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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

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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

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Part I
What is the UMLS?

(1) Introduction

What does UMLS stand for?

- Unified
- Medical
- Language
- System

UMLS®
Unified Medical Language System®
UMLS Metathesaurus®

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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.»

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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

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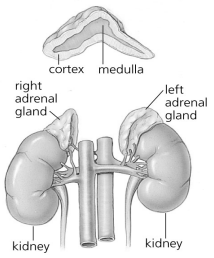
Part I
What is the UMLS?

(2) Overview through an example

Addison's disease

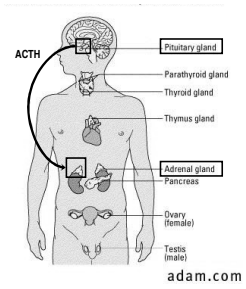
- Addison's disease is a rare endocrine disorder
- Addison's disease occurs when the adrenal glands do not produce enough of the hormone cortisol
- For this reason, the disease is sometimes called chronic adrenal insufficiency, or hypocortisolism



NLM 10

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



NLM 11

Addison's disease: Symptoms

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

NLM 12

AD in medical vocabularies

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

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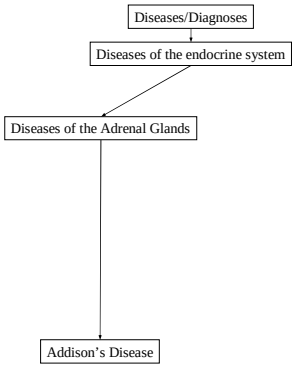
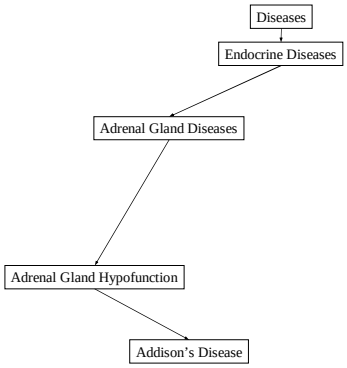
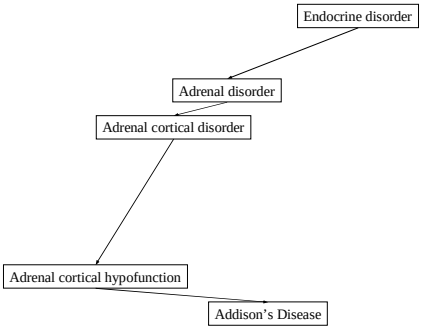
SNOMED International



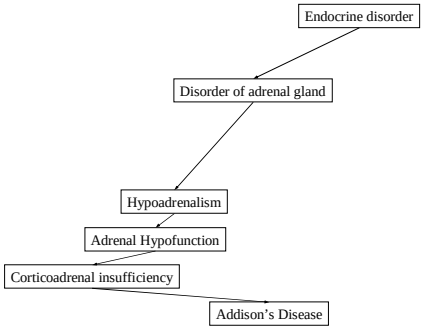
MeSH

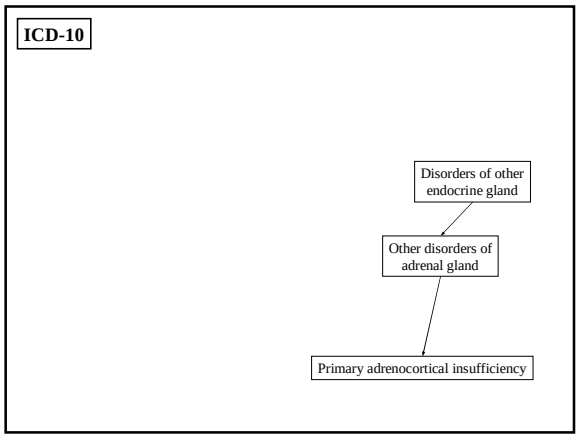


AOD



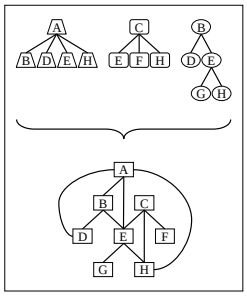
Read Codes





Organize concepts

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



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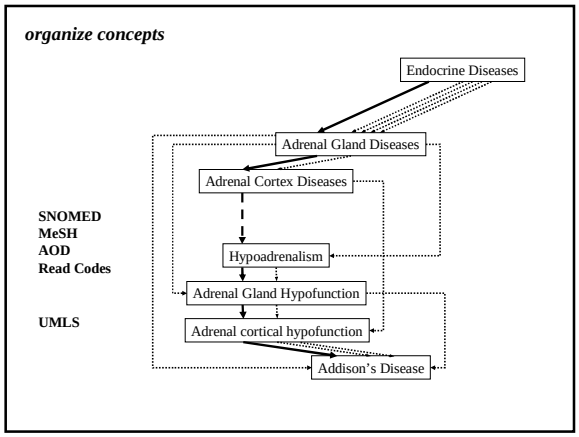
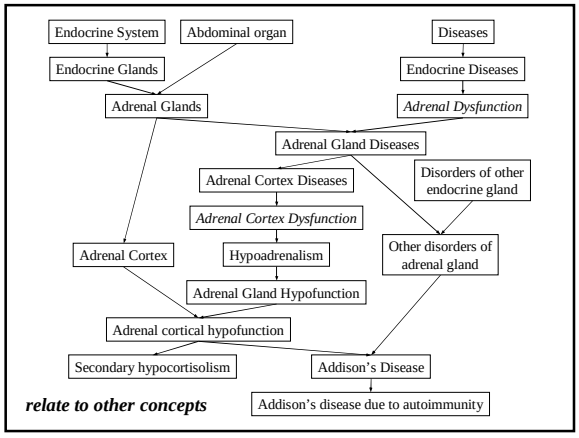


Relate to other concepts

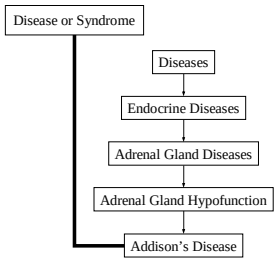
- Additional hierarchical relationships
  - link to other trees
  - make relationships explicit
- Non-hierarchical relationships
- Co-occurring concepts
- Mapping relationships

NLM 22



Categorize concepts

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



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How do they do that?

- Lexical knowledge
- Semantic pre-processing
- UMLS editors

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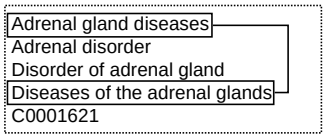
Lexical knowledge



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Semantic pre-processing

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

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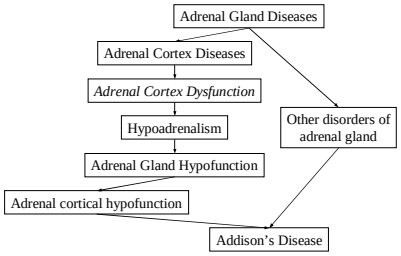
Additional knowledge: UMLS editors



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UMLS Summary

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

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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

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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

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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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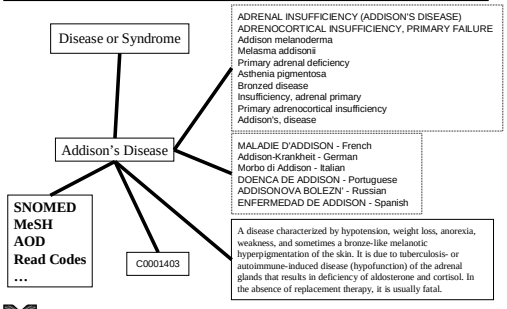
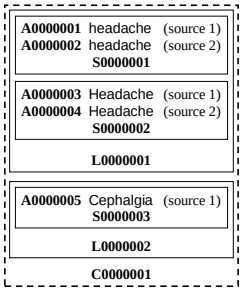
Addison's Disease: Concept



NLM 37

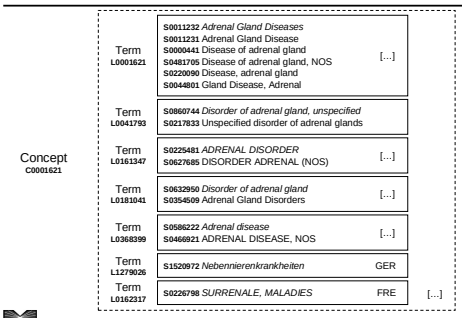
Metathesaurus Concepts (2005AA)

- 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



NLM 38

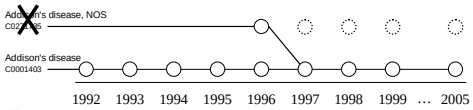
Cluster of synonymous terms



NLM 39

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



NLM 40

Metathesaurus Relationships

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

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

NLM 41

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

NLM 42

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**

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Symbolic relationships Attribute

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

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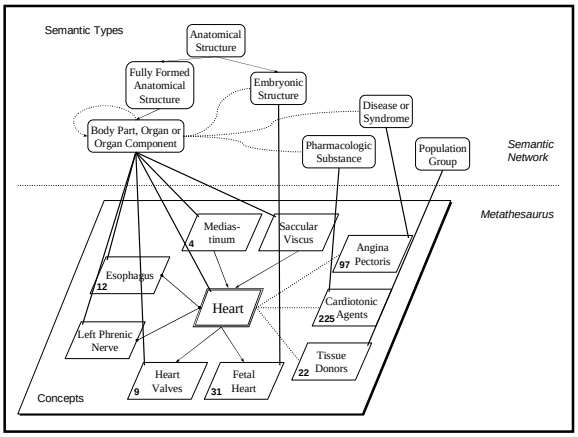

UMLS Semantic Network

Semantic Network

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

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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

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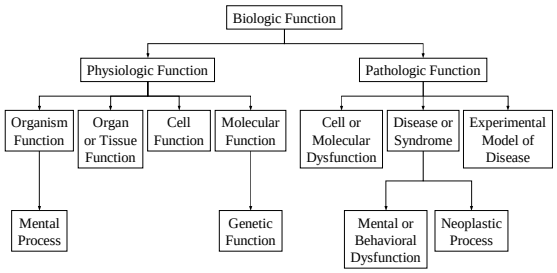
"Biologic Function" hierarchy (isa)



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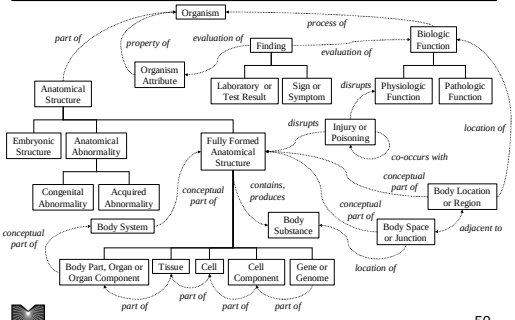
Associative (non-isa) relationships



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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

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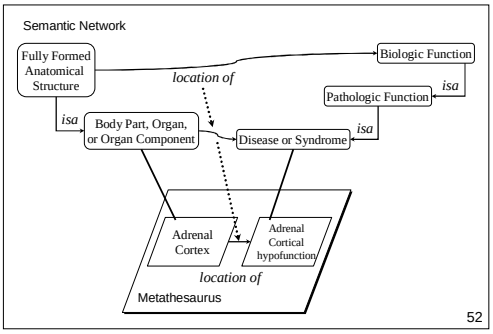
Relationships can inherit semantics



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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

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Morphology

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

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Orthography

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

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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

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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

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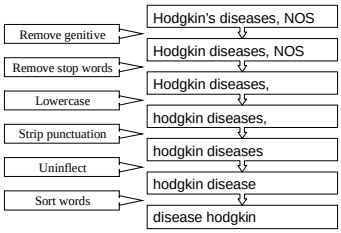
Normalization



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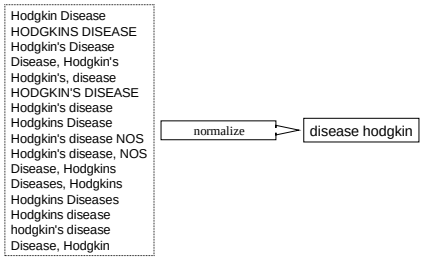
Normalization: Example



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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

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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

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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)

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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

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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
  - Read appendix
  - Print a copy for your records
  - Complete the Web form

  Accept and continue
  Accept
  Printer-friendly Version
  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

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

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Unified Medical Language System

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    12. 2. Category 2:

    12. 2. Category 2:

    All category 1 restrictions AND

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APPENDIX A.1

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

UMLS METATHESAURUS® SOURCE VOCABULARIES -- 2004AB Edition

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 Biling 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

---

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
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76

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)

78

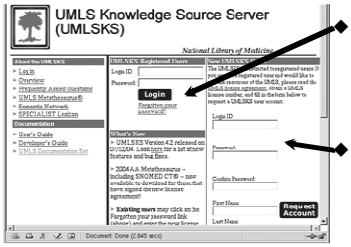
Knowledge Source Server
Web search interface

UMLSKS Web search interface

- Logging in
- Basic searching
- Advanced searching

80

UMLSKS Web search interface log in

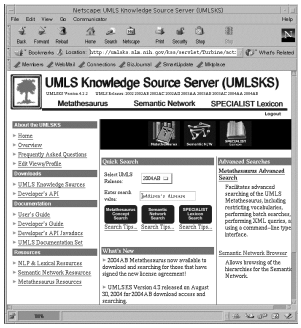


- Returning users log in
- New users create account

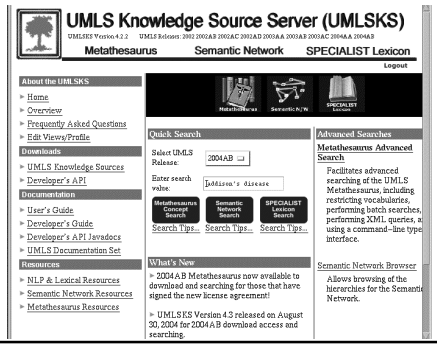
81

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



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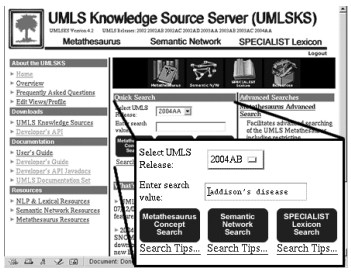
Metathesaurus Basic Search
Addison's disease



- UMLS Release
- Search Term
- UMLS Knowledge Source

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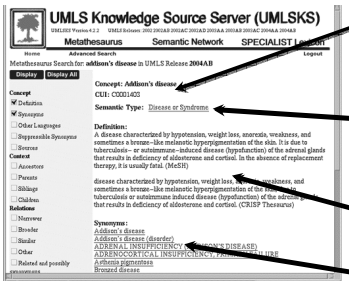
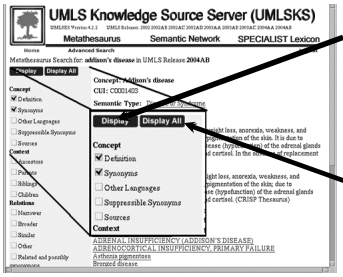
Concept Report *Addison's disease*



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

85

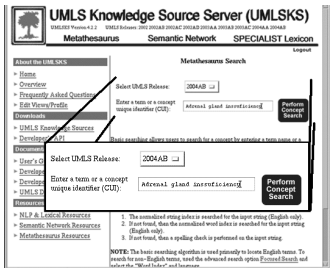
Display All



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

86

Metathesaurus Basic Search *Adrenal gland insufficiency*



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

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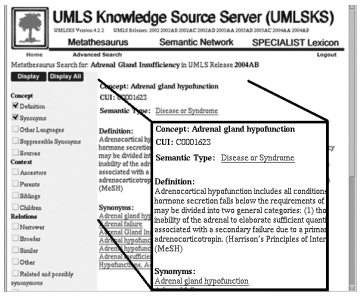
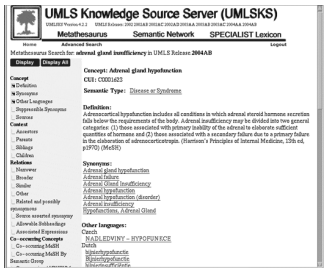
Basic Concept Report *Adrenal gland insufficiency*



88

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

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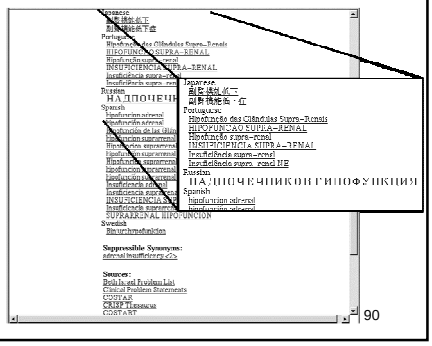
Concept Report Display All (continued)

Synonyms

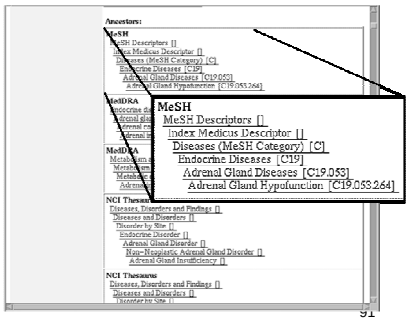
Sources



90

Concept Report Display All (continued)

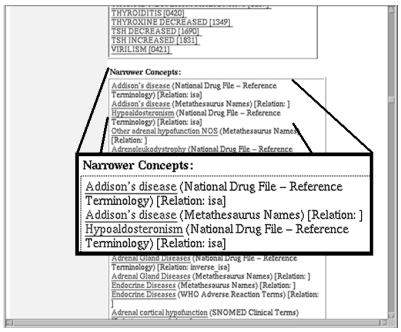
Hierarchies



91

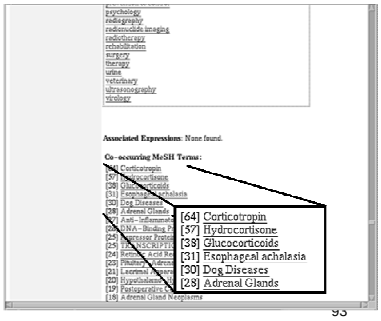
Concept Report Display All (continued)

Relations



Concept Report Display All (continued)

Co-occurrence data



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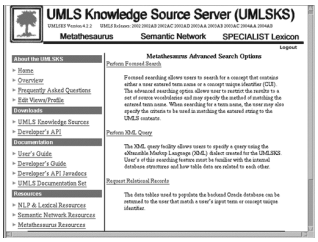
Metathesaurus Advanced Search Options

- Focused Search
- Raw Relational Records



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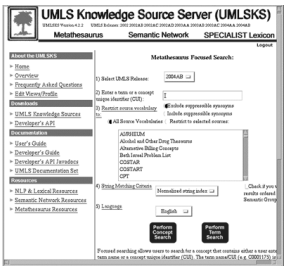
Metathesaurus Advanced Search Feature Focused Search



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

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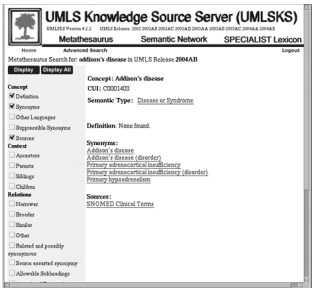
Restricted Source Concept Report *Addison's Disease*



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

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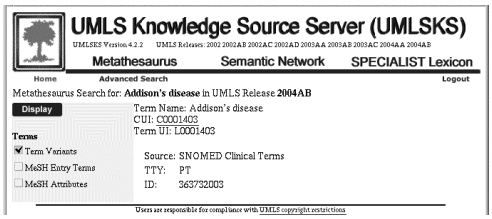
Addison's disease in SNOMED CT Preferred Term and Code



- TTY: Term Type
- ID: Source Code Descriptor

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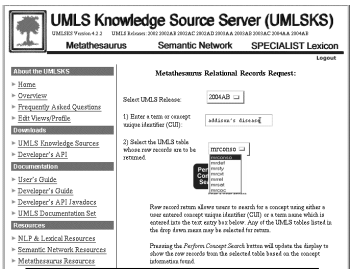
Metathesaurus Advanced Search Feature Relational Record Request

- UMLS Release
- Search Term
- UMLS Relational Table



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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|||I0001130|MDRDUT|LT|I0001130|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|||CD|DUT|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|||I0036696|MDRDUT|LT|I0036696|primair hypoadr
C0001403|DUT|S|L4999270|PF|S5686775|N|A6627493|||I0052381|MDRDUT|LT|I0052381|primaire bijnie
C0001403|DUT|S|L4999270|PF|S5686775|Y|A6627494|||I0052381|MDRDUT|PT|I0052381|primaire bijnie
C0001403|DUT|S|L5012413|PF|S5699917|Y|A6645695|||I0013096|MDRDUT|LT|I0013096|ziekte van Addi
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388276|||ACD|DE|D000000601|2|Addison's disease|0||
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388277|||0060-3321|CSP|PT|0060-3321|Addison's disea
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388279|||LCH|PT|V000061|Addison's disease|0||
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388280|||I0001390|MDR|LT|I0001130|Addison's disease|
C0001403|ENG|P|L0001403|PF|S0354372|N|A0388281|||RCD|PT|C1541|Addison's disease|3||
```

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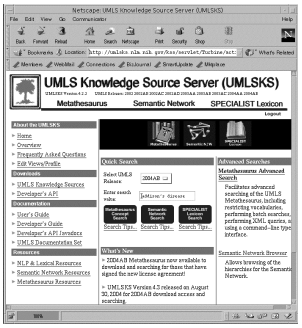
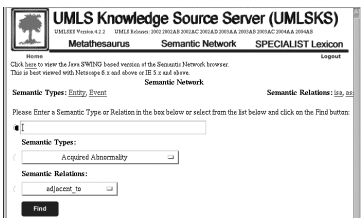
Semantic Network Searching

- 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

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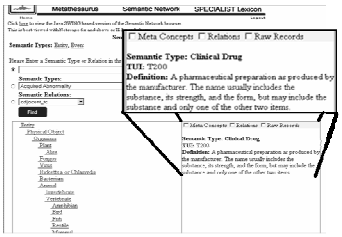
Semantic Type Clinical Drug



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

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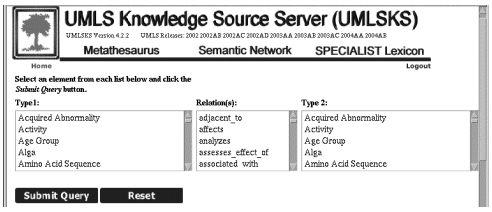
Show Relations Between Types



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

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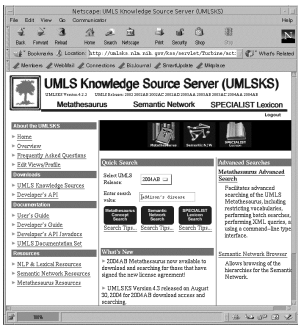
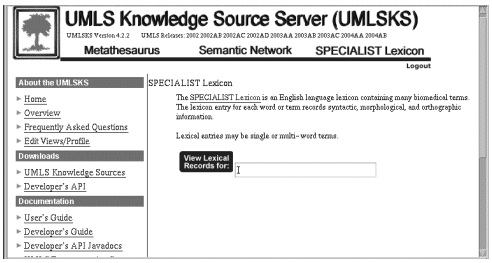
SPECIALIST Lexicon Searching

- Select Tab along top
- Quick search



SPECIALIST Lexicon Search



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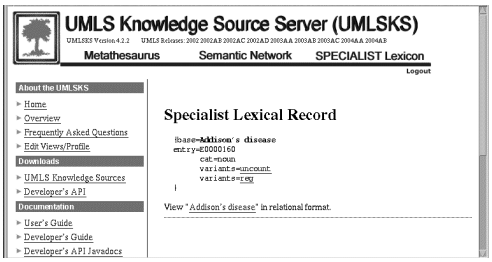
SPECIALIST Lexical Record



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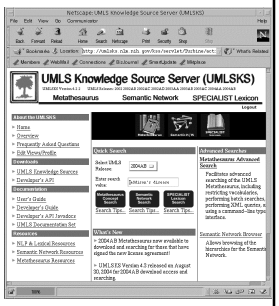
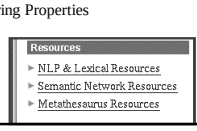
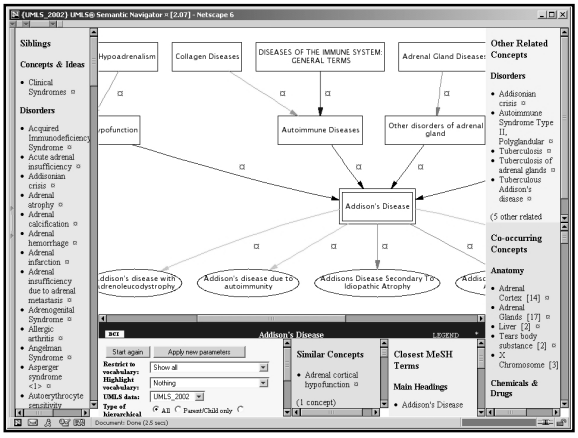
UMLS Resources

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





107



Knowledge Source Server
Application Programming Interface

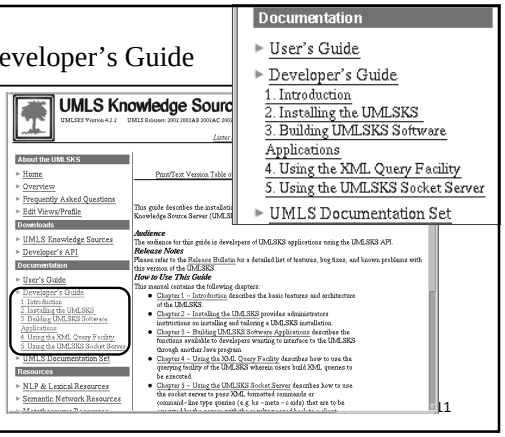
UMLSKS API basics

- Remote server at NLM
- Local application connected through

Java RMI	TCP/IP socket
◆ 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	◆ XML-based queries ◆ Developer's Guide: Chapter 5 ◆ XML schema ◆ Socket server • Host: umlsks.nlm.nih.gov • Port: 8042

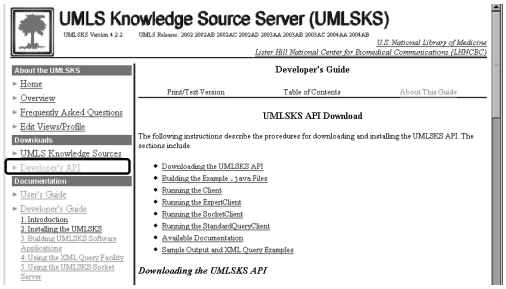
NLM 110

Developer's Guide



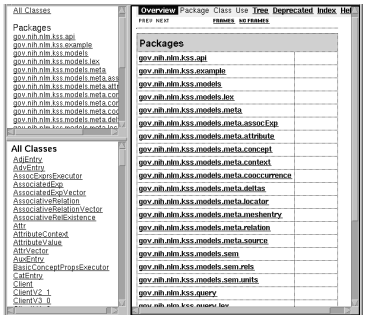
111

Documentation Java API



NLM 112

Documentation Javadocs



NLM 113

Sample XML query (1) Current version

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

⇨

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

NLM 114

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>
```

NLM 115

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>
```

NLM 116

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>
[…]
```

NLM 117

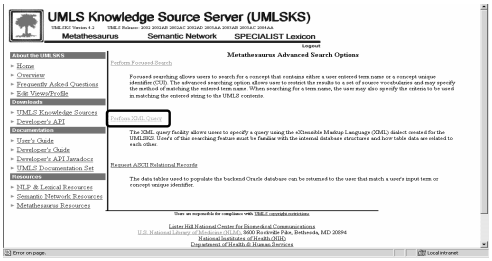
Sample XML query (5) All semantic type IDs

```
<?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>
 […]
```

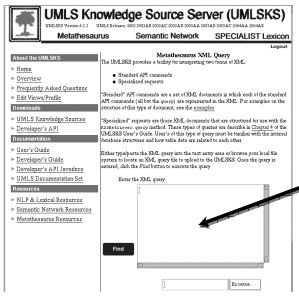
NLM 118

Performing XML queries from UMLSKS



NLM 119

Performing XML queries from UMLSKS




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

NLM 120

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

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Using MetamorphoSys

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

123

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

124

Why use MetamorphoSys?

Changing Default Settings

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

125

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

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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

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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!

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...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

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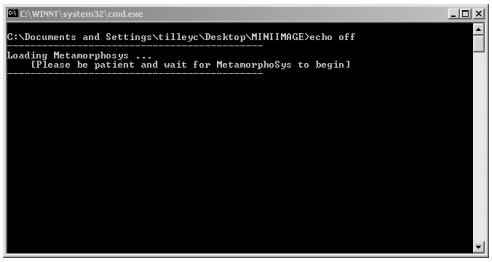
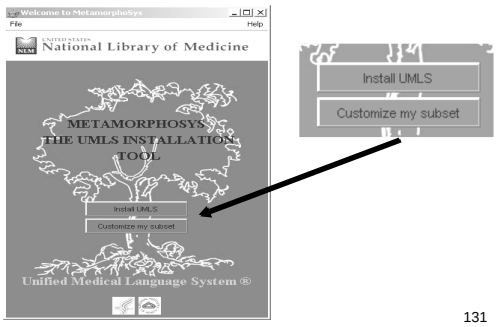
Be patient! A lot of software must load.



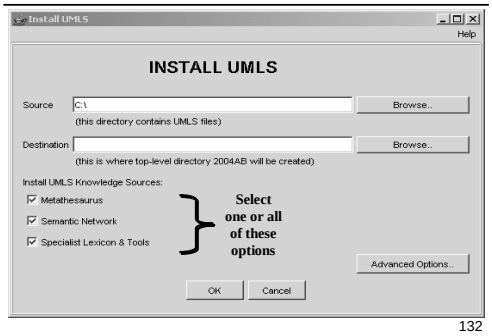
130

Welcome Screen



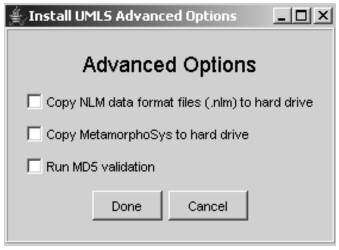
131

Install UMLS



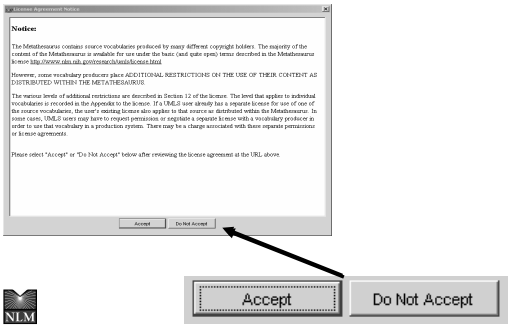
132

Install UMLS Advanced Options

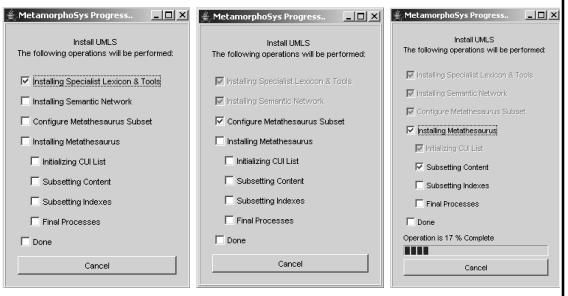


133

UMLS License Notice

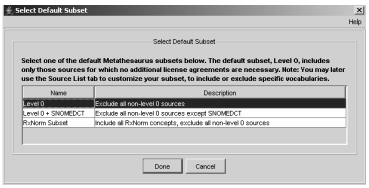


Installation progress monitor



135

Select a default subset



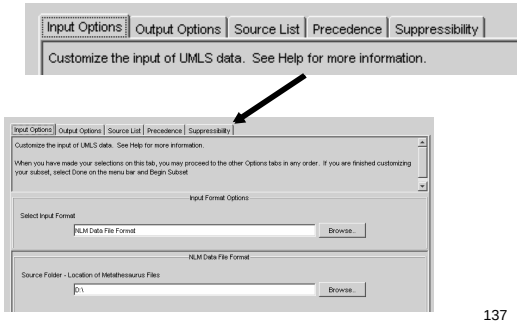
Level 0 → no separate additional license agreements

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

RxNorm → no separate additional license agreements

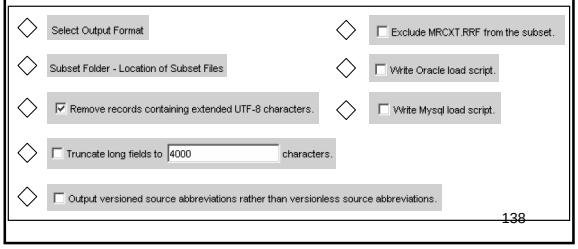
136

Input Options Tab



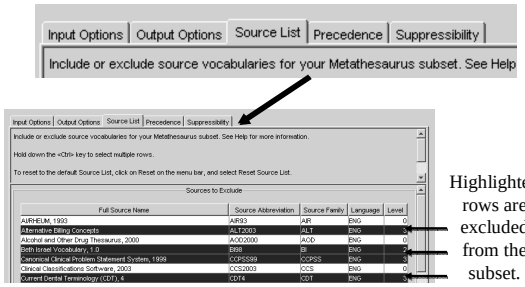
137

Output Options Tab



138

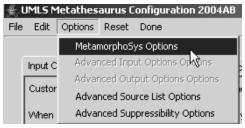
Source List Tab



Highlighted rows are excluded from the subset.

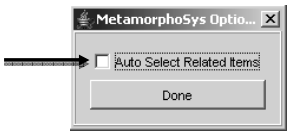
139

MetamorphoSys Option Tab



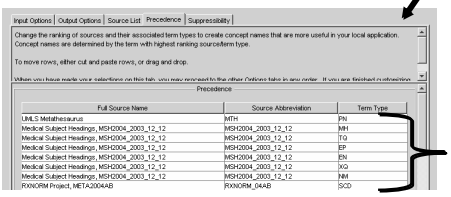
Source list behavior can be changed using the MetamorphoSys Option Tab

If you wish to Auto Select Related Items check this box



Precedence Tab

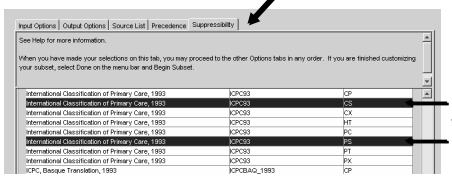
- Ranks names by types of terms within sources
- Highest ranking name determines the Preferred Name



Cut and paste rows to alter the preferred name

141

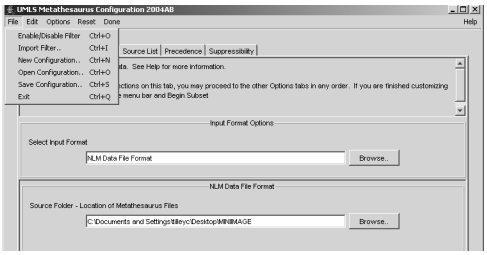
Suppressibility Tab



Highlighted source term types will be marked as suppressible

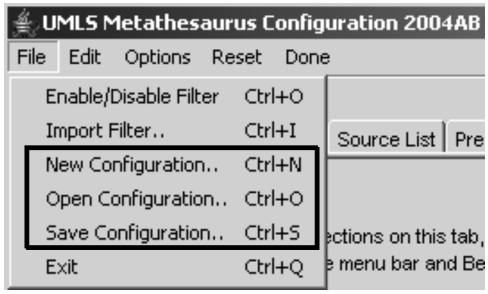
142

File menu



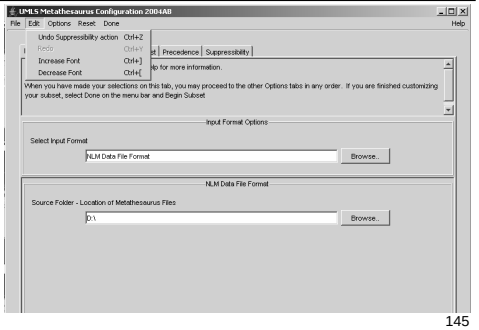
143

File menu



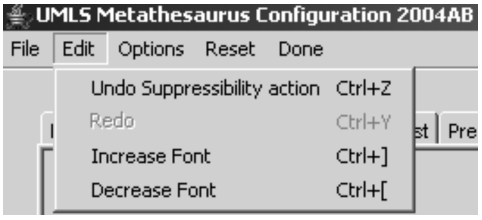
144

Edit menu



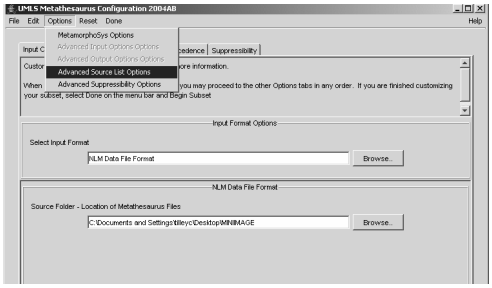
145

Edit menu



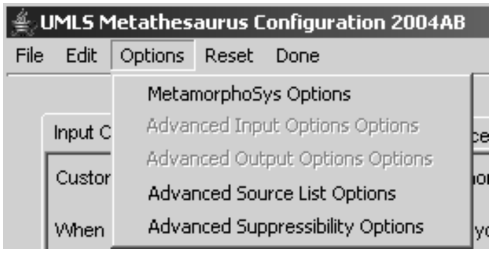
146

Options menu



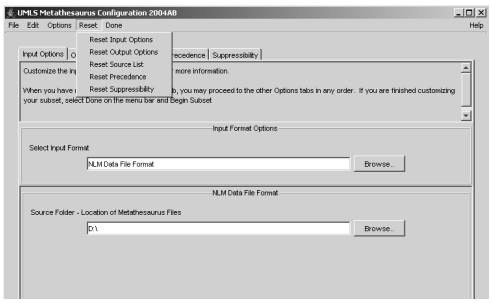
147

Options menu



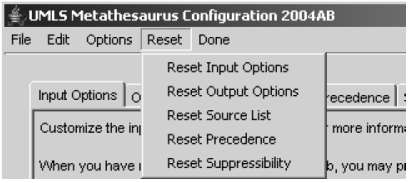
148

Reset menu



149

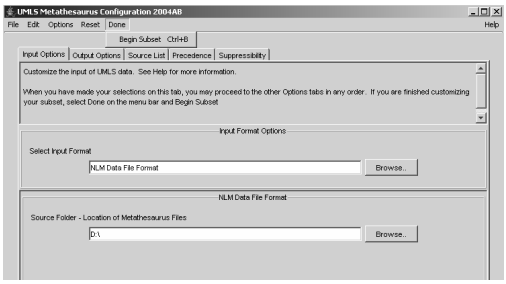
Reset menu



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

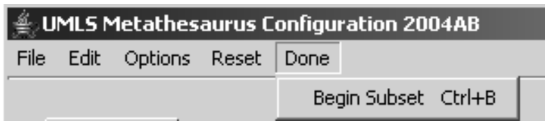
150

Done – Begin Subset



151

Done – Begin Subset



- Compete configuration options
- Done menu
- Begin Subset

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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

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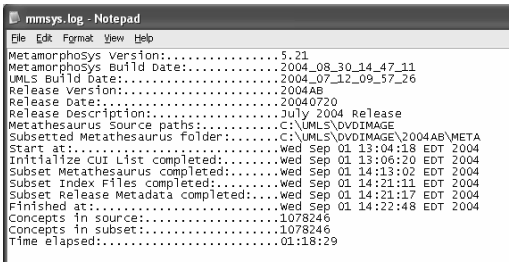
MetamorphoSys log



```
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
```

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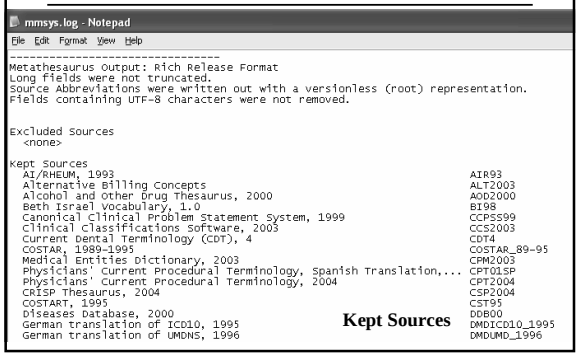
MetamorphoSys log



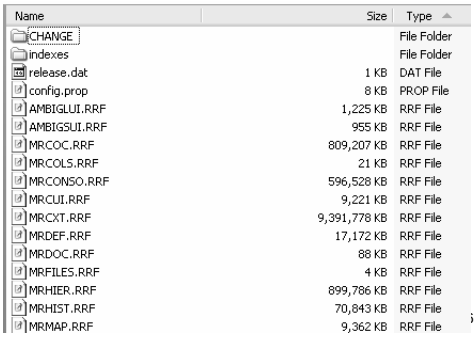
```
mmsys.log - Notepad
File Edit Format View Help
-------------------------------
Metathesaurus Output: Rich Release Format
Long fields were not truncated.
Source Abbreviations were written out with a versionless (root) representation.
Fields containing UTF-8 characters were not removed.


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
```

Kept Sources

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

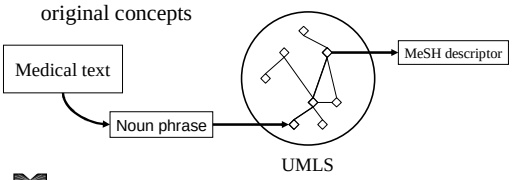
Part II
How to use the UMLS?

(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



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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

159

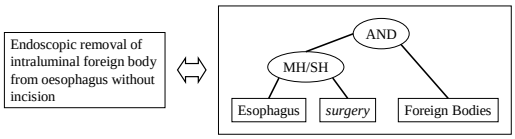
Restrict to MeSH Synonymy

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

160

Restrict to MeSH Assoc. expressions

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



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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

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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)

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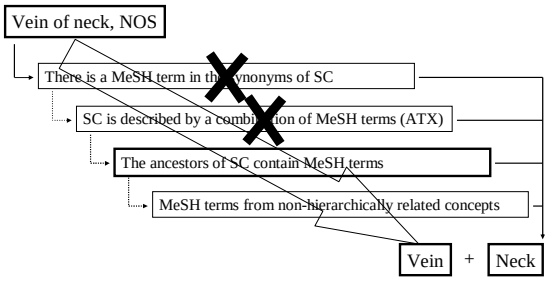
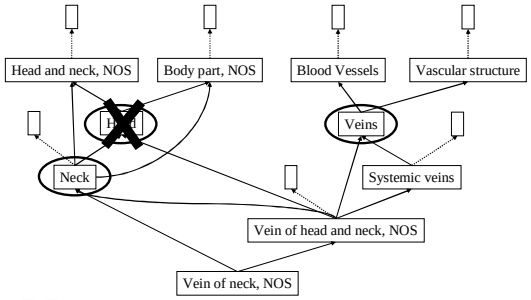
Restrict to MeSH Example



164

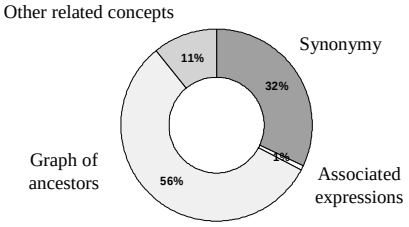
Restrict to MeSH Example



165

Restrict to MeSH Quantitative results

- 82.5% of UMLS concepts mapped to MeSH



166

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

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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

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Direct benefits

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

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UMLA as an enabling resource

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

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Limitations

Limitations

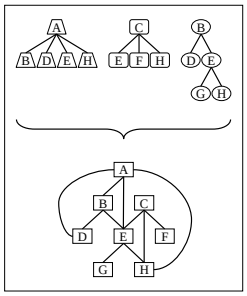
[Cimino, *JAMIA*, 1998]

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

174

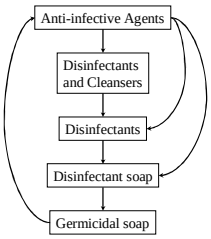
Structural inconsistency From trees to graph

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



175

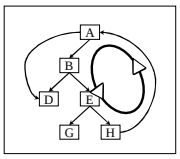
Structural inconsistency There are some cycles



176

Structural inconsistency Issues

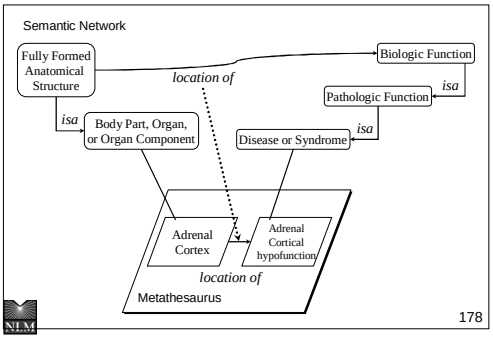
- Theoretical
  - Violate the antisymmetry property of partial ordering relations
- Practical
  - Loops in graph traversal
  - Impossible to perform transitive reduction



[Bodenreider, AMIA 2001]

177

Semantic inconsistency A two-level structure



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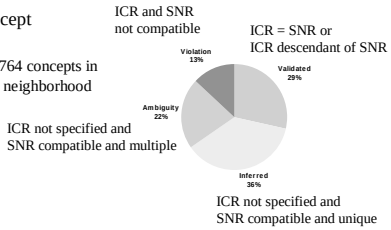
Semantic inconsistency A limited study

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



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.

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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]

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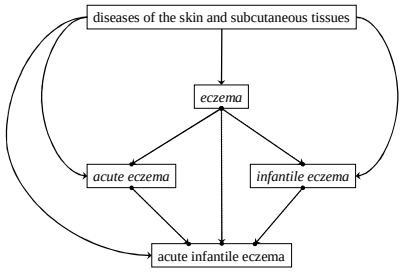
Missing relations Example



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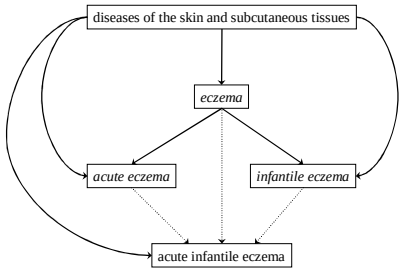
Missing relations Example



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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]

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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

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More limitations

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

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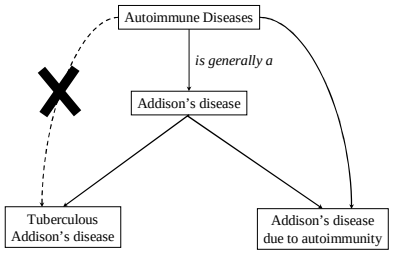
Meaning of *isa*



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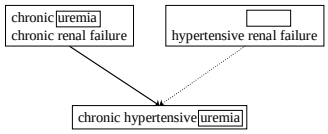
Relations Missing and difficult to detect



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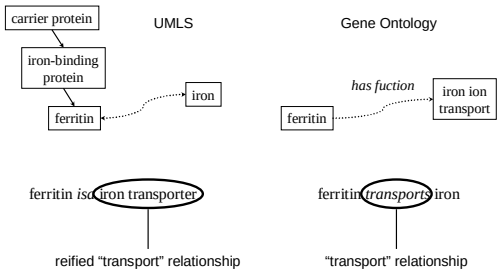
Relations Existing but difficult to find



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How to address these limitations?

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

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Summary

UMLS Overview

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

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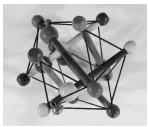

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  http:// www.nlm.nih.gov/research/umls/
- UMLS documentation
  - "Green Book"
  - online documentation
    http://www.nlm.nih.gov/research/umls/UMLSDOC.HTML
- UMLS Information web site
  http://umlsinfo.nlm.nih.gov/

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  - 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.

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  - 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

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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

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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	

NLM Appendix - Metathesaurus relational files (RRF) 200

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病	3	N	
10	MSHPJN	SY	D000224	副腎性黒皮症	3	N	

NLM Appendix - Metathesaurus relational files (RRF) 201

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	

NLM Appendix - Metathesaurus relational files (RRF) 202

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		

NLM Appendix - Metathesaurus relational files (RRF) 203

MRREL (sample rows) (2004AB)

	1	2	3	4	5	6	7
	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

	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	

NLM Appendix - Metathesaurus relational files (RRF) 204

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 CUI	2 LUI	3 SUI	4 METAUI	5 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 ATUI	8 SATUI	9 ATN	10 SAB	11 ATV	14 SUPPRESS	15 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 CUI	2 SOURCEUI	3 SAB	4 SVER	5 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 CHANGEKEY	7 CHANGEVAL	8 REASON	9 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|
[…]
```

Appendix - Semantic Network relational files (ORF) 223

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|
[…]
```

Appendix - Semantic Network relational files (ORF) 224

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|

Appendix - Semantic Network relational files (ORF) 225