih.nlm.kss.models.lex | |
| gov.nih.nlm.kss.models.meta | |
| gov.nih.nlm.kss.models.meta.assocExp | |
| gov.nih.nlm.kss.models.meta.attribute | |
| gov.nih.nlm.kss.models.meta.concept | |
| gov.nih.nlm.kss.models.meta.context | |
| gov.nih.nlm.kss.models.meta.cooccurrence | |
| gov.nih.nlm.kss.models.meta.deltas | |
| gov.nih.nlm.kss.models.meta.locator | |
| gov.nih.nlm.kss.models.meta.meshentry | |
| gov.nih.nlm.kss.models.meta.relation | |
| gov.nih.nlm.kss.models.meta.source | |
| gov.nih.nlm.kss.models.sem | |
| gov.nih.nlm.kss.models.sem.rels | |
| gov.nih.nlm.kss.models.sem.units | |
| gov.nih.nlm.kss.query | |

NLM

# Sample XML query (1) Current version

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

```
<?xml version="1.0"?>
<CurrentUMLSYear version="1.0">
      2004AB
</CurrentUMLSYear>
```

# Sample XML query (2) Concepts by string

```
<?xml version="1.0"?>
<findCUI version="1.0">
<conceptName>appendicectomy</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>
```

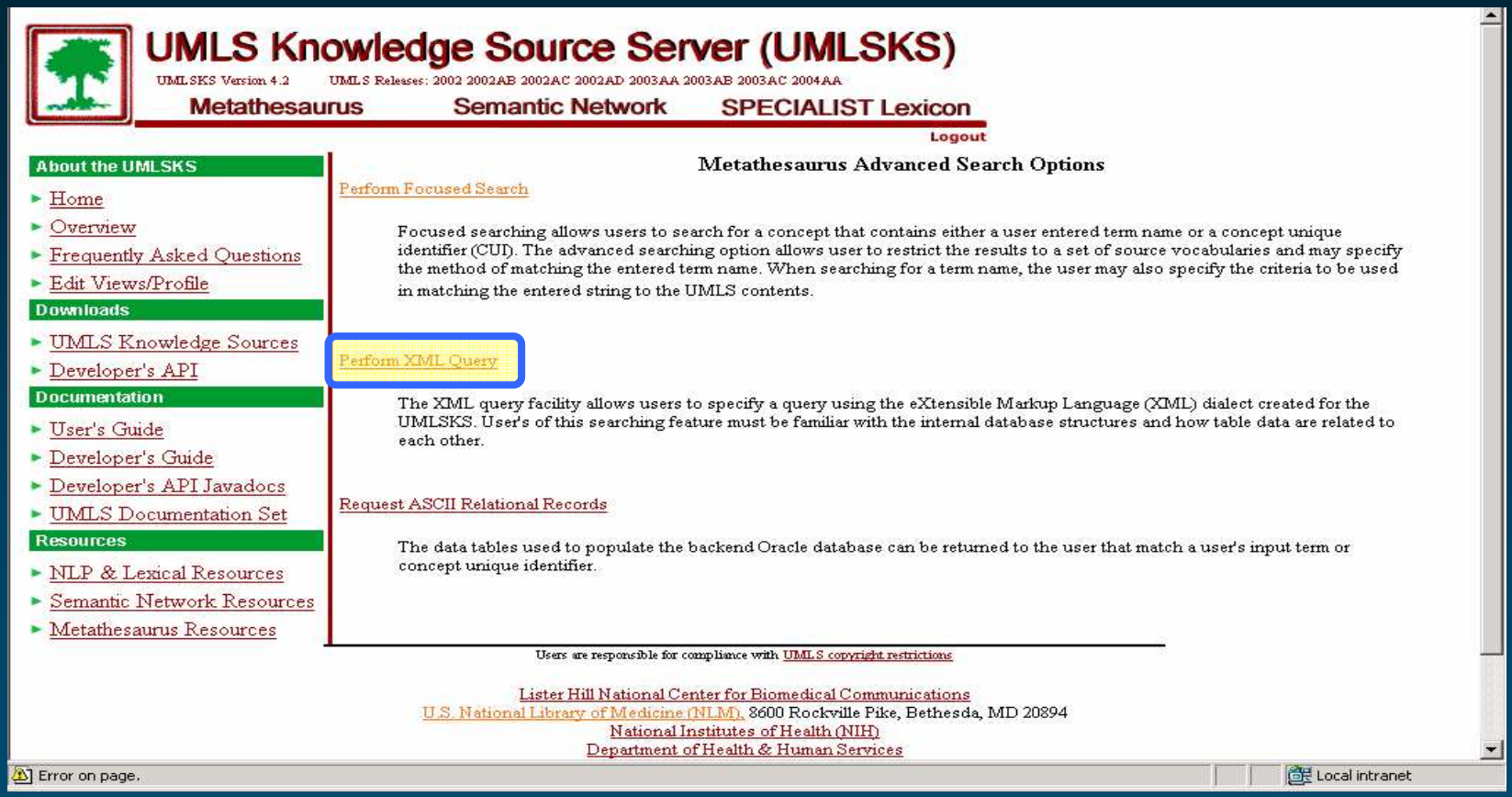
```
<?xml version="1.0"?>
<RelationCollection version="1.0">
[…]
      <relation>
         <rel>RO</rel>
         <cui2>C0005001</cui2>
         <cn2>Benign prostatic hyperplasia</cn2>
         <rela>has_finding_site</rela>
         <sab>SNOMEDCT</sab>
         <sl>SNOMEDCT</sl>
</relation>
[…]
```

# Sample XML query (5) All semantic type IDs

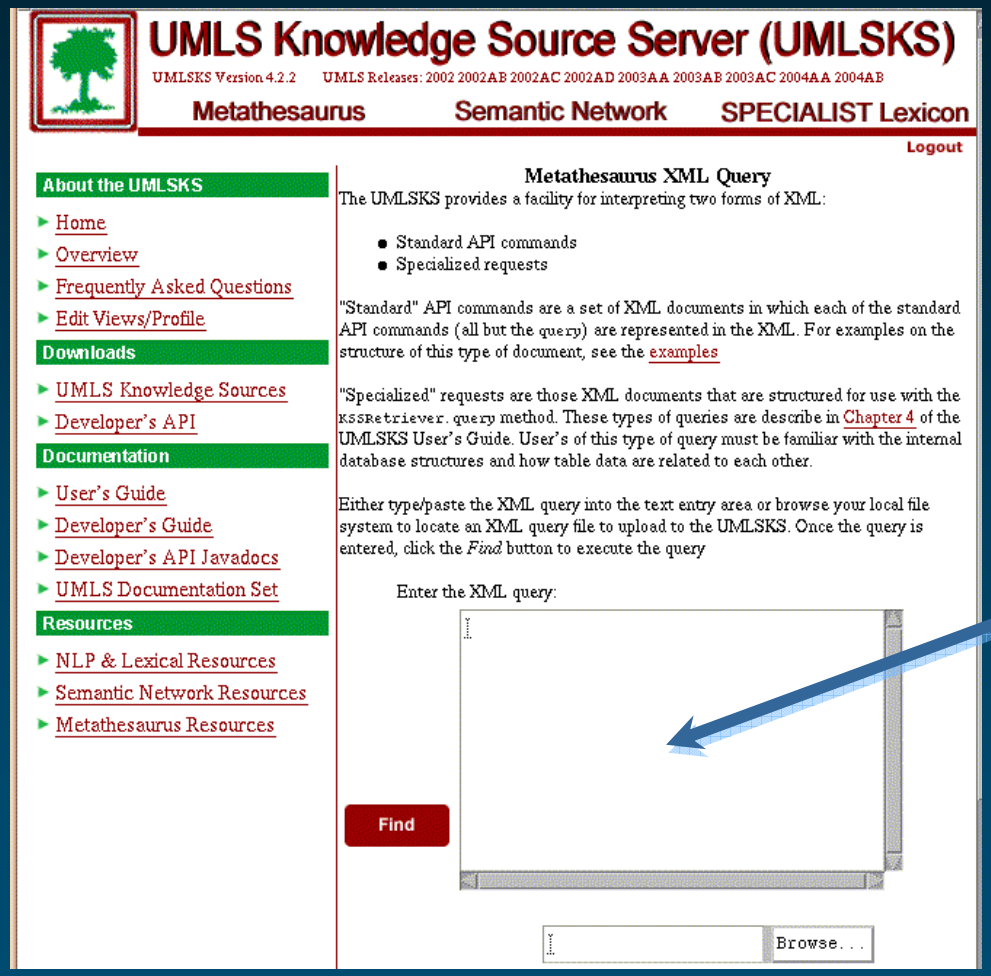
```
<?xml version="1.0"?>
<listSemTypeIds version="1.0">
</listSemTypeIds>
```

```
<?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



# Performing XML queries from UMLSKS




```
<?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
- 2004AB UMLS Files

  2004AB.CHK
  2004AB.MD5
  2004ab-1-meta.nlm
  2004ab-2-meta.nlm
  2004ab-3-meta.nlm
  mmsys.zip
  Copyright_Notice.txt
  README.txt

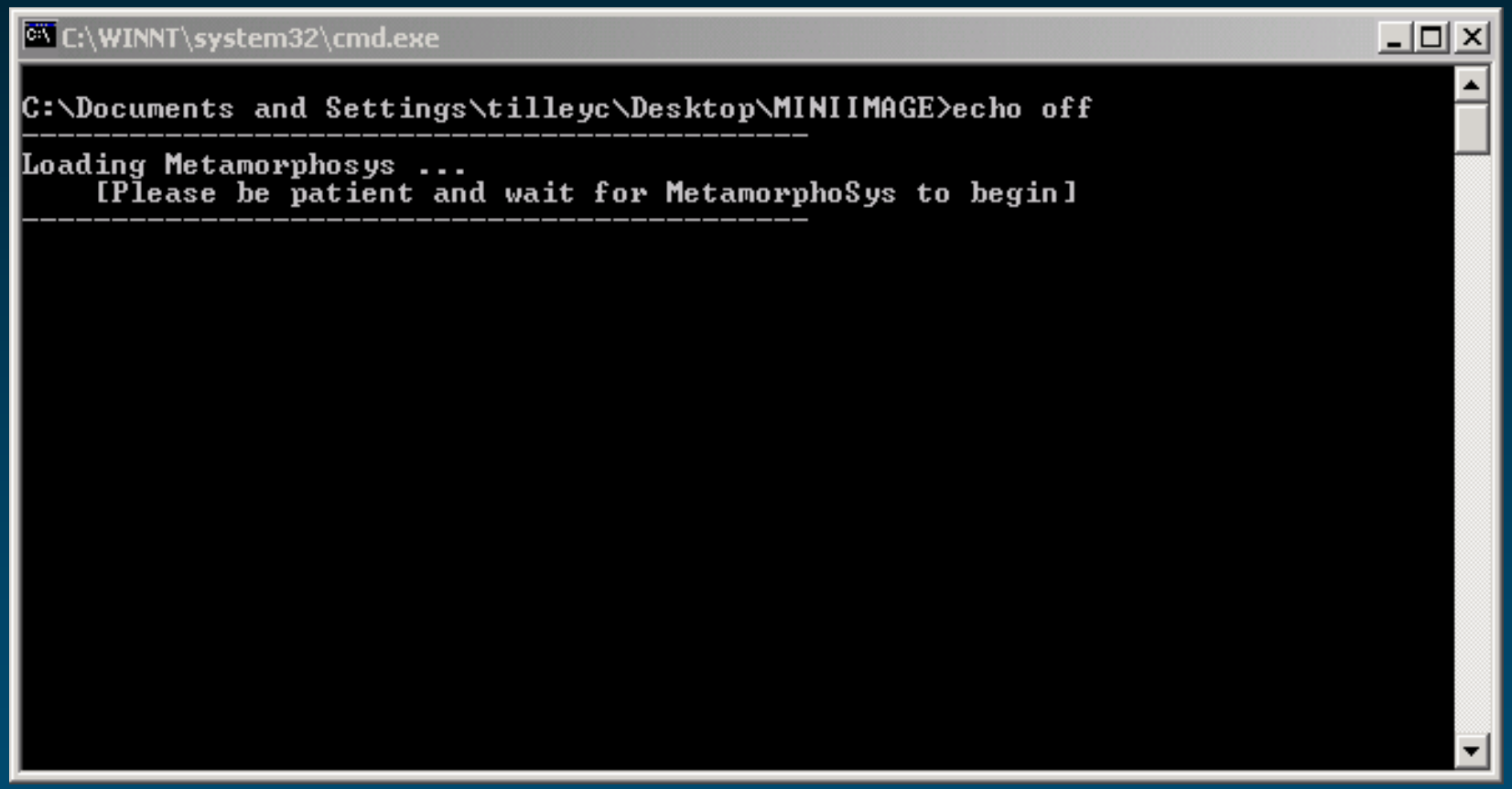
Please README!

# ...or DVD?

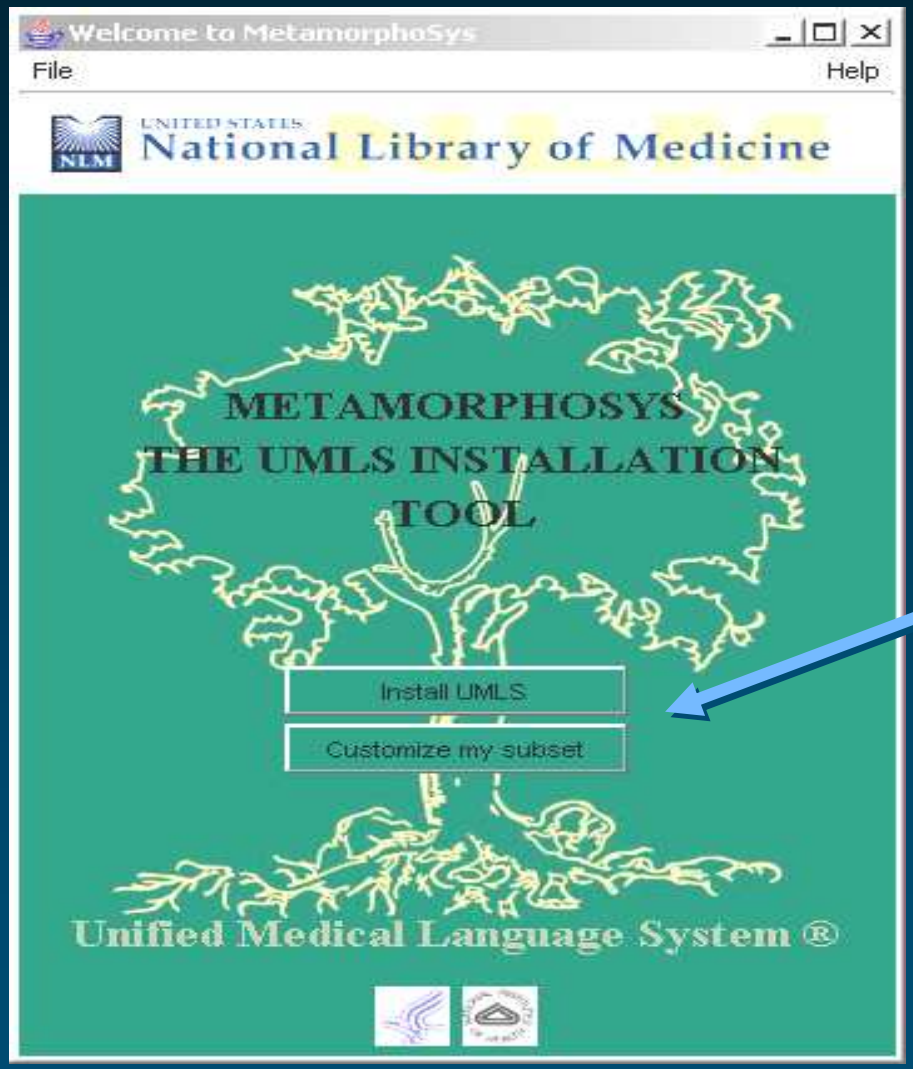
- Order at: umls_support@nlm.nih.gov
- **Include your license number**

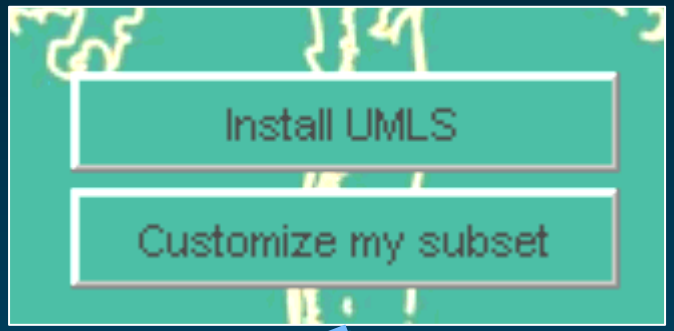
- Run MetamorphoSys from DVD
  - Windows
    - Autorun; or go to root directory and click on “windows_mmsys.bat”
  - Linux, Solaris, Macintosh
    - open a terminal window, change to the root directory and type appropriate command: ./linux_mmsys.sh, ./solaris_mmsys.sh, ./macintosh_mmsys.sh

# Be patient! A lot of software must load.

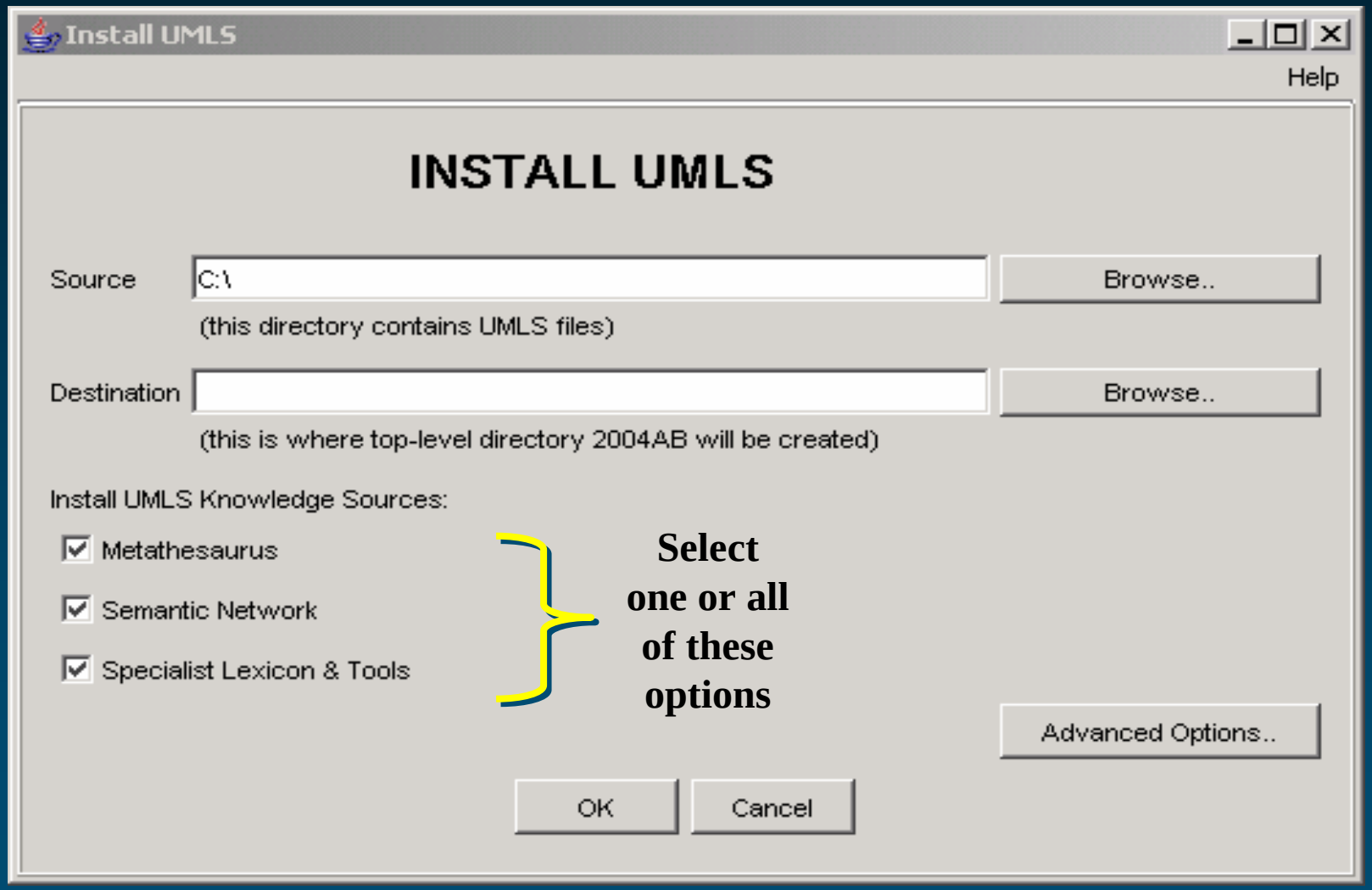


# Welcome Screen



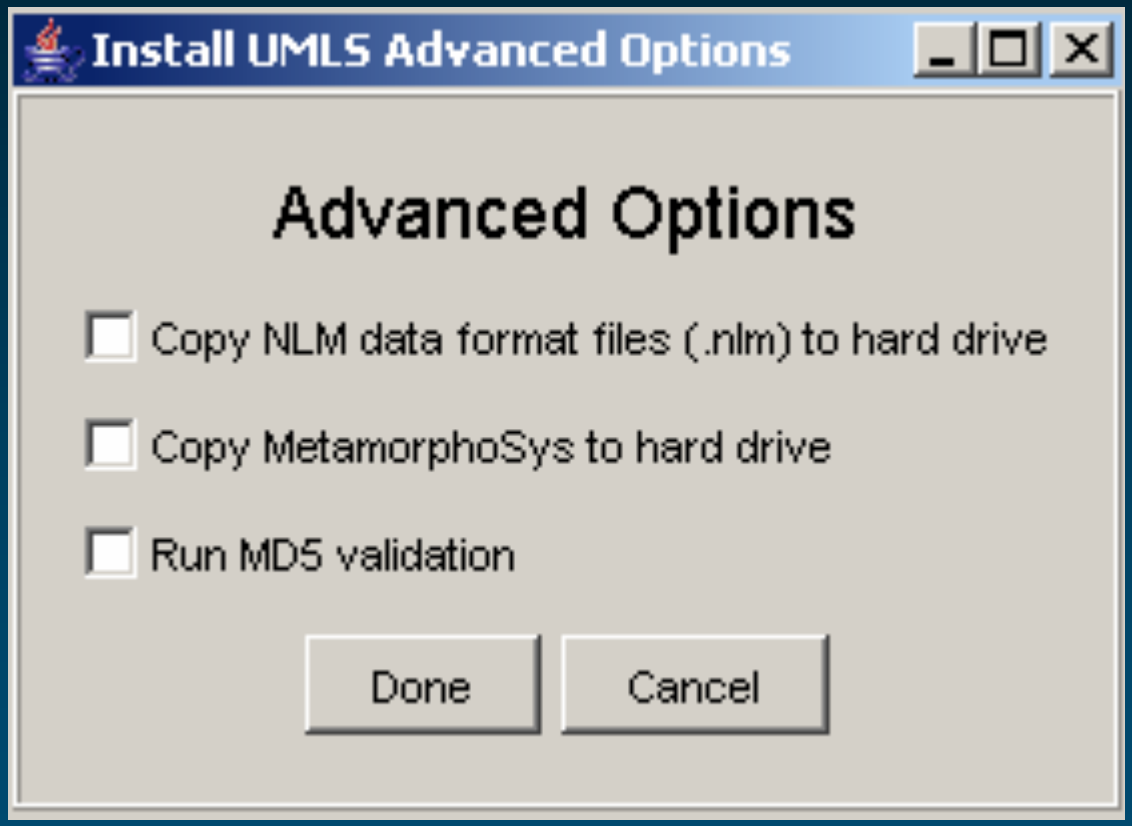



# Install UMLS

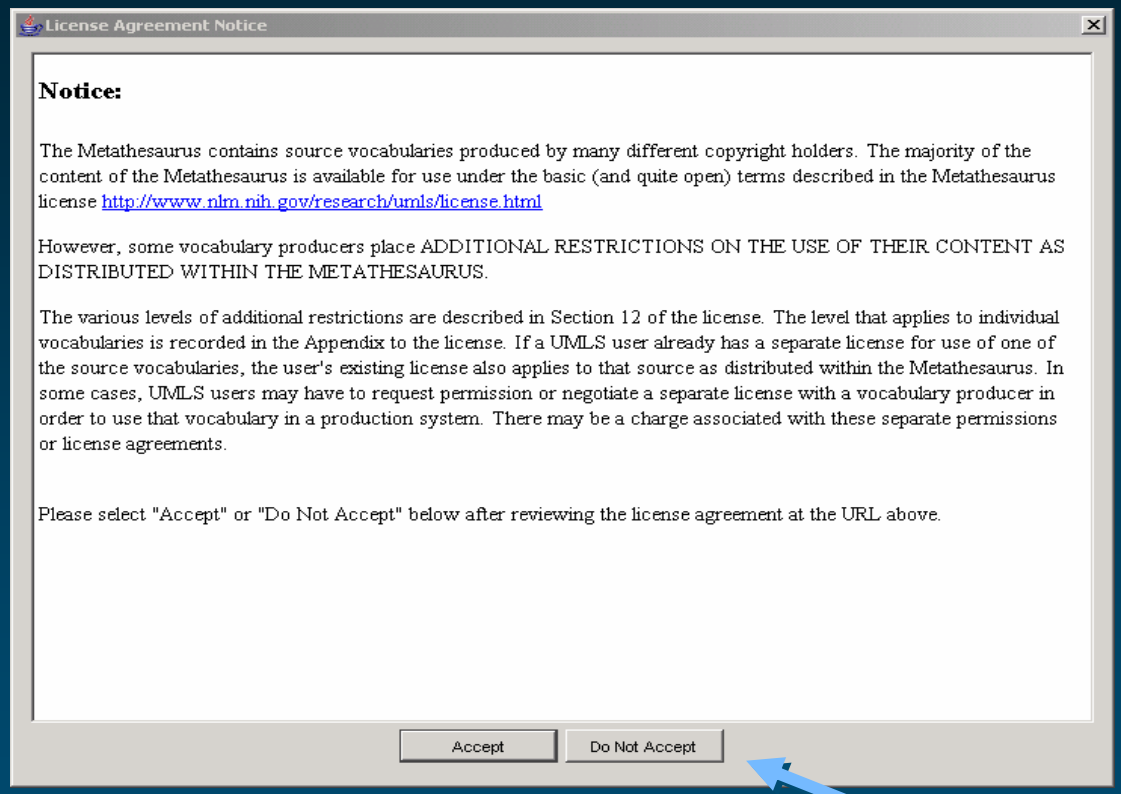


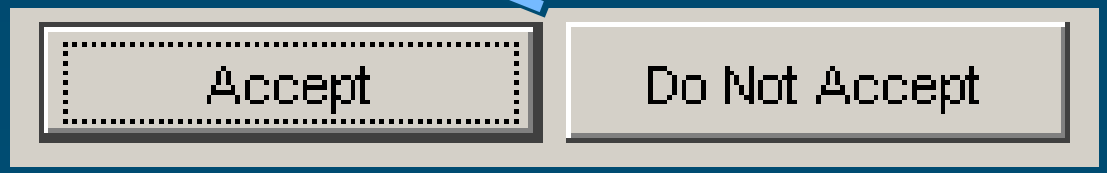

**Select one or all of these options**

# Install UMLS Advanced Options

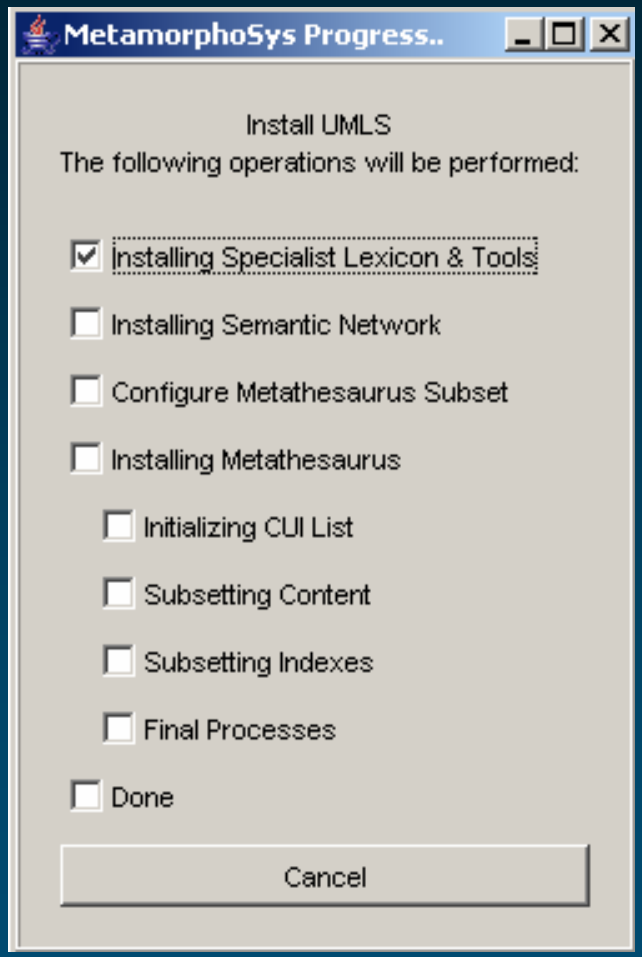


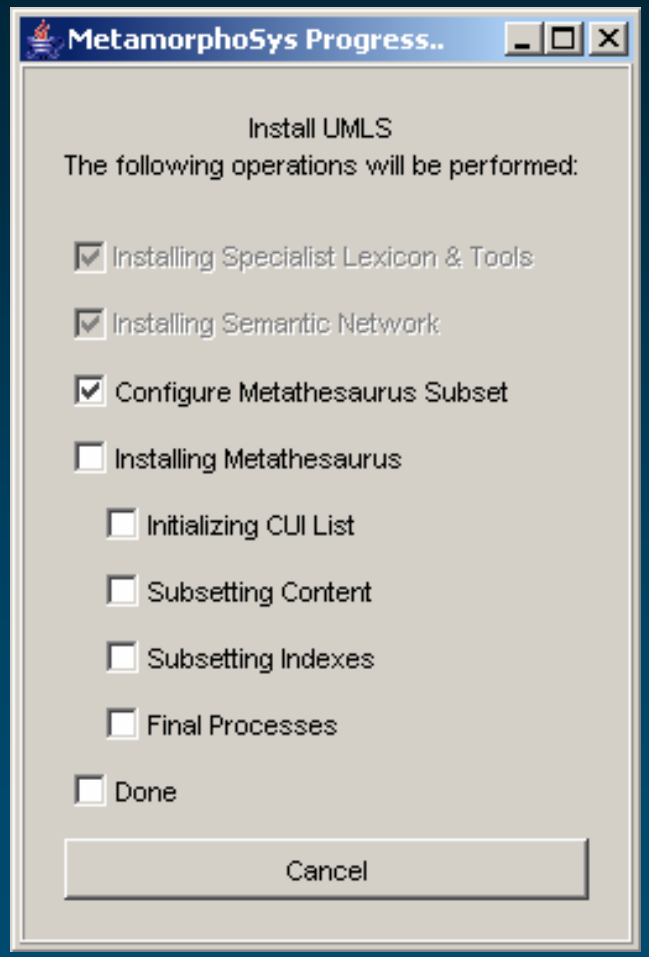
# UMLS License Notice

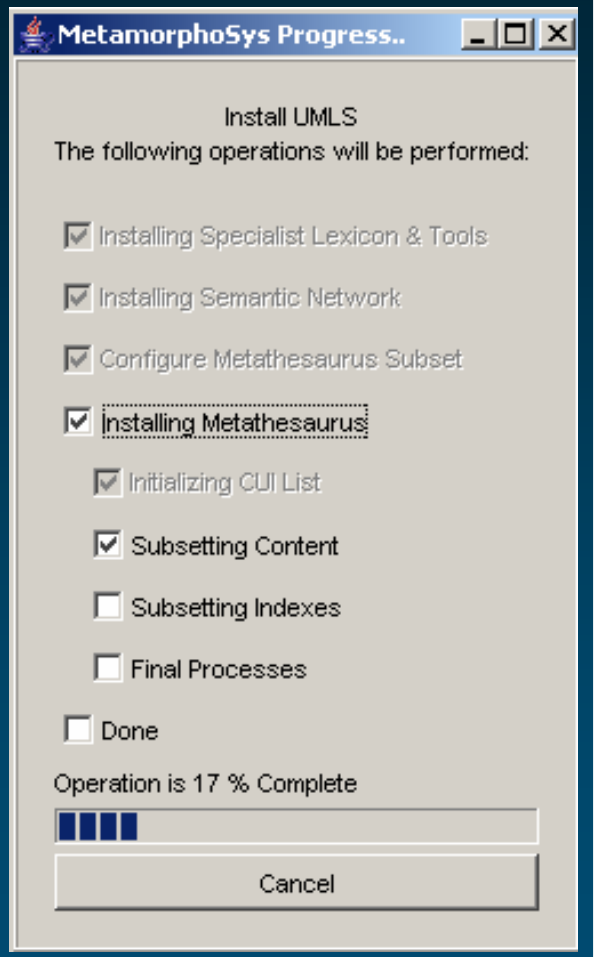


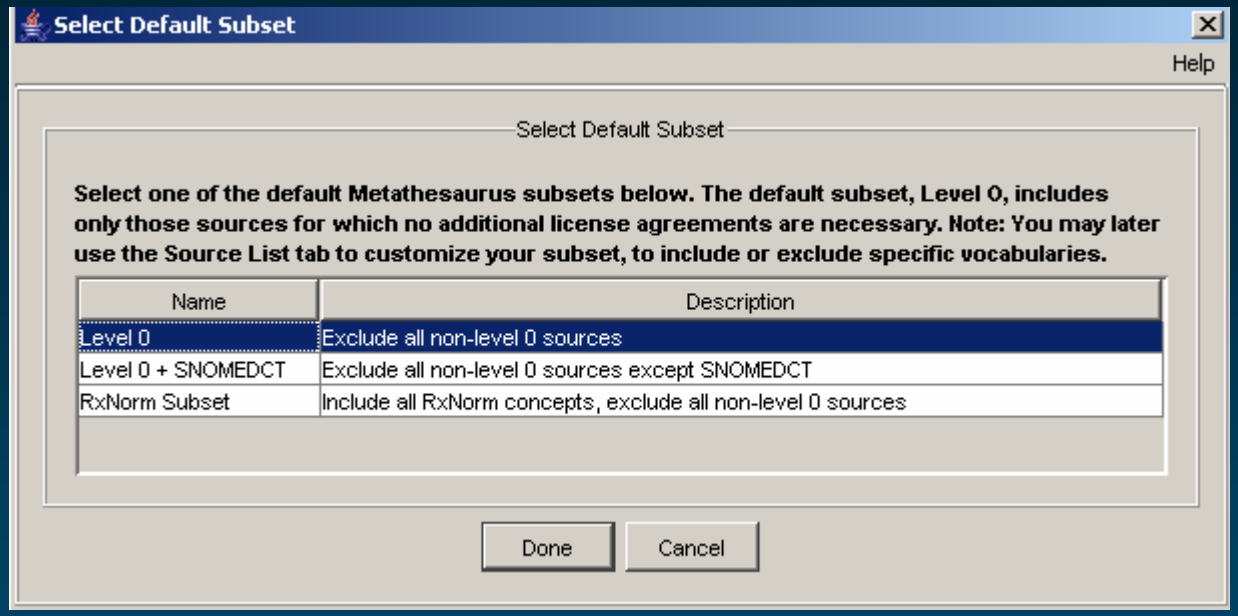
# Installation progress monitor



# Select a default subset

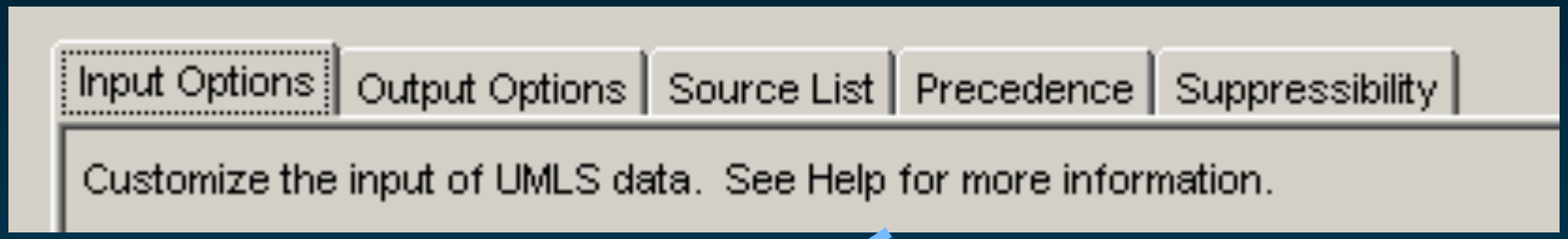


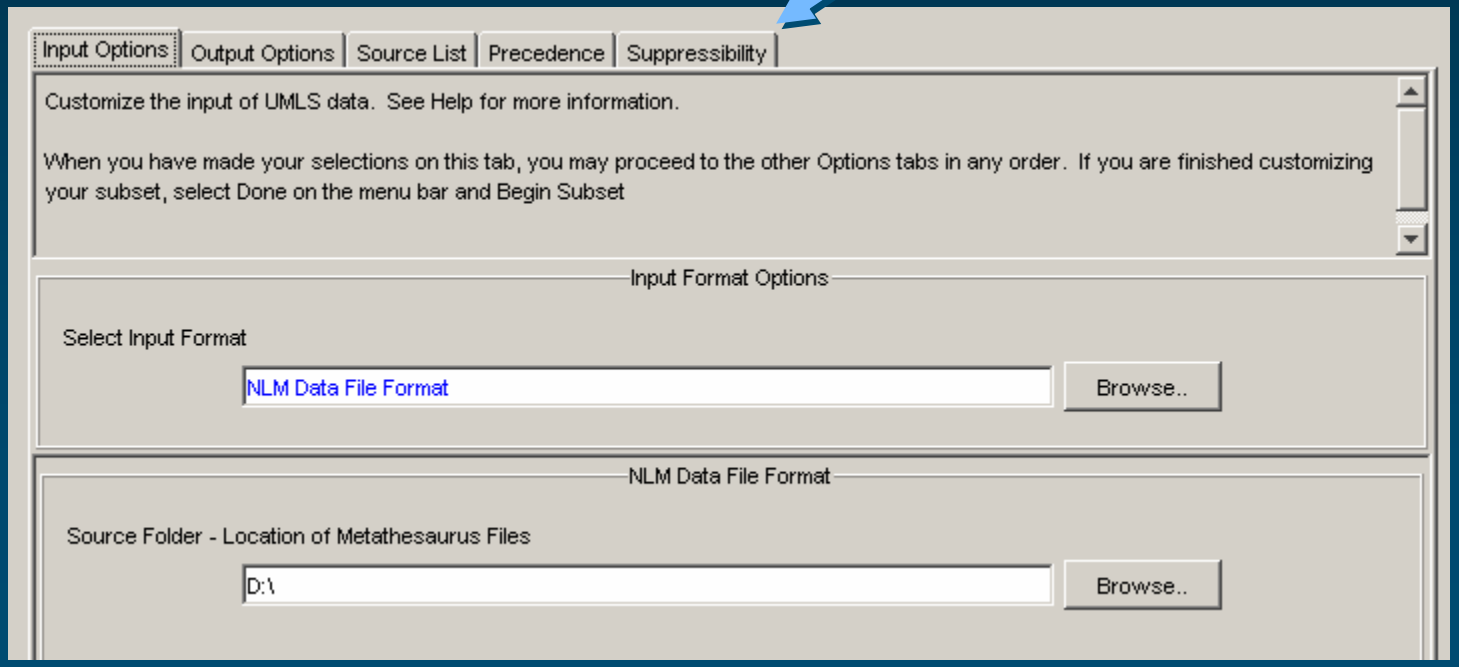

Level 0 → no separate additional license agreements

Level 0 + SNOMEDCT → Non-U.S. users must have separate license agreements

RxNorm → no separate additional license agreements

# Input Options Tab



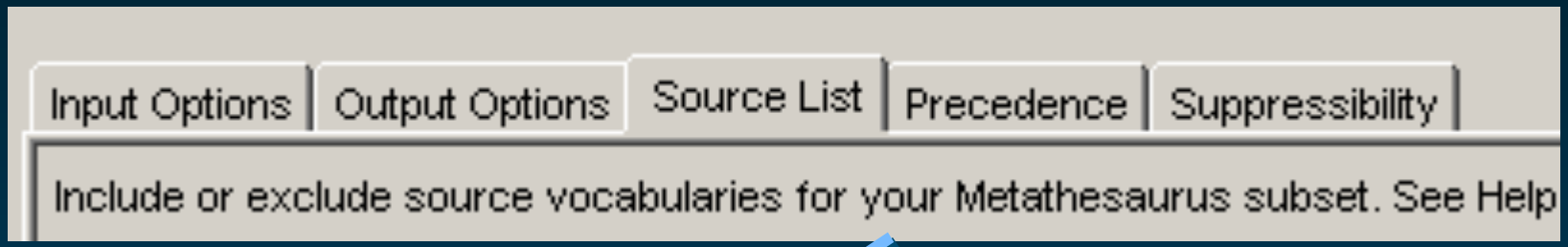
# Output Options Tab

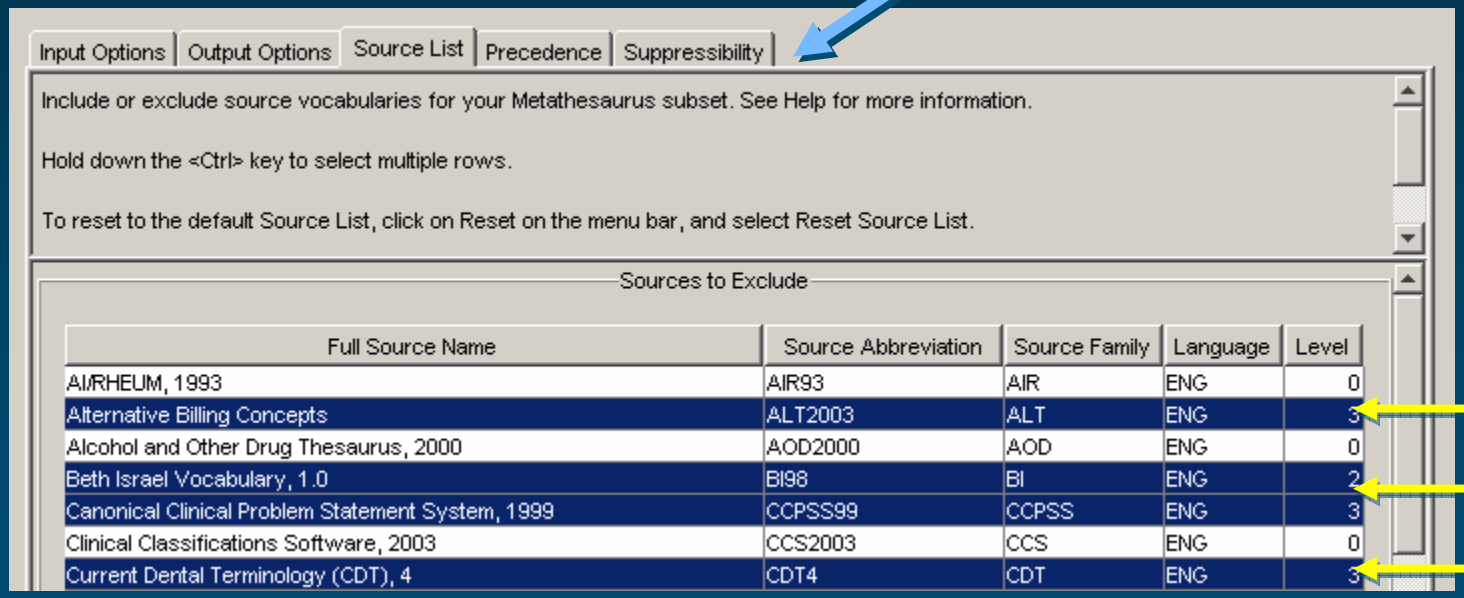
Input Options | Output Options | Source List | Precedence | Suppressibility

Select data output options for your local application. See Help for more information.

- Select Output Format
- Subset Folder - Location of Subset Files
- ☑ Remove records containing extended UTF-8 characters.
- ☐ Truncate long fields to 4000 characters.
- ☐ Output versioned source abbreviations rather than versionless source abbreviations.
- ☐ Exclude MRCXT.RRF from the subset.
- ☐ Write Oracle load script.
- ☐ Write Mysql load script.

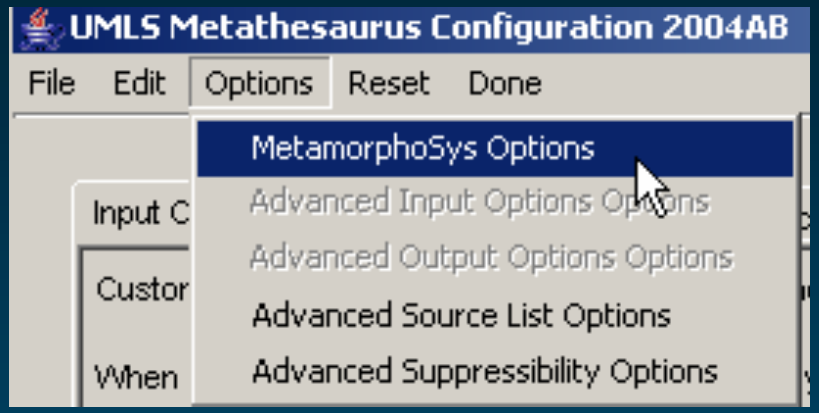
# Source List Tab



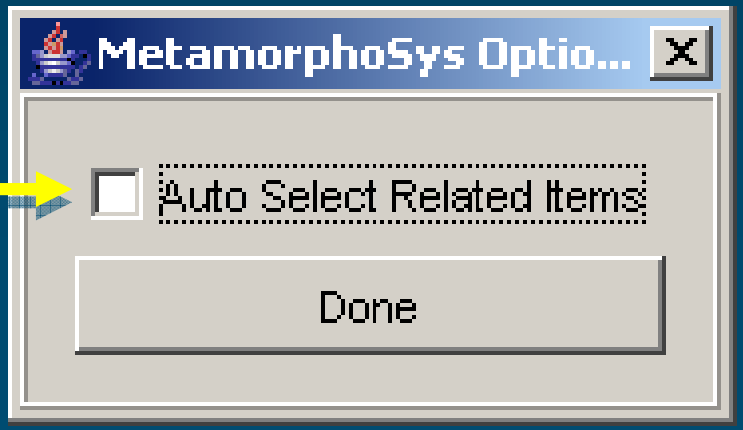

Highlighted rows are excluded from the subset.

# MetamorphoSys Option Tab

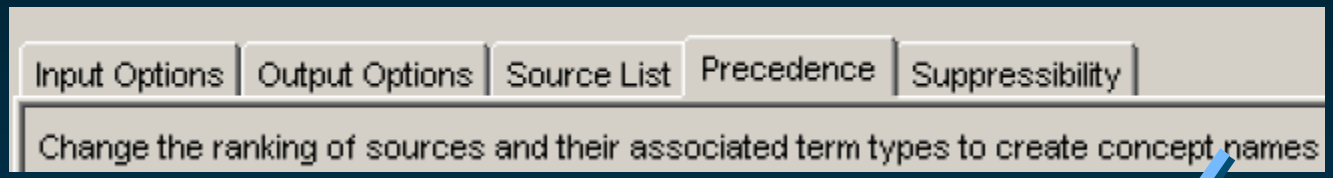



Source list behavior can be changed using the MetamorphoSys Option Tab

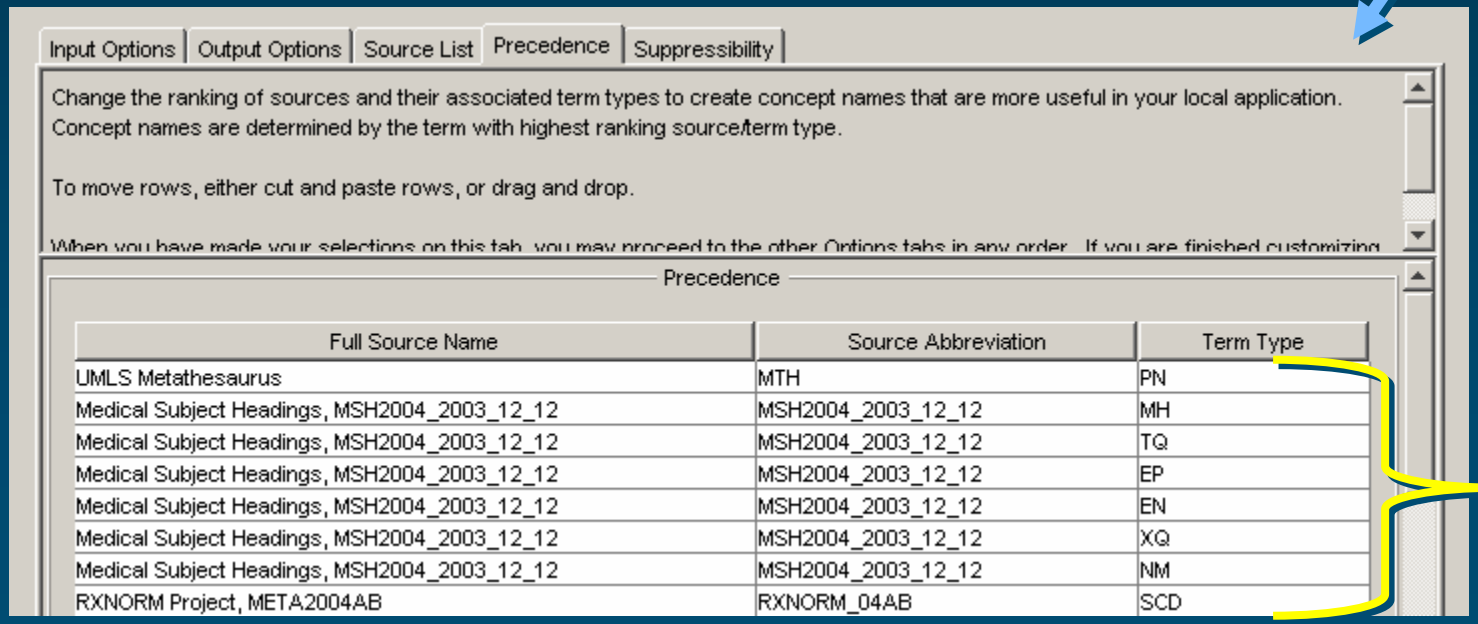
If you wish to Auto Select Related Items check this box



# Precedence Tab




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




| Full Source Name | Source Abbreviation | Term Type |
|---|---|---|
| UMLS Metathesaurus | MTH | PN |
| Medical Subject Headings, MSH2004_2003_12_12 | MSH2004_2003_12_12 | MH |
| Medical Subject Headings, MSH2004_2003_12_12 | MSH2004_2003_12_12 | TQ |
| Medical Subject Headings, MSH2004_2003_12_12 | MSH2004_2003_12_12 | EP |
| Medical Subject Headings, MSH2004_2003_12_12 | MSH2004_2003_12_12 | EN |
| Medical Subject Headings, MSH2004_2003_12_12 | MSH2004_2003_12_12 | XQ |
| Medical Subject Headings, MSH2004_2003_12_12 | MSH2004_2003_12_12 | NM |
| RXNORM Project, META2004AB | RXNORM_04AB | SCD |

Cut and paste rows to alter the preferred name

# Suppressibility Tab

Input Options | Output Options | Source List | Precedence | Suppressibility

See Help for more information.

Input Options | Output Options | Source List | Precedence | Suppressibility

See Help for more information.

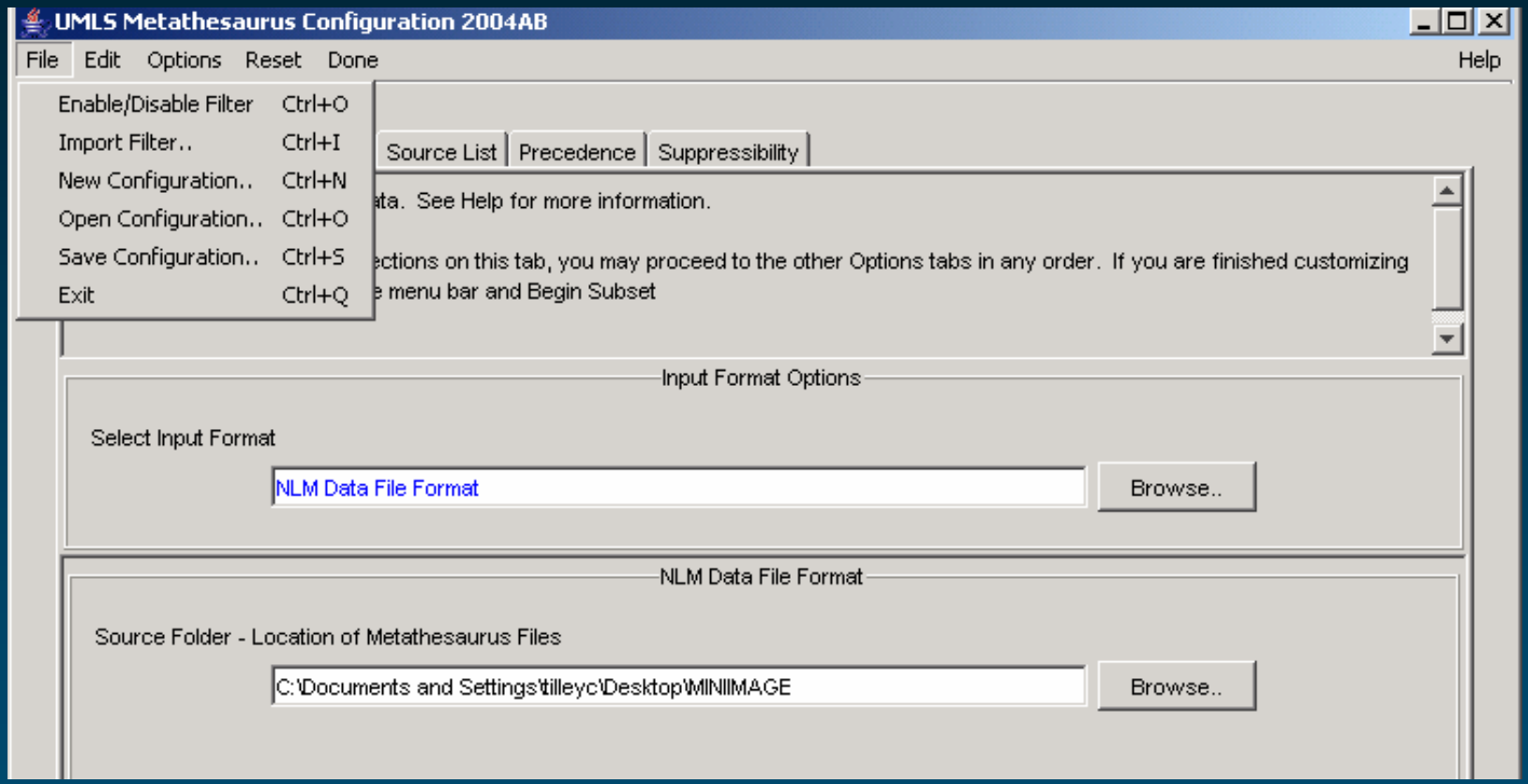
When you have made your selections on this tab, you may proceed to the other Options tabs in any order. If you are finished customizing your subset, select Done on the menu bar and Begin Subset.

| | | |
|---|---|---|
| International Classification of Primary Care, 1993 | ICPC93 | CP |
| International Classification of Primary Care, 1993 | ICPC93 | CS |
| International Classification of Primary Care, 1993 | ICPC93 | CX |
| International Classification of Primary Care, 1993 | ICPC93 | HT |
| International Classification of Primary Care, 1993 | ICPC93 | PC |
| International Classification of Primary Care, 1993 | ICPC93 | PS |
| International Classification of Primary Care, 1993 | ICPC93 | PT |
| International Classification of Primary Care, 1993 | ICPC93 | PX |
| ICPC, Basque Translation, 1993 | ICPCBAQ_1993 | CP |

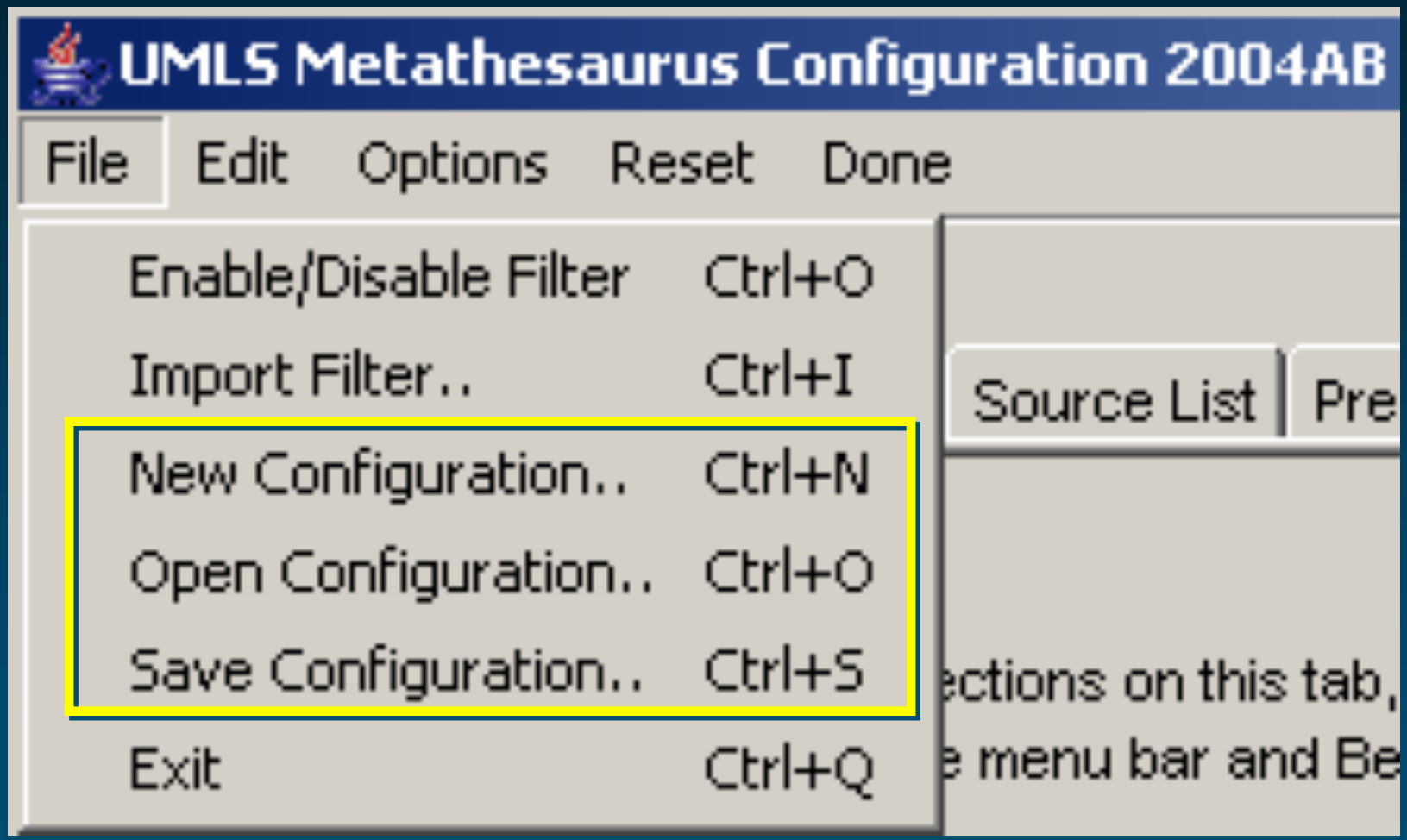
Highlighted source term types will be marked as suppressible

NLM

# File menu


UMLS Metathesaurus Configuration 2004AB
File
Edit
Options
Reset
Done
Help
Enable/Disable Filter Ctrl+O
Import Filter.. Ctrl+I
New Configuration.. Ctrl+N
Open Configuration.. Ctrl+O
Save Configuration.. Ctrl+S
Exit Ctrl+Q
Source List
Precedence
Suppressibility
ata. See Help for more information.
ections on this tab, you may proceed to the other Options tabs in any order. If you are finished customizing
e menu bar and Begin Subset
Input Format Options
Select Input Format
NLM Data File Format
Browse..
NLM Data File Format
Source Folder - Location of Metathesaurus Files
C:\Documents and Settings\tilleyc\Desktop\MINIIMAGE
Browse..

# File menu


UMLS Metathesaurus Configuration 2004AB
File
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Options
Reset
Done
Enable/Disable Filter Ctrl+O
Import Filter.. Ctrl+I
New Configuration.. Ctrl+N
Open Configuration.. Ctrl+O
Save Configuration.. Ctrl+S
Exit Ctrl+Q
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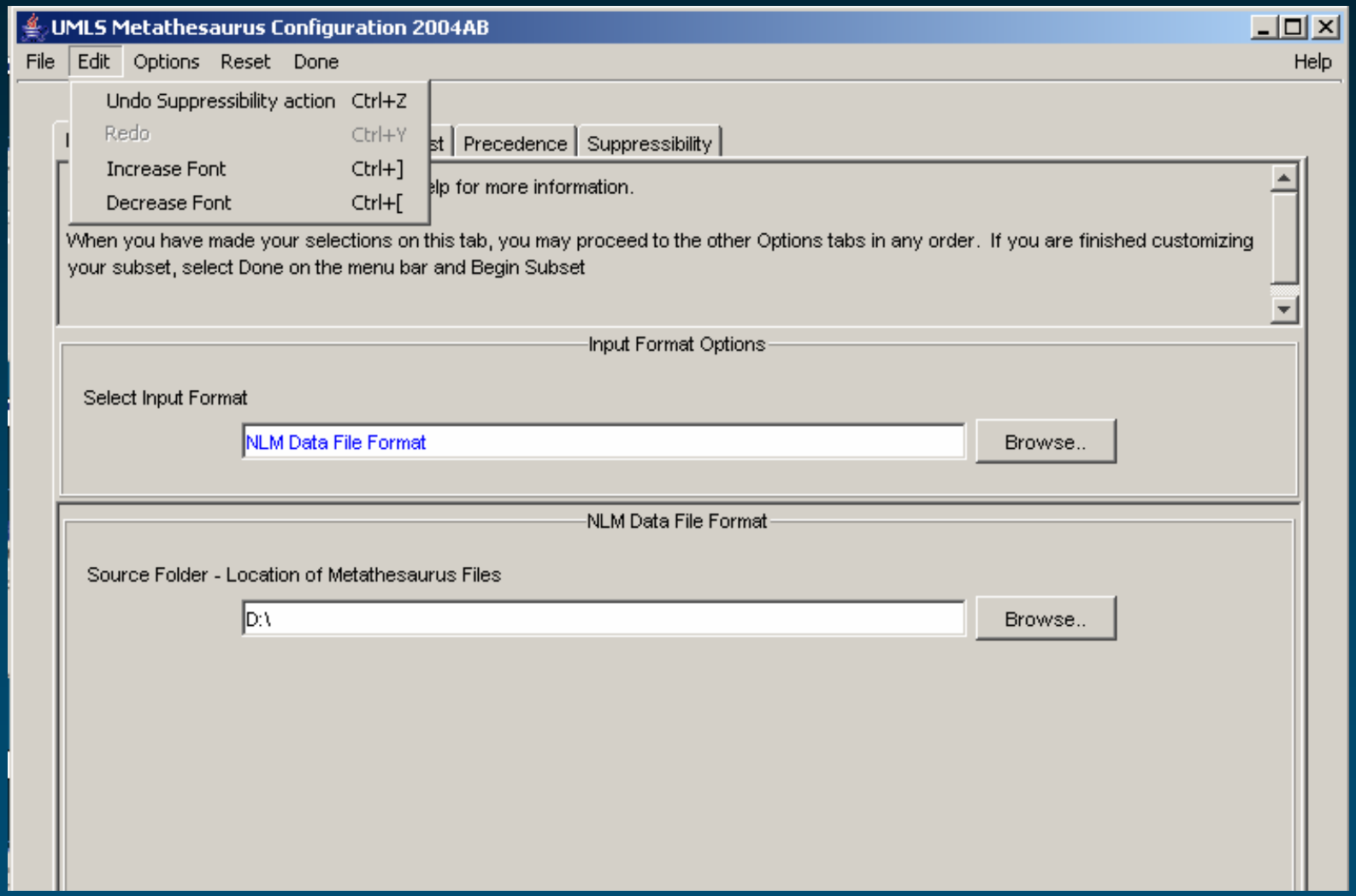
# Edit menu


UMLS Metathesaurus Configuration 2004AB
File
Edit
Options
Reset
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Help
Undo Suppressibility action Ctrl+Z
Redo Ctrl+Y
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st
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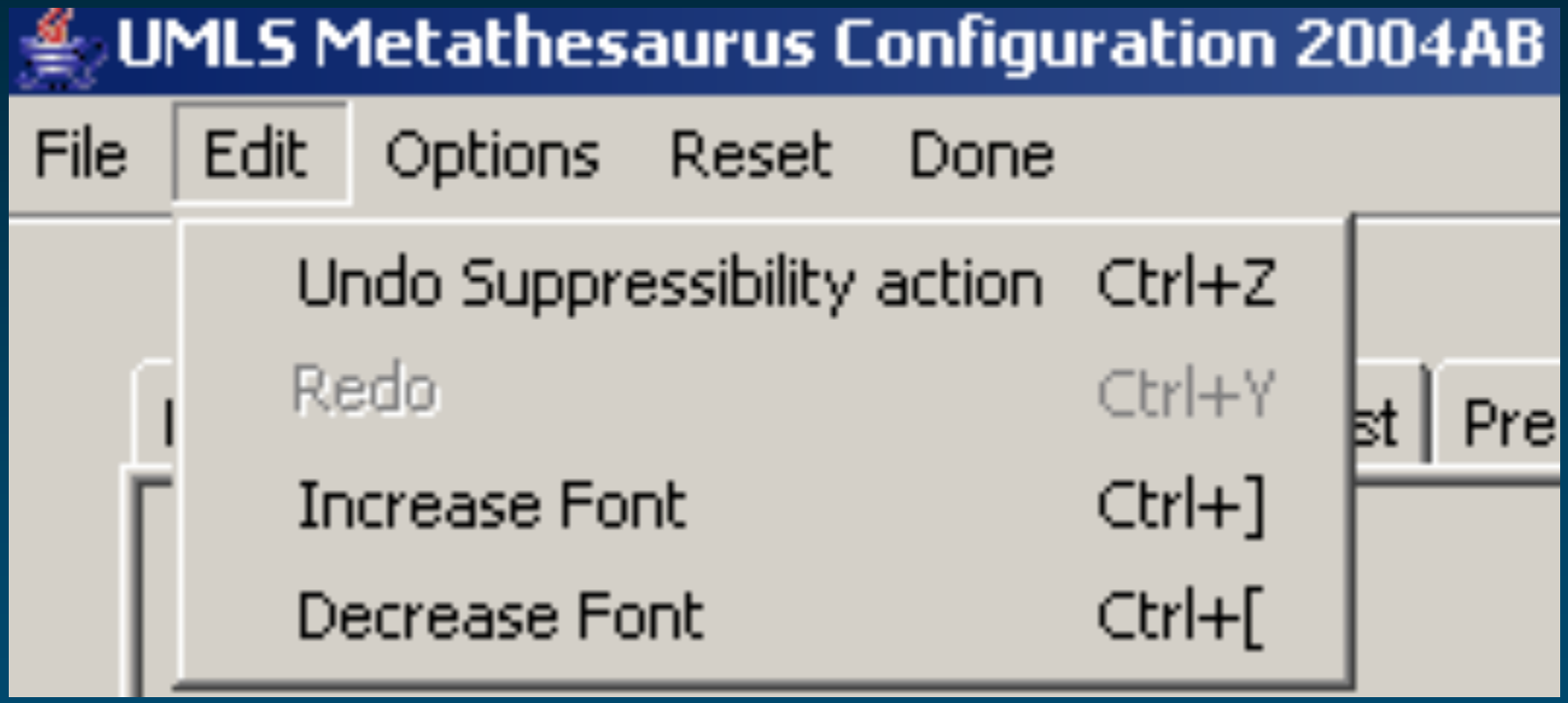
lp for more information.
When you have made your selections on this tab, you may proceed to the other Options tabs in any order. If you are finished customizing your subset, select Done on the menu bar and Begin Subset
Input Format Options
Select Input Format
NLM Data File Format
Browse..
NLM Data File Format
Source Folder - Location of Metathesaurus Files
D:\
Browse..

# Edit menu


UMLS Metathesaurus Configuration 2004AB
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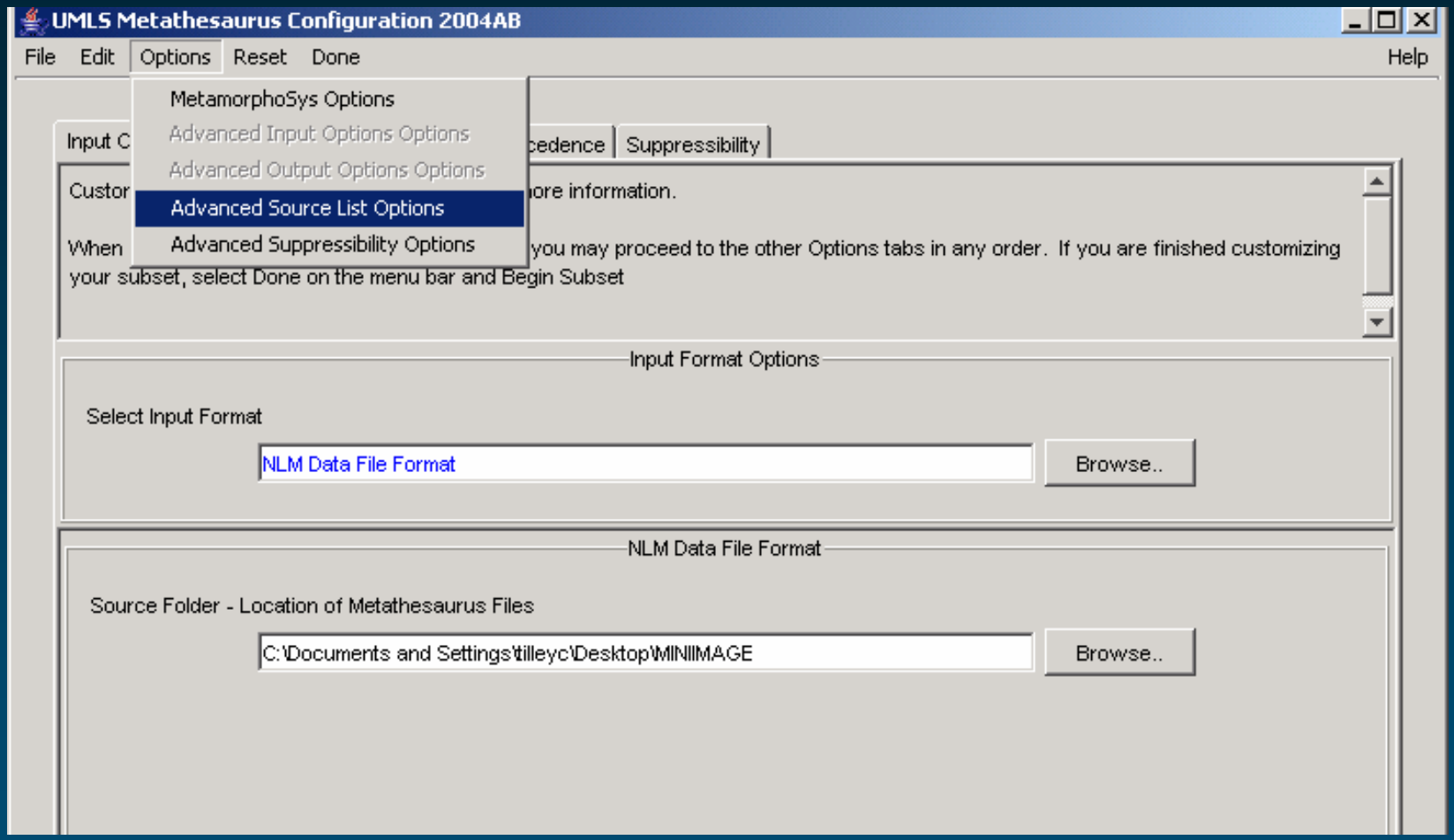
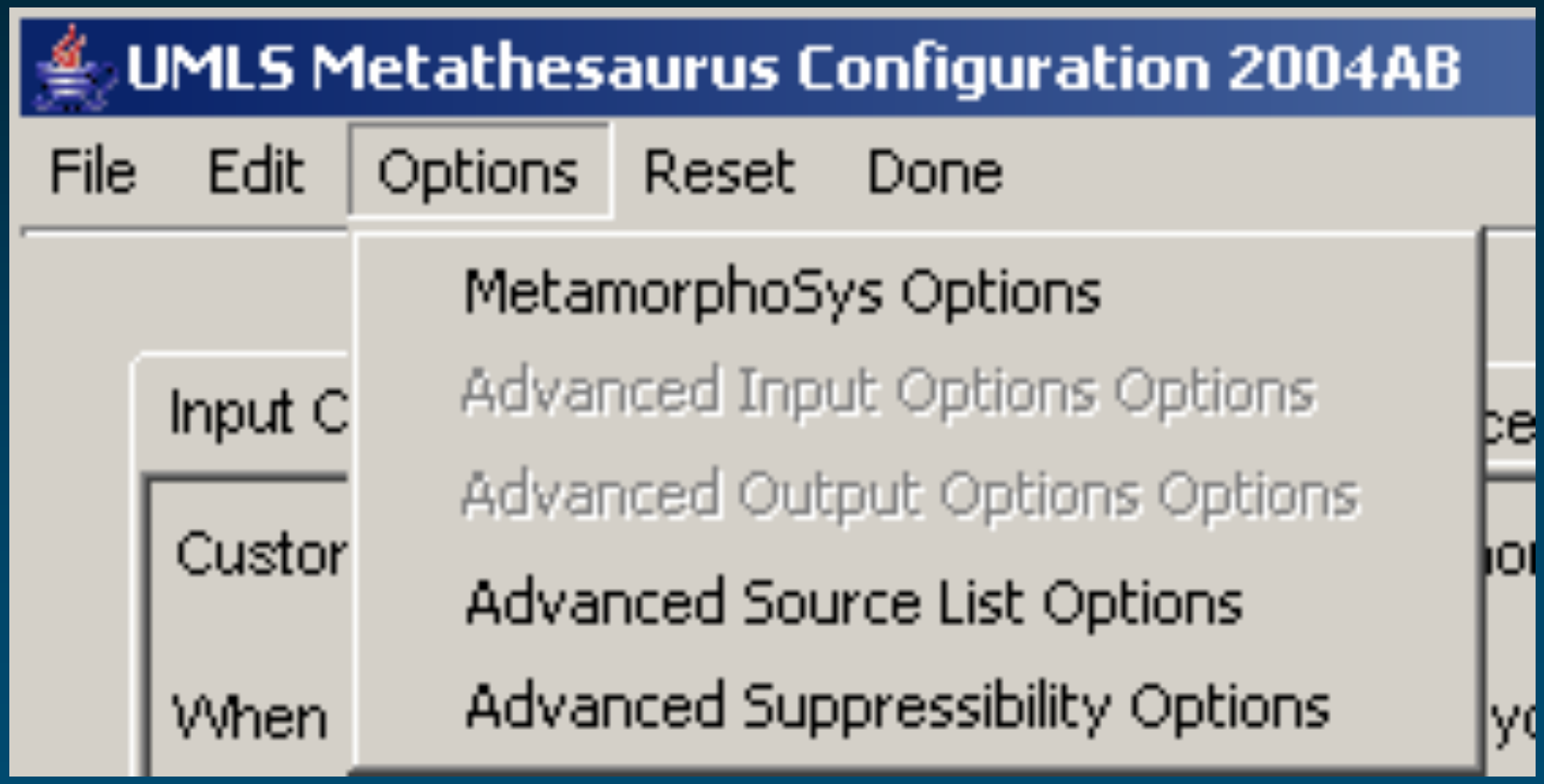
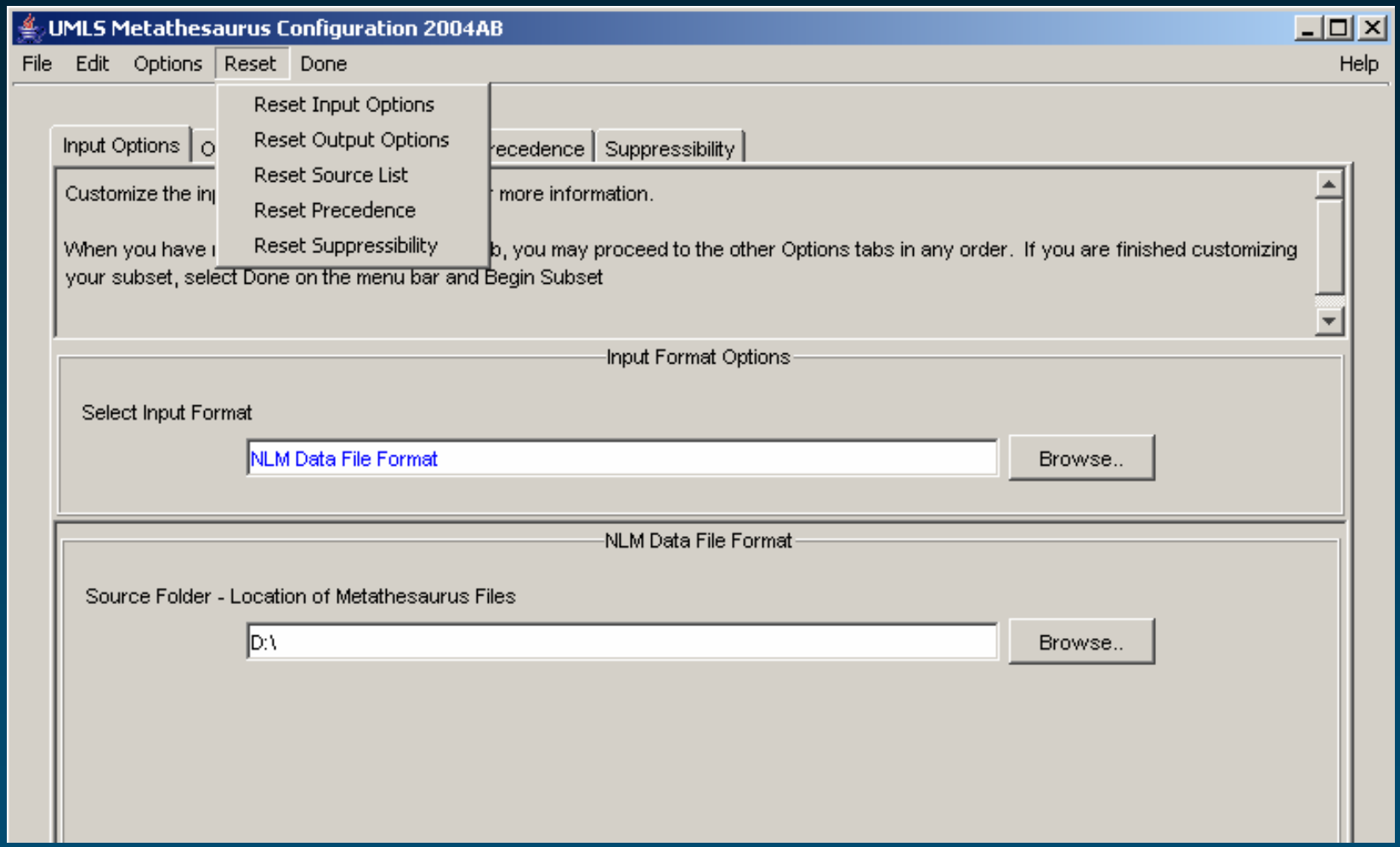
# Options menu



# Options menu



# Reset menu


UMLS Metathesaurus Configuration 2004AB
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Help
Reset Input Options
Reset Output Options
Reset Source List
Reset Precedence
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more information.
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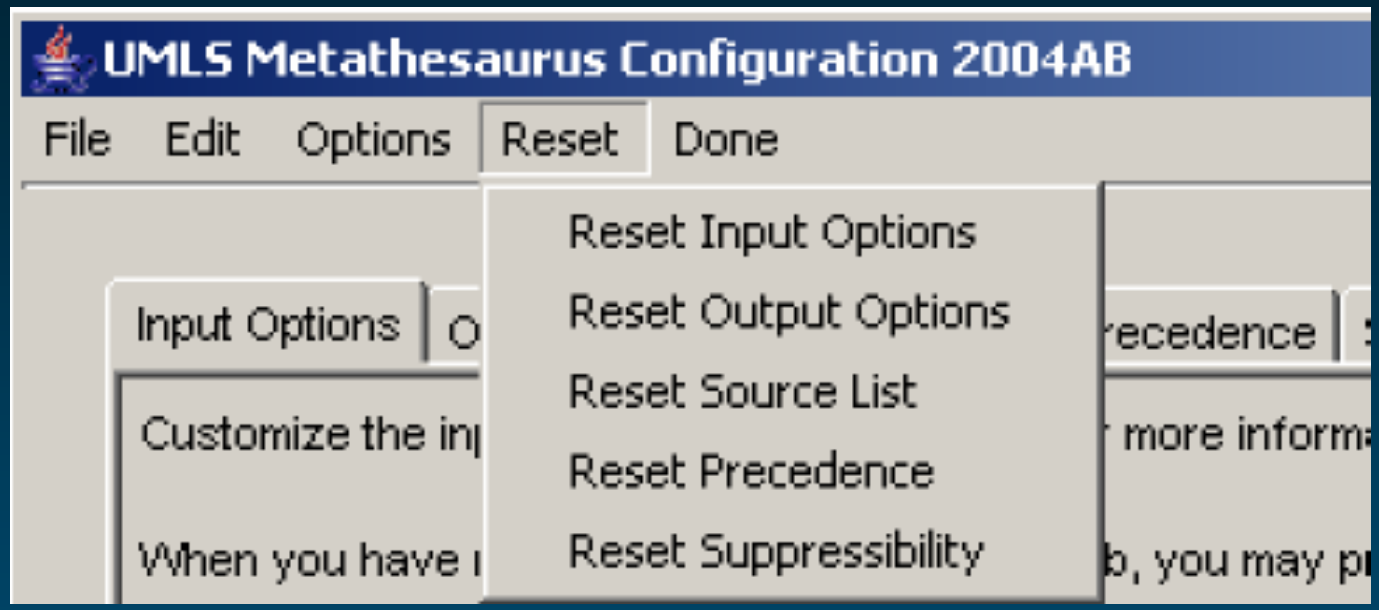
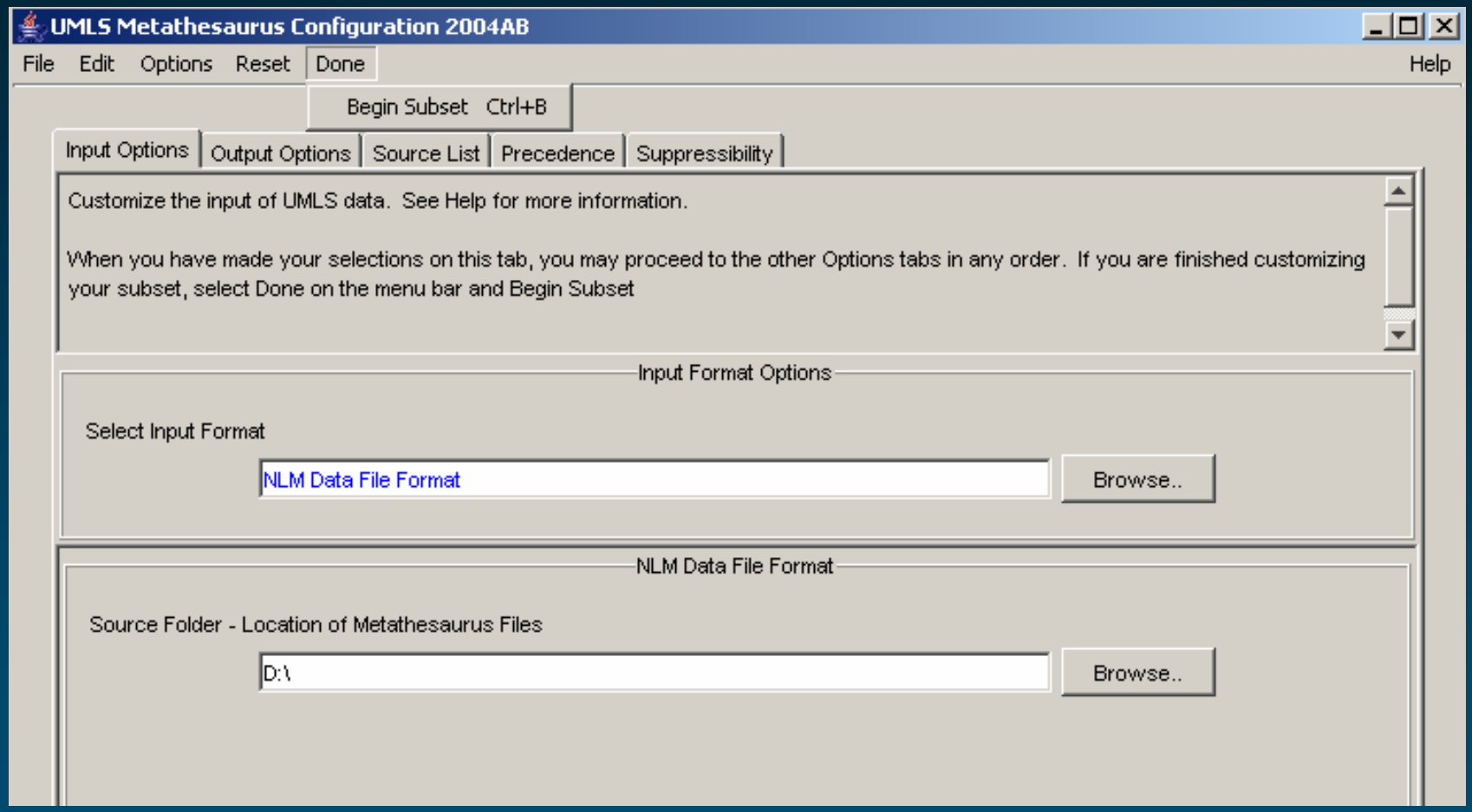
b, you may proceed to the other Options tabs in any order. If you are finished customizing your subset, select Done on the menu bar and Begin Subset
Input Format Options
Select Input Format
NLM Data File Format
Browse..
NLM Data File Format
Source Folder - Location of Metathesaurus Files
D:\
Browse..

# Reset menu

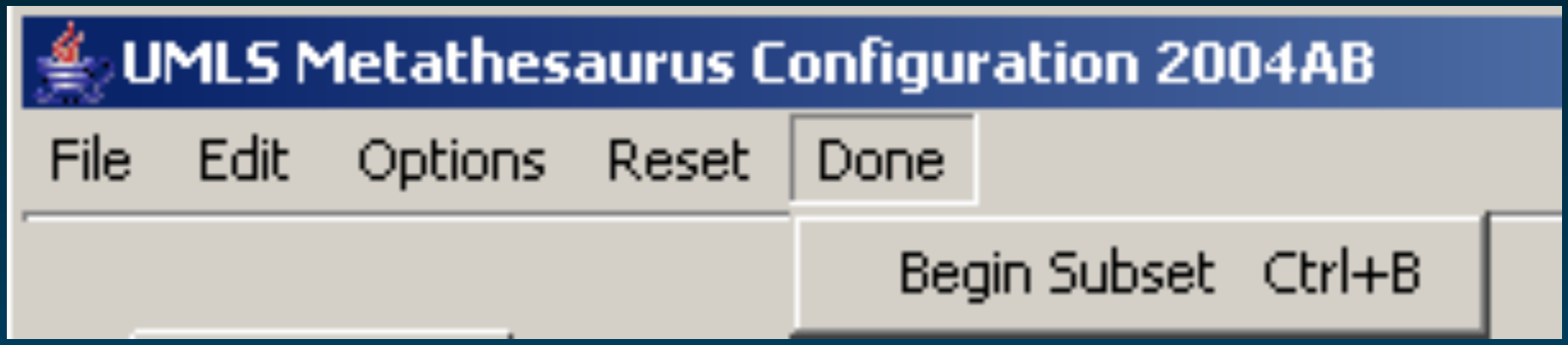



- Returns all filters to default selections
- Default selections in “mmsys.prop.default file” in config folder
- mmsys.prop.default contains properties in last run

# Done – Begin Subset



# Done – Begin Subset




- Compete configuration options
- Done menu
- Begin Subset

# 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

mmsys.log - Notepad

File Edit Format View Help

```
MetamorphoSys Version:................5.21
MetamorphoSys Build Date:.............2004_08_30_14_47_11
UMLS Build Date:......................2004_07_12_09_57_26
Release Version:......................2004AB
Release Date:........................20040720
Release Description:..................July 2004 Release
Metathesaurus Source paths:...........C:\UMLS\DVDIMAGE
Subsetted Metathesaurus folder:.......C:\UMLS\DVDIMAGE\2004AB\META
Start at:............................Wed Sep 01 13:04:18 EDT 2004
Initialize CUI List completed:........Wed Sep 01 13:06:20 EDT 2004
Subset Metathesaurus completed:.......Wed Sep 01 14:13:02 EDT 2004
Subset Index Files completed:.........Wed Sep 01 14:21:11 EDT 2004
Subset Release Metadata completed:....Wed Sep 01 14:21:17 EDT 2004
Finished at:.........................Wed Sep 01 14:22:48 EDT 2004
Concepts in source:...................1078246
Concepts in subset:...................1078246
Time elapsed:........................01:18:29
```

# MetamorphoSys log

mmsys.log - Notepad

File Edit Format View Help

```
--------------------------------
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.


Excluded Sources
  <none>

Kept Sources
  AI/RHEUM, 1993                                                    AIR93
  Alternative Billing Concepts                                      ALT2003
  Alcohol and Other Drug Thesaurus, 2000                            AOD2000
  Beth Israel Vocabulary, 1.0                                       BI98
  Canonical Clinical Problem Statement System, 1999                 CCPSS99
  Clinical Classifications Software, 2003                           CCS2003
  Current Dental Terminology (CDT), 4                               CDT4
  COSTAR, 1989-1995                                                 COSTAR_89-95
  Medical Entities Dictionary, 2003                                 CPM2003
  Physicians' Current Procedural Terminology, Spanish Translation,... CPT01SP
  Physicians' Current Procedural Terminology, 2004                  CPT2004
  CRISP Thesaurus, 2004                                             CSP2004
  COSTART, 1995                                                     CST95
  Diseases Database, 2000                                           DDB00
  German translation of ICD10, 1995                                 DMDICD10_1995
  German translation of UMDNS, 1996                                 DMDUMD_1996
```

# Output directory contents

| Name | Size | Type |
|---|---|---|
| CHANGE | | File Folder |
| indexes | | File Folder |
| release.dat | 1 KB | DAT File |
| config.prop | 8 KB | PROP File |
| AMBIGLUI.RRF | 1,225 KB | RRF File |
| AMBIGSUI.RRF | 955 KB | RRF File |
| MRCOC.RRF | 809,207 KB | RRF File |
| MRCOLS.RRF | 21 KB | RRF File |
| MRCONSO.RRF | 596,528 KB | RRF File |
| MRCUI.RRF | 9,221 KB | RRF File |
| MRCXT.RRF | 9,391,778 KB | RRF File |
| MRDEF.RRF | 17,172 KB | RRF File |
| MRDOC.RRF | 88 KB | RRF File |
| MRFILES.RRF | 4 KB | RRF File |
| MRHIER.RRF | 899,786 KB | RRF File |
| MRHIST.RRF | 70,843 KB | RRF File |
| MRMAP.RRF | 9,362 KB | RRF File |

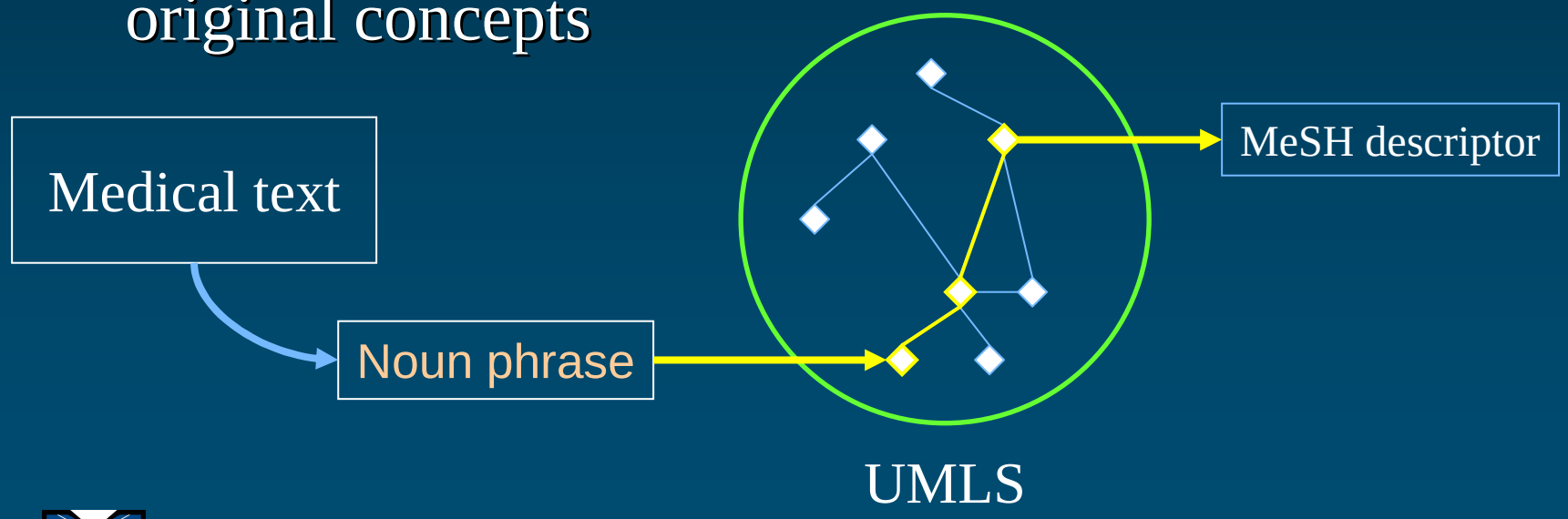
# Part II
# How to use the UMLS?

## *(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 — MRCON + MRSO
  - Use Associated expressions (ATXs) — MRATX
  - Explore the Ancestors — MRREL + SN
  - Explore the Other related concepts — MRREL + SN

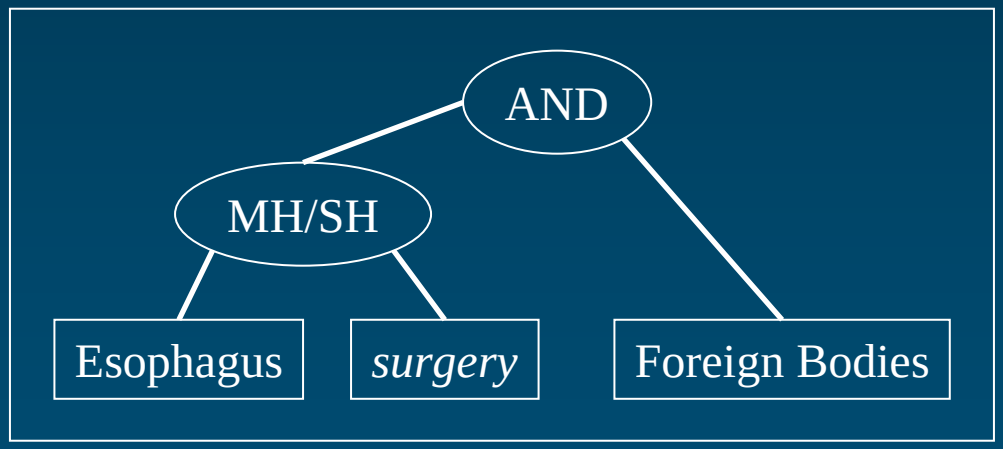
# Restrict to MeSH Synonymy

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

# Restrict to MeSH Assoc. expressions

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

Endoscopic removal of intraluminal foreign body from oesophagus without incision



# Restrict to MeSH Ancestors

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

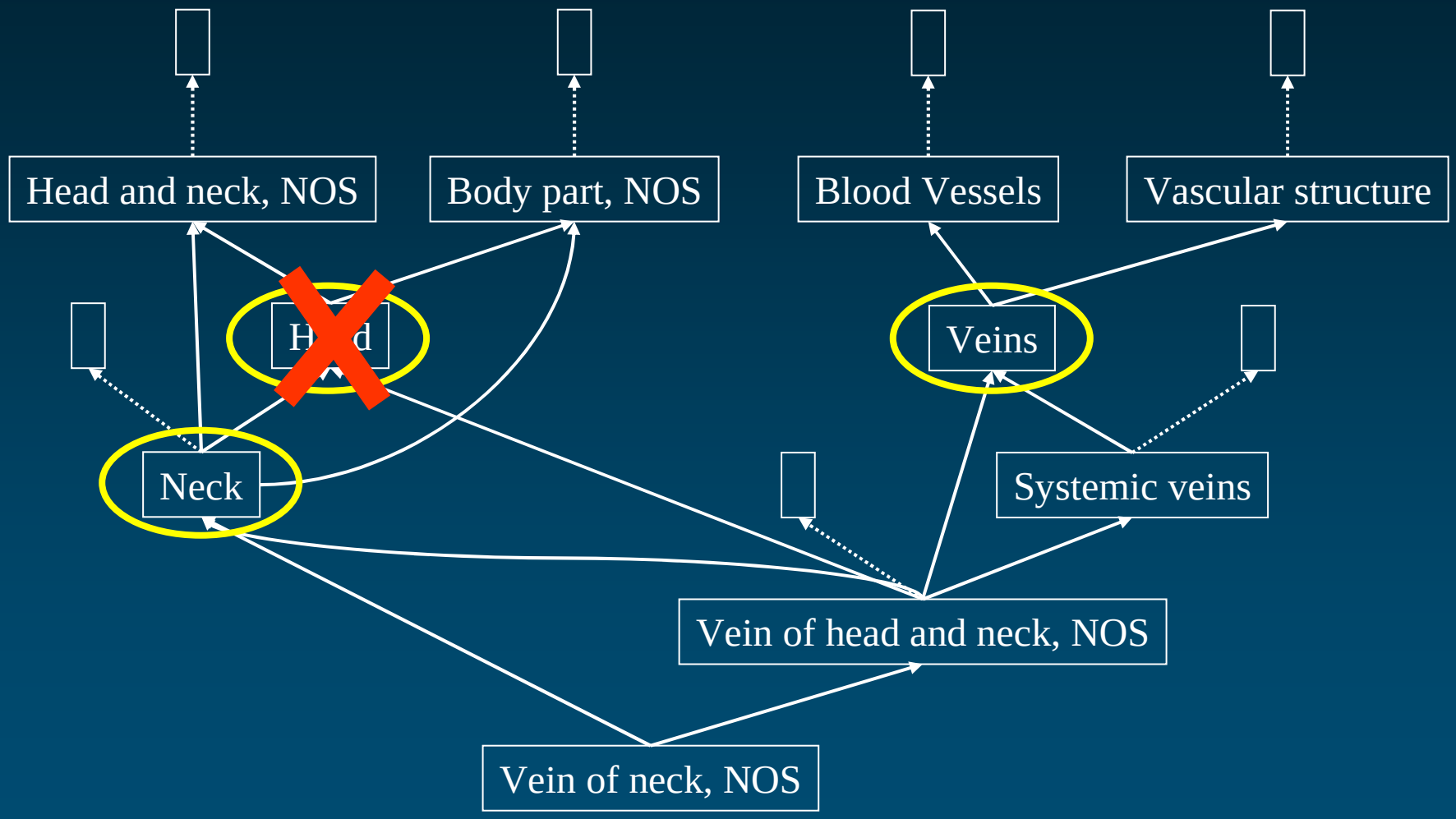
# Restrict to MeSH Example

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

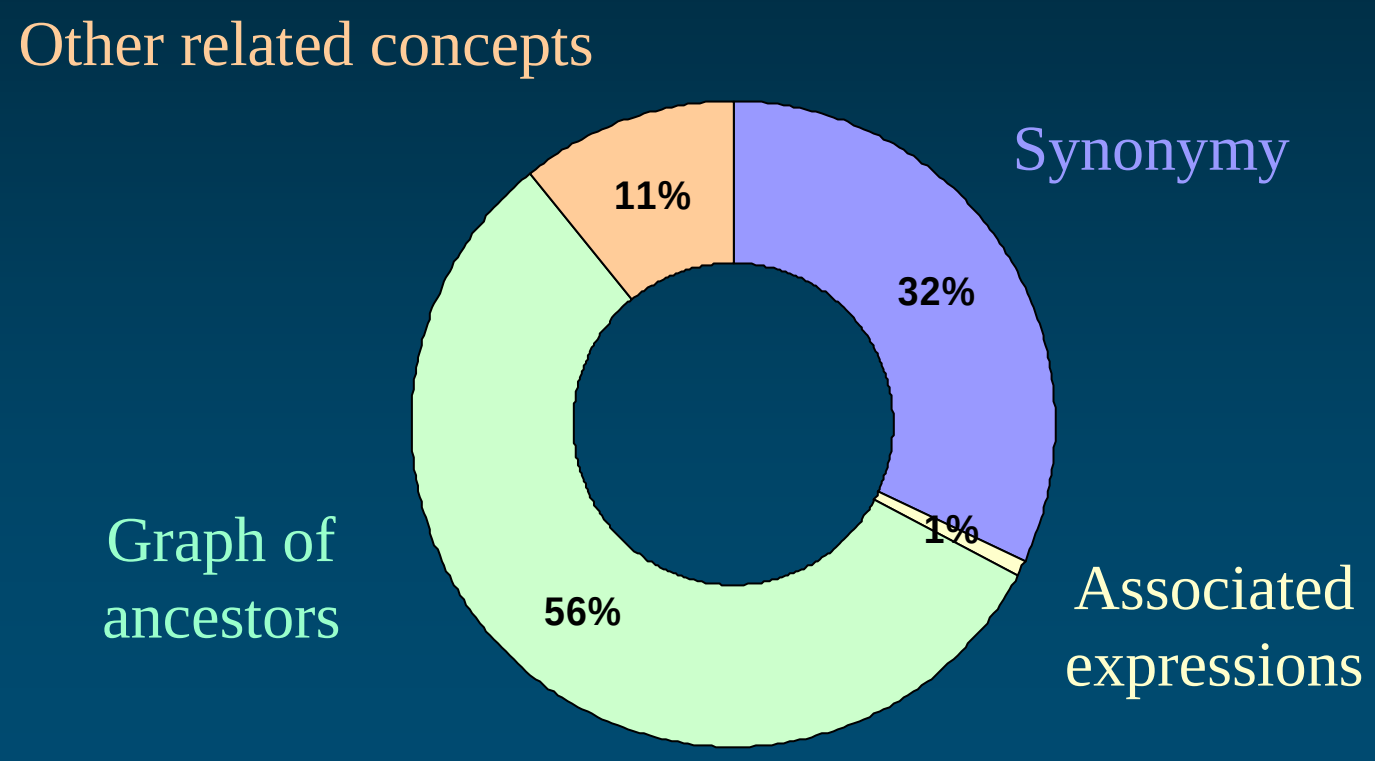
Vein + Neck

# Restrict to MeSH Example



# Restrict to MeSH Quantitative results

- 82.5% of UMLS concepts mapped to MeSH



# 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

NLM

# 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

# UMLA as an enabling resource

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

# Limitations

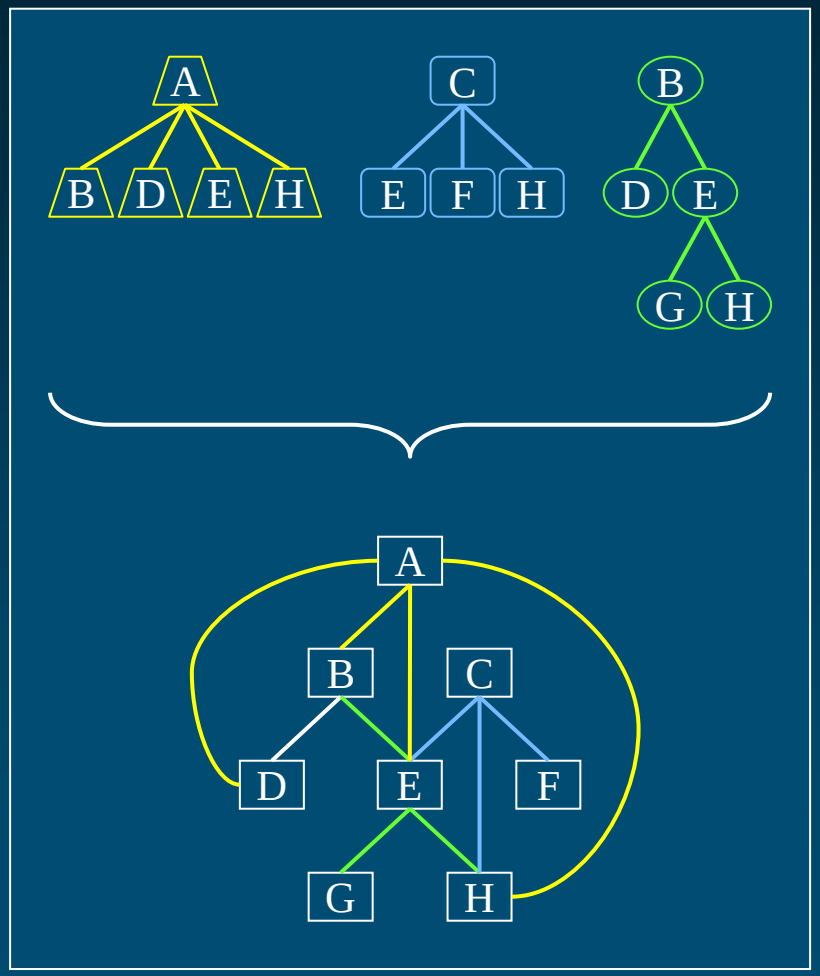
# Limitations

[Cimino, *JAMIA*, 1998]

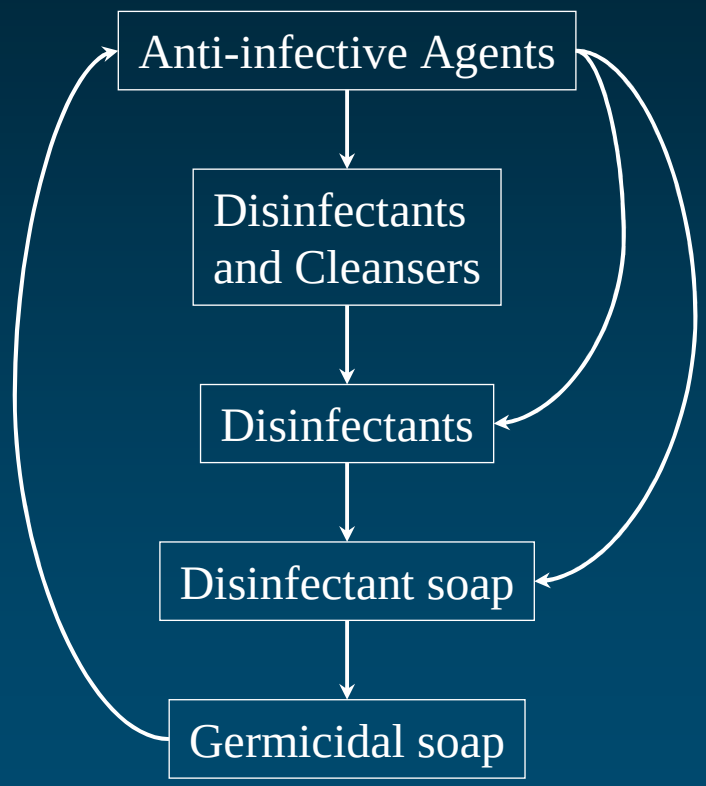
- Structural inconsistency
  - Cycles in the graph of hierarchical relations
- Semantic inconsistency
  - Between Metathesaurus and Semantic Network
- Missing relations
  - Synonymy
  - Hierarchical relations (missing or underspecified)

# Structural inconsistency From trees to graph

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

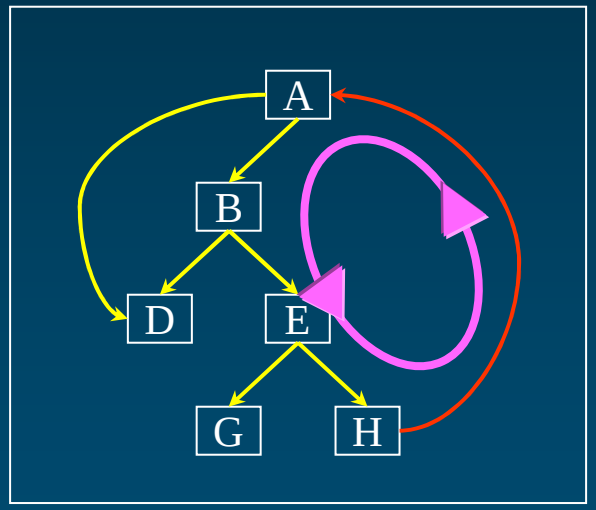


# Structural inconsistency There are some cycles



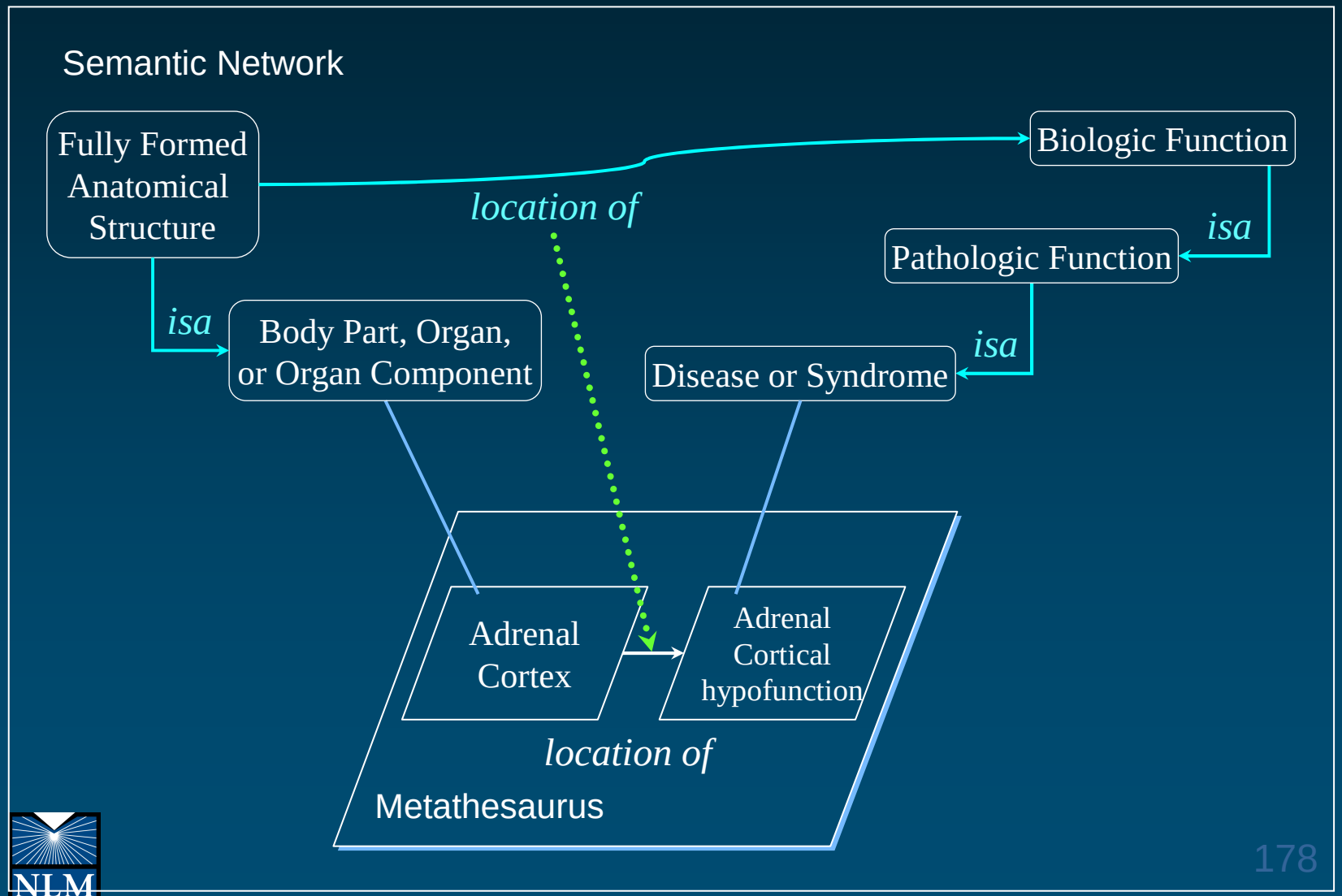
# Structural inconsistency Issues

- Theoretical
  - Violate the antisymmetry property of partial ordering relations
- Practical
  - Loops in graph traversal
  - Impossible to perform transitive reduction




[Bodenreider, AMIA 2001]

# Semantic inconsistency A two-level structure

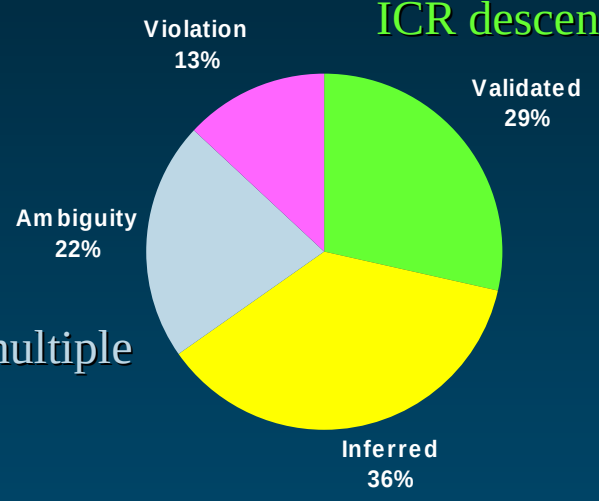


# Semantic inconsistency A limited study

- 6894 interconcept relationships
  - among the 3764 concepts in the semantic neighborhood of "Heart"

ICR and SNR not compatible

ICR = SNR or ICR descendant of SNR




ICR not specified and SNR compatible and multiple

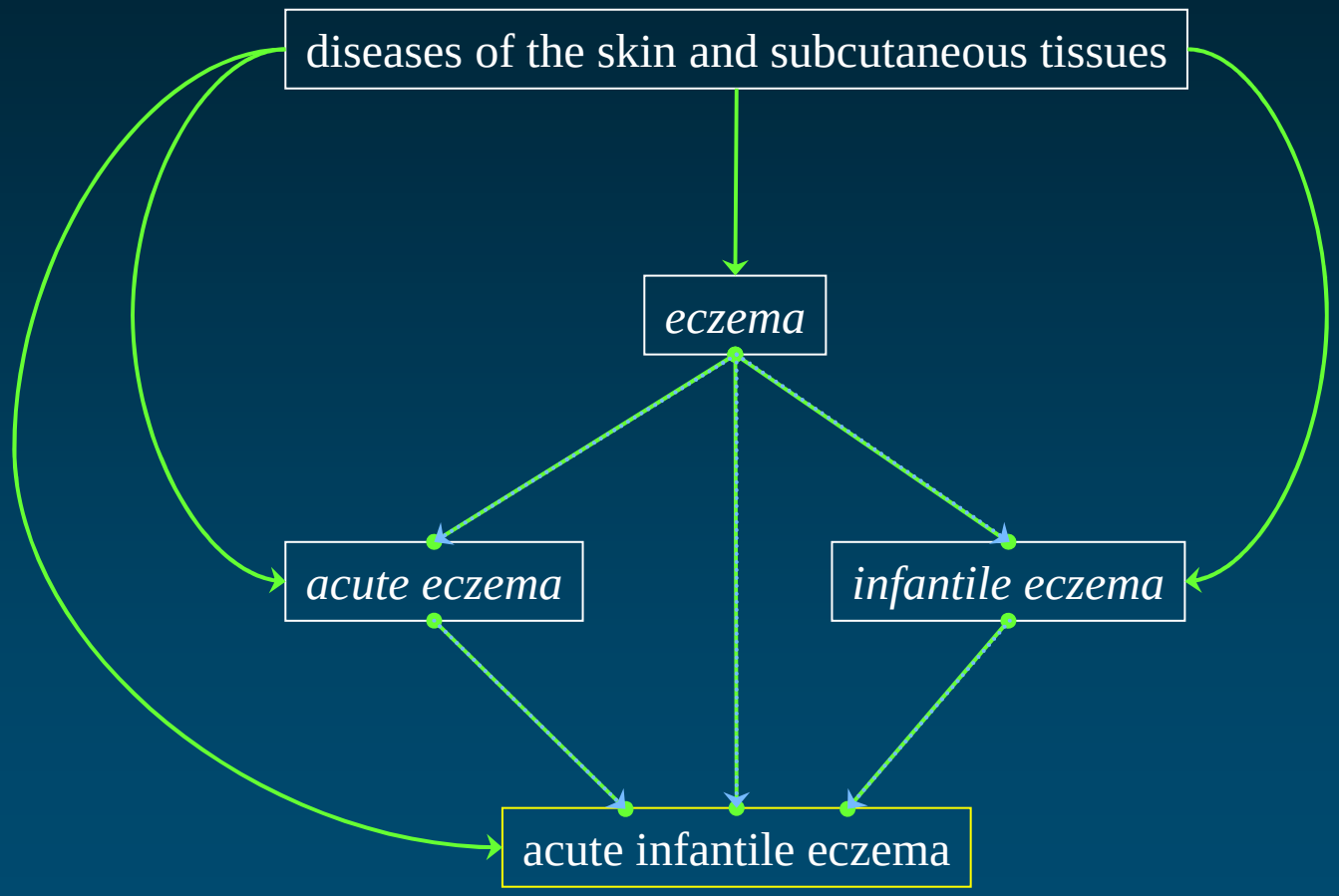
ICR not specified and SNR compatible and unique

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.

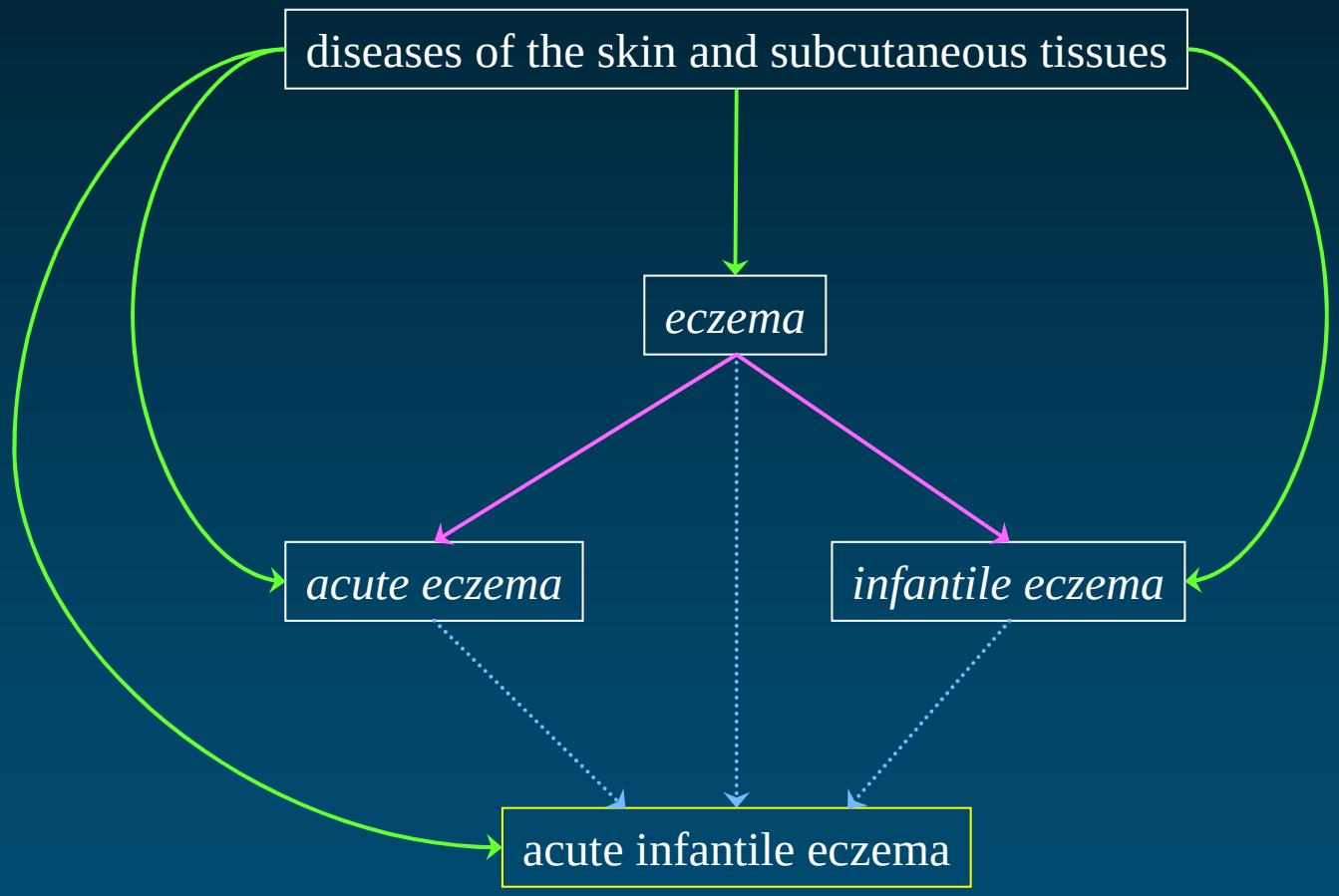
# Semantic inconsistency Issues

- 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]

# Missing relations Example



# Missing relations Example



# Missing relations A limited study

- 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]

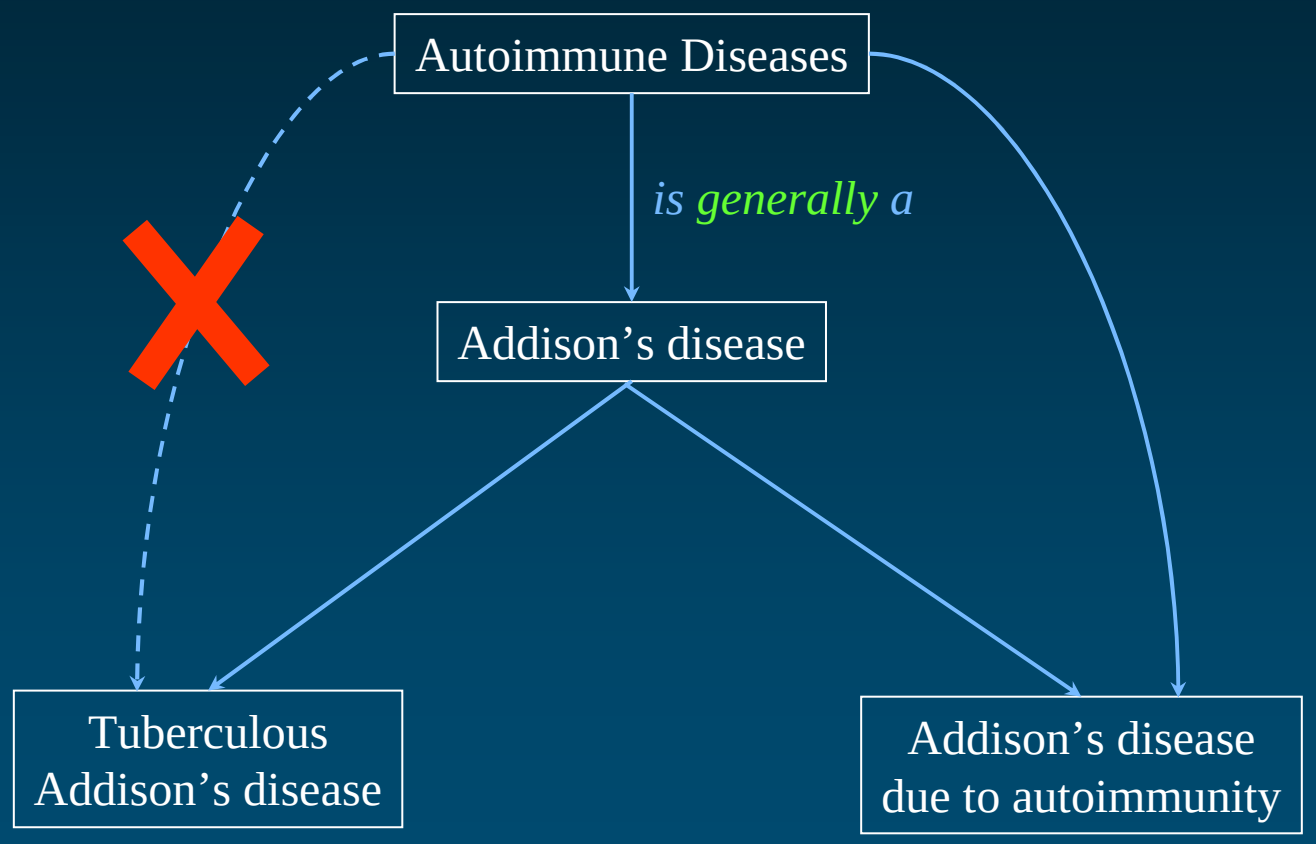
# Compensation mechanisms

- 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

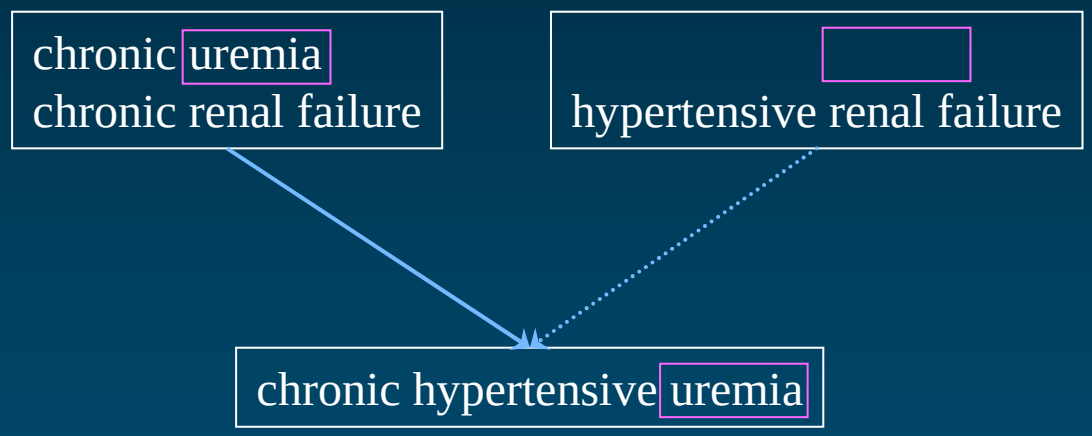
# More limitations

- Meaning of *isa*
- Some missing / wrong relations are hard to detect
- Some relations are present but hard to find

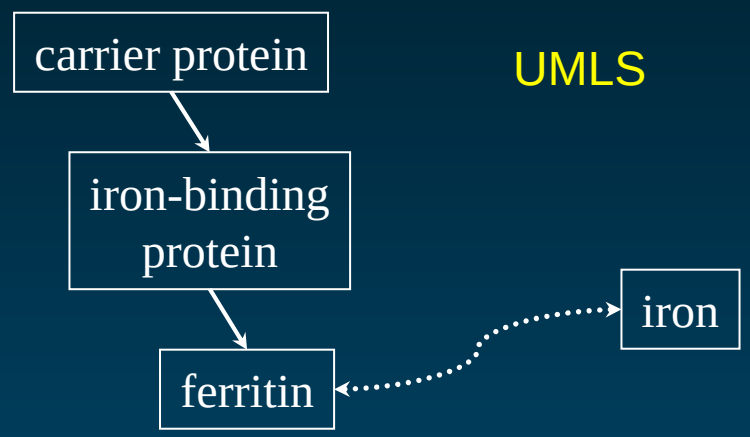
# Meaning of *isa*



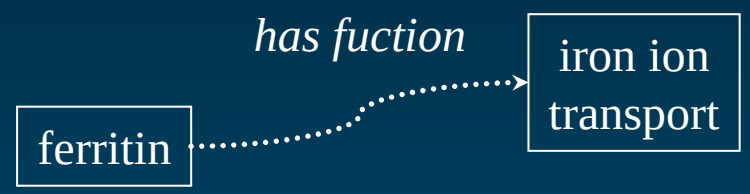
# Relations Missing and difficult to detect



# Relations Existing but difficult to find




UMLS




Gene Ontology

ferritin *isa* iron transporter

reified “transport” relationship

ferritin *transports* iron

“transport” relationship

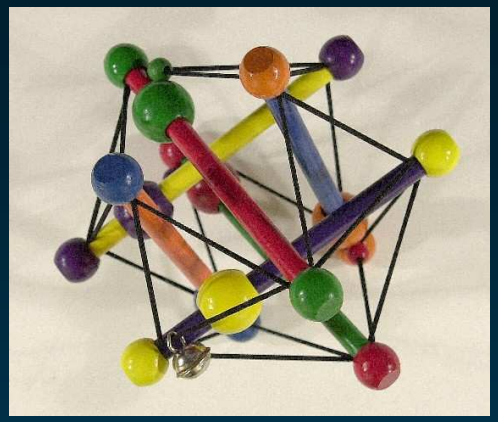
# How to address these limitations?

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

# Summary

# UMLS Overview

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



# Medical Ontology Research


Contact: olivier@nlm.nih.gov

Web: mor.nlm.nih.gov

*Olivier Bodenreider*

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

# Bibliography

# References: UMLS home page

- UMLS home page

  http:// www.nlm.nih.gov/research/umls/

- UMLS documentation
  - "Green Book"
  - online documentation

    http://www.nlm.nih.gov/research/umls/UMLSDOC.HTML

- UMLS Information web site

  http://umlsinfo.nlm.nih.gov/

# References

- UMLS as a research project
  - Lindberg, D. A., Humphreys, B. L., & McCray, A. T. (1993). The Unified Medical Language System. *Methods Inf Med*, *32*(4), 281-91.
  - Humphreys, B. L., Lindberg, D. A., Schoolman, H. M., & Barnett, G. O. (1998). The Unified Medical Language System: an informatics research collaboration. *J Am Med Inform Assoc*, *5*(1), 1-11.
- Short presentation
  - Bodenreider, O. (2004) The Unified Medical Language System (UMLS): integrating biomedical terminology. *Nucleic Acids Res*, 32(Database issue), D267-70.

# References

- Technical papers
  - McCray, A. T., & Nelson, S. J. (1995). The representation of meaning in the UMLS. *Methods Inf Med*, *34*(1-2), 193-201.
  - Campbell, K. E., Oliver, D. E., Spackman, K. A., & Shortliffe, E. H. (1998). Representing thoughts, words, and things in the UMLS. *J Am Med Inform Assoc*, *5*(5), 421-31.
- Comprehensive bibliography 1986-96

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

# Documentation and Support

# UMLS documentation and support

- UMLS homepage http://umlsinfo.nlm.nih.gov/
  - with links to all other UMLS information
- UMLSKS homepage http://umlsks.nlm.nih.gov/
  - with links to the User's and Developer's guides
- Email address for support custserv@nlm.nih.gov

# Appendix 1

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 | ISPREF | 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 | ISPREF | 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 | 3 | N | |
| 10 | MSHPJN | SY | D000224 | | 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 | | |

# Appendix 2

UMLS files in
Original Release Format

# MRCON Concepts

(2003AA)

```
CUI      LAT TS  LUI    STT   SUI   STR                LRL
C0001403|ENG|P|L0001403|PF|S0010794|Addison's Disease|0|
C0001403|ENG|P|L0001403|VC|S0352253|ADDISON'S DISEASE|0|
C0001403|ENG|P|L0001403|VO|S0010792|Addison Disease|0|
C0001403|ENG|P|L0001403|VO|S0033587|Disease, Addison|0|
C0001403|ENG|P|L0001403|VO|S0469271|Addison's disease, NOS|3|
C0001403|ENG|S|L0278071|PF|S0352321|ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)|0|
C0001403|ENG|S|L0278422|PF|S0352329|ADRENOCORTICAL INSUFFICIENCY, PRIMARY FAILURE|0|
C0001403|ENG|S|L0367999|PF|S0469267|Addison melanoderma|3|
C0001403|ENG|S|L0368000|PF|S0496840|Melasma addisonii|3|
C0001403|ENG|S|L0368398|PF|S0506528|Primary adrenal deficiency|3|
C0001403|ENG|S|L0373744|PF|S0471237|Asthenia pigmentosa|3|
C0001403|ENG|S|L0377831|PF|S0473611|Bronzed disease|3|
C0001403|ENG|S|L0494940|PF|S0718028|Primary adrenocortical insufficiency|3|
C0001403|ENG|s|L0494937|PF|S0718027|Primary adrenocortical insuff|3|
C0001403|FIN|P|L1510041|PF|S1805950|Addisonin tauti|3|
C0001403|FRE|S|L1272481|PF|S1514427|MALADIE D'ADDISON|2|
C0001403|GER|P|L1229627|PF|S1471573|Addison-Krankheit|3|
C0001403|GER|S|L1288823|PF|S1530769|Primaere Nebennierenrindeninsuffizienz|1|
C0001403|ITA|P|L1276837|PF|S1518783|Morbo di Addison|3|
C0001403|POR|P|L0324623|PF|S0432928|DOENCA DE ADDISON|2|
C0001403|RUS|P|L0889403|PF|S1093220|ADDISONOVA BOLEZN'|3|
C0001403|SPA|P|L0342625|PF|S0450930|ENFERMEDAD DE ADDISON|3|
[…]
```

# MRSO Sources

(2003AA)

```
CUI      LUI      SUI      SAB TTY  SCD   SRL
C0001403|L0001403|S0010792|MSH|EN|D000224|0|
C0001403|L0001403|S0010794|MSH|MH|D000224|0|
C0001403|L0001403|S0010796|MSH|PM|D000224|0|
C0001403|L0001403|S0010796|PSY|PT|00810|3|
C0001403|L0001403|S0033587|MSH|PM|D000224|0|
C0001403|L0001403|S0220088|MSH|PM|D000224|0|
C0001403|L0001403|S0352252|CCPSS|PT|0022753|3|
C0001403|L0001403|S0352252|DXP|SY|NOCODE|0|
C0001403|L0001403|S0352253|CST|GT|ADREN INSUFFIC|0|
C0001403|L0001403|S0352253|WHO|IT|0410|2|
C0001403|L0001403|S0354372|AOD|DE|0000005430|0|
C0001403|L0001403|S0354372|CSP|PT|0060-3321|0|
C0001403|L0001403|S0354372|LCH|PT|U000061|0|
C0001403|L0001403|S0354372|MDR|LT|10001130|3|
C0001403|L0001403|S0354372|RCD|PT|C1541|3|
C0001403|L0001403|S0354372|SNM|SY|D-2332|3|
C0001403|L0001403|S0365923|CST|GT|ADREN INSUFFIC|0|
C0001403|L0001403|S0469271|SNMI|PT|DB-70620|3|
C0001403|L0001403|S1619433|MDR|LT|10001130|3|
C0001403|L0001403|S1911394|ICPC2P|PT|T99002|3|
C0001403|L0001403|S1921523|MTHICD9|ET|255.4|0|
C0001403|L0001403|S1932462|ICPC2P|SF|T99002|3|
[…]
```

# MRDEF Definitions

(2003AA)

```
CUI      SAB     DEF
C0001403|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.|
[…]
```

# MRSTY Semantic Types (2003AA)

```
CUI      TUI  STY
C0001400|T040|Organism Function|
C0001403|T047|Disease or Syndrome|
C0001406|T083|Geographic Area|
C0001407|T114|Nucleic Acid, Nucleoside, or Nucleotide|
C0001407|T123|Biologically Active Substance|
[…]
```

# MRATX Associated Expressions (2003AA)

```
CUI       SAB REL     ATX
```

*Closed fracture of malar and maxillary bones, NOS*

```
C0009045|MSH|RB|<Zygomatic Fractures> OR <Maxillary Fractures>|
```

*Unilateral congenital dislocation of hip*

```
C0009702|MSH|RB|<Hip Dislocation, Congenital> AND <Femur Head>/<abnormalities>|
```

*Suture of bladder*

```
C0010700|MSH|RB|<Bladder>/<surgery>|
```

*Corneal abrasion*

```
C0010032|MSH|RO|<Cornea>/<injuries>|
```

*CORRECTIVE LENS PROBLEM*

```
C0010099|MSH|RO|<Contact Lenses>/<adverse effects>|
```

*Chronic cough*

```
C0010201|MSH|SY|<Cough> AND <Chronic Disease>|
```

*Cyst and pseudocyst of pancreas*

```
C0010623|MSH|SY|<Pancreatic Cyst> OR <Pancreatic Pseudocyst>|
```

*Cystitis*

```
C0010692|LCH|RU|<Bladder>/<Inflammation>|
[…]
```

# MRCXT Contexts

(2003AA)

```
CUI      SUI      SAB  SCD    CXN CXL RNK   CXS                CUI2      HCD REL XC
C0001403|S0469271|SNMI|DB-70620|1|ANC|1|SNOMED International|C1140118||||
C0001403|S0469271|SNMI|DB-70620|1|ANC|2|DISEASES/DIAGNOSES|C0338067||||
C0001403|S0469271|SNMI|DB-70620|1|ANC|3|DISEASES OF THE END. SYSTEM|C0014130||||
C0001403|S0469271|SNMI|DB-70620|1|ANC|4|DISEASES OF THE ADRENAL GLANDS|C0001621||||
C0001403|S0469271|SNMI|DB-70620|1|CCP||Addison's disease, NOS|C0001403|DB-70620|||

(* = C0001403|S0718028|ICD10)
*|E27.1|1|ANC|1|ICD…, Tenth Revision (ICD-10)|C1140143||||
*|E27.1|1|ANC|2|Endocrine, nutritional and metabolic diseases|C0694452|E00-E90.9|||
*|E27.1|1|ANC|3|Disorders of other endocrine glands|C0178257|E20-E35.9|||
*|E27.1|1|ANC|4|Other disorders of adrenal gland|C0494313|E27|||
*|E27.1|1|CCP||Primary adrenocortical insufficiency|C0001403|E27.1|||

(* = C0001403|S0010794|MSH)
*|D000224|1|ANC|1|MeSH|C1135584||||
*|D000224|1|ANC|2|MeSH Descriptors|C1135587||||
*|D000224|1|ANC|3|Index Medicus Descriptor|C1135589||||
*|D000224|1|ANC|4|Diseases (MeSH Category)|C0012674|C|||
*|D000224|1|ANC|5|Endocrine Diseases|C0014130|C19|||
*|D000224|1|ANC|6|Adrenal Gland Diseases|C0001621|C19.53|||
*|D000224|1|ANC|7|Adrenal Gland Hypofunction|C0001623|C19.53.264|||
*|D000224|1|CCP||Addison's Disease|C0001403|C19.53.264.263|||
*|D000224|1|SIB||Adrenoleukodystrophy|C0001661|C19.53.264.270|||
*|D000224|1|SIB||Hypoaldosteronism|C0020595|C19.53.264.480|||
```

# MRSAT Simple concept attributes (2003AA)

```
CUI      LUI      SUI      SCD    ATN SAB     ATV
C0001403|L0001403|S0010792|D000224|DID|MSH|D000224|
C0001403|L0001403|S0010792|D000224|EV|MSH|ADDISON DIS|
C0001403|L0001403|S0010792|D000224|MUI|MSH|M0000346|
C0001403|L0001403|S0010792|D000224|TH|MSH|UNK (19XX)|
C0001403|L0001403|S0010794|D000224|AN|MSH|an autoimmune dis with adrenal hypofunction|
C0001403|L0001403|S0010794|D000224|AQL|MSH|BL CF CI CL CN CO DH DI DT EC EH EM EN …|
C0001403|L0001403|S0010794|D000224|DC|MSH|1|
C0001403|L0001403|S0010794|D000224|DID|MSH|D000224|
C0001403|L0001403|S0010794|D000224|EV|MSH|ADDISON DIS|
C0001403|L0001403|S0010794|D000224|MDA|MSH|19990101|
C0001403|L0001403|S0010794|D000224|MED1963|NLM-MED|*2|
C0001403|L0001403|S0010794|D000224|MED1963|NLM-MED|2|
[…]
C0001403|L0001403|S0010794|D000224|MED2002|NLM-MED|*19|
C0001403|L0001403|S0010794|D000224|MED2002|NLM-MED|23|
[…]
C0001403|L0001403|S0010794|D000224|MN|MSH|C19.53.264.263|
C0001403|L0001403|S0010794|D000224|MN|MSH|C20.111.163|
[…]
C0001403|L0001403|S0469271|DB-70620|SIC|SNMI|255.4|
[…]
C0001403||||DA|MTH|19900930|
C0001403||||MR|MTH|20021026|
C0001403||||ST|MTH|R|
```

# MRRANK Name Ranking

(2003AA)

```
RANK SAB TTY SUPRES
0401|MTH|PN|N|
0400|MTH|MM|N|
0399|MSH|MH|N|
0398|MSH|TQ|N|
0397|MSH|EP|N|
0396|MSH|EN|N|
0395|MSH|XQ|N|
0394|MSH|NM|N|
0393|RXNORM|SCD|N|
0392|RXNORM|SCDC|N|
0391|DSM4|PT|N|
0390|DSM3R|PT|N|
0389|SNMI|PT|N|
0388|SNMI|PX|Y|
0387|SNMI|HT|N|
0386|SNMI|HX|Y|
0385|VANDF|CD|N|
0384|VANDF|HT|N|
0383|VANDF|IN|N|
0382|MDDB|CD|N|
0381|MMX|CD|N|
0380|MMX|IN|N|
0379|RCDSA|PT|N|
[…]
```

# MRREL Inter-concept Relationships (2003AA)

```
CUI1     REL CUI2      RELA   SAB  SL       MG
C0001403|AQ|C0348026||MSH|MSH||
C0001403|CHD|C0342477||RCD|RCD||
C0001403|CHD|C0546992||RCD|RCD||
C0001403|PAR|C0001621||PSY|PSY||
C0001403|PAR|C0001621||SNMI|SNMI||
C0001403|PAR|C0001623||MSH|MSH||
C0001403|PAR|C0935495|has_member|PSY|PSY||
C0001403|RB|C0001621||PSY|PSY||
C0001403|RB|C0001623||MTH|MTH||
C0001403|RB|C0004364||CSP|CSP||
C0001403|RB|C0004364||MTH|MTH||
C0001403|RL|C0405580|mapped_from|SNMI|SNMI||
C0001403|RN|C0518933||MTH|MTH||
C0001403|RN|C0518934||MTH|MTH||
C0001403|RO|C0152889|associated_with|SNMI|SNMI||
C0001403|RO|C0546992||MTH|MTH||
C0001403|RQ|C0020615|clinically_associated_with|CCPSS|CCPSS||
C0001403|RQ|C0151467|clinically_similar|RAM|RAM||
C0001403|RQ|C0300942|classifies|MDR|MDR||
C0001403|RQ|C0405580|mapped_from|CST|CST||
C0001403|RQ|C0405580|mapped_to|HLREL|HLREL||
C0001403|RQ|C0740740|inverse_isa|CCPSS|CCPSS||
C0001403|SIB|C0001206||MDR|MDR||
[…]
```

# MRCOC Co-occurrences (2003AA)

```
CUI1     CUI2    SOC COT COF COA
C0001403|C0000727|MED|L|1|CO=1,DI=1,ME=1|
C0001403|C0000737|MBD|L|1|CO=1,DI=1|
C0001403|C0000833|MED|L|2|MI=2,DT=1,RA=1|
C0001403|C0001175|MBD|L|1|CO=1|
C0001403|C0001418|MED|L|1|ET=1|
C0001403|C0001430|MBD|L|1|BL=1,CO=1|
C0001403|C0001551|MED|L|3|DT=3|
C0001403|C0001613|MBD|L|6|ET=2,IM=2,CL=1,CN=1,DI=1,PA=1,PP=1|
C0001403|C0001613|MED|L|6|IM=4,PP=3,CO=2,BL=1,DI=1,TH=1|
C0001403|C0001614|MBD|L|1|BL=1,CI=1|
C0001403|C0001617|MBD|L|1|BL=1|
C0001403|C0001618|MBD|L|2|BL=2,CO=1,ET=1|
C0001403|C0001618|MED|L|1|CO=1,PA=1|
[…]
C0018099|C0151373|AIR|KP|||
C0018099|C0151407|AIR|KP|||
C0018099|C0151463|CCPSS|PP|1||
C0018099|C0205082|CCPSS|MP|1||
C0018099|C0205090|CCPSS|MP|8||
C0018099|C0205091|CCPSS|MP|2||
C0018099|C0221598|AIR|KP|||
[…]
```

# MRCON Suppressible synonyms (2003AA)

```
CUI      LAT TS  LUI    STT   SUI    STR                LRL
C0001403|ENG|P|L0001403|PF|S0010794|Addison's Disease|0|
C0001403|ENG|P|L0001403|VC|S0352253|ADDISON'S DISEASE|0|
C0001403|ENG|P|L0001403|VO|S0010792|Addison Disease|0|
C0001403|ENG|P|L0001403|VO|S0033587|Disease, Addison|0|
C0001403|ENG|P|L0001403|VO|S0469271|Addison's disease, NOS|3|
C0001403|ENG|S|L0278071|PF|S0352321|ADRENAL INSUFFICIENCY (ADDISON'S DISEASE)|0|
C0001403|ENG|S|L0278422|PF|S0352329|ADRENOCORTICAL INSUFFICIENCY, PRIMARY FAILURE|0|
C0001403|ENG|S|L0367999|PF|S0469267|Addison melanoderma|3|
C0001403|ENG|S|L0368000|PF|S0496840|Melasma addisonii|3|
C0001403|ENG|S|L0368398|PF|S0506528|Primary adrenal deficiency|3|
C0001403|ENG|S|L0373744|PF|S0471237|Asthenia pigmentosa|3|
C0001403|ENG|S|L0377831|PF|S0473611|Bronzed disease|3|
C0001403|ENG|S|L0494940|PF|S0718028|Primary adrenocortical insufficiency|3|
C0001403|ENG|S|L0494937|PF|S0718027|Primary adrenocortical insuff|3|
C0001403|FIN|P|L1510041|PF|S1805950|Addisonin tauti|3|
C0001403|FRE|S|L1272481|PF|S1514427|MALADIE D'ADDISON|2|
C0001403|GER|P|L1229627|PF|S1471573|Addison-Krankheit|3|
C0001403|GER|S|L1288823|PF|S1530769|Primaere Nebennierenrindeninsuffizienz|1|
C0001403|ITA|P|L1276837|PF|S1518783|Morbo di Addison|3|
C0001403|POR|P|L0324623|PF|S0432928|DOENCA DE ADDISON|2|
C0001403|RUS|P|L0889403|PF|S1093220|ADDISONOVA BOLEZN'|3|
C0001403|SPA|P|L0342625|PF|S0450930|ENFERMEDAD DE ADDISON|3|
[…]
```

# MRCUI Concept history

(2003AA)

```
CUI1     VER    CREL  CUI2  MAPIN
C0241779|1996AA|SY|C0001403|Y|
C0271735|1996AA|SY|C0001403|Y|
[…]
```

# MRSAB Source information (2003AA)

```
VCUI     RCUI     VSAB    RSAB SON SF SVER MSTART MEND IMETA RMETA SLC SCC SRL TFR
C1140103|C1140104|INS2002|INS|French translation of the Medical Subject Headings,
2002|MSH|2002|2002_04_11||2002AB||Dr. Annie Advocat; e-mail: advocat@inserm-
dicdoc.u-strasbg.fr|Dr. Annie Advocat; e-mail: advocat@inserm-dicdoc.u-
strasbg.fr|3|30883|20692||MH,SY||FRE|ISO646-US|Y|Y|

C1140132|C1140133|BRMP2002|BRMP|Portuguese translation of the Medical Subject
Headings, 2002|MSH|2002|2001_12_04||2002AA||Elenice de Castro; e-mail:
elenice@brm.bireme.br|Elenice de Castro; e-mail:
elenice@brm.bireme.br|3|41853|27195||EP,MH,SY||POR|ISO646-US|Y|Y|

C1140297|C1140298|DUT2001|DUT|Dutch Translation of the Medical Subject Headings,
2001|MSH|2001|2001_12_04||2002AB||A.J.P.M.Overbeke, overbeke@ntvg.nl, * 20 662
0150|A.J.P.M.Overbeke, overbeke@ntvg.nl, * 20 662
0150|3|35705|17733||EP,MH,SY||DUT|ISO646-US|Y|Y|

C1142630|C1135584|MSH2003_2002_10_24|MSH|Medical Subject Headings,
2002_10_24|MSH|2003_2002_10_24|2002_11_05||2003AA||Stuart Nelson, M.D., Head, MeSH
Section; e-mail: nelson@nlm.nih.gov|Stuart Nelson, M.D., Head, MeSH Section; e-
mail: nelson@nlm.nih.gov|0|516015|231458|FULL-
MULTIPLE|CE,EN,EP,HS,HT,MH,N1,NM,PM,TQ,XQ|AN,AQL,CX,DC,DID,DQ,DS,DX,EC,EV,FR,FX,HM
,HN,II,LT,MDA,MMR,MN,MUI,OL,PA,PI,PM,QA,QE,QS,RN,RR,SOS,SRC,TH|ENG|ISO646-US|Y|Y|
```

# SRDEF Basic information

(2003AA)

```
RT  TUI  STY/RL  STN/RTN  DEF     EX     UN     NH     ABR     RIN
STY|T001|Organism|A1.1|Generally, a living individual, including all plants and
animals.|Homozygote; Radiation Chimera; Sporocyst|||||
STY|T002|Plant|A1.1.1|An organism having cellulose cell walls, growing by
synthesis of inorganic substances, generally distinguished by the presence of
chlorophyll, and lacking the power of locomotion.  Plant parts are included here
as well.|Pollen; Potatoes; Vegetables|||||
STY|T003|Alga|A1.1.1.1|A chiefly aquatic plant that contains chlorophyll, but does
not form embryos during development and lacks vascular tissue.|Chlorella;
Laminaria; Seaweed|||||
STY|T004|Fungus|A1.1.2|A eukaryotic organism characterized by the absence of
chlorophyll and the presence of a rigid cell wall.  Included here are both slime
molds and true fungi such as yeasts, molds, mildews, and mushrooms.|Aspergillus
clavatus; Blastomyces; Helminthosporium; Neurospora|||||
[…]
RL|T132|physically_related_to|R1|Related by virtue of some physical attribute or
characteristic.||||PR|physically_related_to|
RL|T133|part_of|R1.1|Composes, with one or more other physical units, some larger
whole.  This includes component of, division of, portion of, fragment of, section
of, and layer of.||||PT|has_part|
[…]
RL|T186|isa|H|The basic hierarchical link in the Network.  If one item "isa"
another item then the first item is more specific in meaning than the second
item.||||IS|inverse_isa|
[…]
```

# SRSTR Structure

(2003AA)

```
STY/RL                RL      STY/RL                                  LS
Biologic Function|affects|Organism|D|
Biologic Function|isa|Natural Phenomenon or Process|D|
Biologic Function|process_of|Organism|D|
Biologic Function|produces|Biologically Active Substance|D|
Biologic Function|produces|Body Substance|D|
[…]
Disease or Syndrome|conceptually_related_to|Experimental Model of Disease|DNI|
Disease or Syndrome|isa|Pathologic Function|D|
Disease or Syndrome|produces|Tissue|D|
[…]
Medical Device|isa|Manufactured Object|D|
Medical Device|prevents|Injury or Poisoning|D|
Medical Device|prevents|Pathologic Function|D|
Medical Device|treats|Anatomical Abnormality|D|
Medical Device|treats|Injury or Poisoning|D|
Medical Device|treats|Pathologic Function|D|
Medical Device|treats|Sign or Symptom|D|
[…]
Mental Process|process_of|Plant|B|  blocks  Biologic Function|process_of|Organism|D|
[…]
part_of|isa|physically_related_to|D|
[…]
```

# SRSTRE2 Structure (expanded) (2003AA)

```
STY                    RL          STY
Disease or Syndrome|isa|Pathologic Function|
Disease or Syndrome|isa|Biologic Function|
Disease or Syndrome|isa|Natural Phen. or Pr.|
Disease or Syndrome|isa|Phenomenon or Process|
Disease or Syndrome|isa|Event|
Disease or Syndrome|affects|Alga|
Disease or Syndrome|affects|Amphibian|
Disease or Syndrome|affects|Animal|
Disease or Syndrome|affects|Archaeon|
Disease or Syndrome|affects|Bacterium|
Disease or Syndrome|affects|Biologic Function|
Disease or Syndrome|affects|Bird|
Disease or Syndrome|affects|Cell Function|
Disease or Syndrome|affects|Cell or Molecular Dysfunction|
[…]
```

```
Pathologic Function |isa|Biologic Function|
Biologic Function|isa|Natural Phen. or Process|
Natural Phen. or Process|isa|Phen. or Process|
Phenomenon or Process|isa|Event|
```

from `Biologic Function|affects|Organism|D|`