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®
UML S 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.»
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
  - Bronzed disease
  - Addison melanoderma
  - Asthenia pigmentosa
  - Primary adrenal deficiency
  - Primary adrenal insufficiency
  - Primary adrenocortical insufficiency
  - Chronic adrenocortical insufficiency

◆ **Contexts: different hierarchies**
  - eponym
  - symptoms
  - clinical
  - variants
Organize terms

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

<table>
<thead>
<tr>
<th>Adrenal gland diseases</th>
<th>MeSH</th>
<th>D000307</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adrenal disorder</td>
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<tr>
<td>Disorder of adrenal gland</td>
<td>Read</td>
<td>C15z.</td>
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<td>Diseases of the adrenal glands</td>
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<td>DB-70000</td>
</tr>
<tr>
<td>C0001621</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Diseases of the endocrine system

Diseases of the Adrenal Glands

Addison’s Disease
Endocrine Diseases

Diseases

Adrenal Gland Diseases

Adrenal Gland Hypofunction

Addison’s Disease
Endocrine disorder

Adrenal disorder

Adrenal cortical disorder

Adrenal cortical hypofunction

Addison’s Disease
Primary adrenocortical insufficiency

Other disorders of adrenal gland

Disorders of other endocrine gland
Organize concepts

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

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Addison’s Disease

Endocrine Diseases

Adrenal Gland Diseases

Adrenal Cortex Diseases

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Addison’s Disease

organize concepts
Relate to other concepts

- Additional hierarchical relationships
  - link to other trees
  - make relationships explicit

- Non-hierarchical relationships

- Co-occurring concepts

- Mapping relationships
Endocrine Diseases

Adrenal Gland Diseases

Adrenal Cortex Diseases

Hypoadrenalism

Adrenal Gland Dysfunction

Adrenal cortical hypofunction

Secondary hypocortisolism

Addison’s Disease

Addison’s disease due to autoimmunity

relate to other concepts
Categorize concepts

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

Disease or Syndrome

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

Adrenal Cortex Diseases

Adrenal Cortex Dysfunction

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Addison’s Disease

Other disorders of adrenal gland
UML 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

- 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
Biomedical terminologies

◆ General vocabularies
  • anatomy (UWDA, Neuronames)
  • drugs (RxNorm, First DataBank, Micromedex)
  • medical devices (UMD, SPN)

◆ Several perspectives
  • clinical terms (SNOMED CT)
  • information sciences (MeSH, CRISP)
  • administrative terminologies (ICD-9-CM, CPT-4)
  • data exchange terminologies (HL7, LOINC)
Biomedical terminologies (cont’d)

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

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

The UMLS serves as a vehicle for the regulatory standards (HIPAA, CHI)
Addison’s Disease: Concept

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.

SNOMED
MeSH
AOD
Read Codes

C0001403

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
Metathesaurus Concepts

- **Concept** (~ 1.2M) CUI
  - Set of synonymous concept names
- **Term** (~ 4.2M) 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
  - S0011231 Adrenal Gland Disease
  - S0000441 Disease of adrenal gland
  - S0481705 Disease of adrenal gland, NOS
  - S0220090 Disease, adrenal gland
  - S0044801 Gland Disease, Adrenal

- Term L0041793
  - S0860744 Disorder of adrenal gland, unspecified
  - S0217833 Unspecified disorder of adrenal glands

- Term L0161347
  - S0225481 ADRENAL DISORDER
  - S0627685 DISORDER ADRENAL (NOS)

- Term L0181041
  - S0632950 Disorder of adrenal gland
  - S0354509 Adrenal Gland Disorders

- Term L0368399
  - S0586222 Adrenal disease
  - S0466921 ADRENAL DISEASE, NOS

- Term L1279026
  - S1520972 Nebennierenkrankheiten

- Term L0162317
  - S0226798 SURRENALE, MALADIES

[...]
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

Addison's disease
C0001403

Addison's disease, NOS
C0271735

**Metathesaurus Relationships**

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

**Categorization:** Relationships between concepts and semantic types from the Semantic Network
Symbolic relations

◆ Relation
  - Pair of “atom” identifiers
  - Type
  - Attribute (if any)
  - List of sources (for type and attribute)

◆ Semantics of the relationship:
  defined by its type [and attribute]

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

Type

- Hierarchical
  - Parent / Child
  - Broader / Narrower than

- Derived from hierarchies
  - Siblings (children of parents)

- Associative
  - Other

- Various flavors of near-synonymy
  - Similar
  - Source asserted synonymy
  - Possible synonymy
Symbolic relationships

Attribute

◆ Hierarchical
  ● isa (is-a-kind-of)
  ● part-of

◆ Associative
  ● location-of
  ● caused-by
  ● treats
  ● …

◆ Cross-references (mapping)
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)

- Biologic Function
  - Physiologic Function
    - Organism Function
    - Organ or Tissue Function
    - Cell Function
    - Molecular Function
      - Mental Process
      - Genetic Function
  - Pathologic Function
    - Cell or Molecular Dysfunction
    - Disease or Syndrome
    - Experimental Model of Disease
      - Mental or Behavioral Dysfunction
      - Neoplastic Process
Associative (non-isa) relationships

- Organism
  - Anatomical Structure
    - Embryonic Structure
    - Anatomical Abnormality
      - Congenital Abnormality
      - Acquired Abnormality
    - Body System
      - Body Part, Organ or Organ Component
      - Tissue
      - Cell
      - Cell Component
      - Gene or Genome
    - Fully Formed Anatomical Structure
      - contains, produces
  - Organism Attribute
  - Finding
    - Laboratory or Test Result
    - Sign or Symptom
    - Pathologic Function
    - Physiologic Function
  - Biologic Function
  - Injury or Poisoning
    - disrupts
  - Body Location or Region
    - location of
  - Body Space or Junction
    - adjacent to
  - Biologic Function
  - Body Substance
    - conceptual part of
    - contains, produces
  - Injury or Poisoning
    - disrupts
    - co-occurs with
Why a semantic network?

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

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

Semantic Network

- Fully Formed Anatomical Structure
- Biologic Function
- Pathologic Function
- Disease or Syndrome
- Body Part, Organ, or Organ Component
- Adrenal Cortex
- Adrenal Cortical hypofunction
- Metathesaurus

location of
isa
SPECIALIST Lexicon and lexical tools
SPECIALIST Lexicon

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

◆ Inflection
  • noun  nucleus, nuclei
  • verb  cauterize, cauterizes, cauterized, cauterizing
  • adjective  red, redder, reddest

◆ Derivation
  • verb ↔ noun  cauterize -- cauterization
  • adjective ↔ noun  red -- redness
Orthography

◆ Spelling variants

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

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

◆ Position for adjectives

Valve of coronary sinus
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

- Remove genitive: Hodgkin’s diseases, NOS
- Remove stop words: Hodgkin diseases, NOS
- Lowercase: Hodgkin diseases,
- Strip punctuation: hodgkin diseases,
- Uninflect: hodgkin diseases
- Sort words: hodgkin disease
- Final result: disease hodgkin
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:


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

- Verify:
  - receive e-mail from NLM; go to Web site within 72 hours and enter first and last name
  - NLM official will countersign (turn-around time of a few days)
  - Receive 2nd e-mail from NLM with new license number
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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.


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


CATEGORIES OF RESTRICTIONS APPLY

Contact: Alternative Link LLC; 6121 Indian School Road NE, Suite 131; Albuquerque, NM 87110; phone: 877-521-5465; http://www.alternative-link.com; e-mail: mail@alternative-link.com

*NOTE: Now a CATEGORY 0

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


CATEGORY 2 RESTRICTIONS APPLY

The Metathesaurus includes translations of WHO97 in French (WHOFRE_1997), German (WHODER_1997), Portuguese (WHOPOR_1997), and Spanish (WHOSPES_1997).

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

Last updated: 20 July 2004
First published: 26 March 2004
Permanence level: Permanence Not Guaranteed
License Restriction Levels 0-4

- **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

There may be additional restrictions, or separate license fees, associated with usage of specific vocabularies. Read the UMLS License, including the Appendix!
Part II
How to use the UMLS?

(2) Remote access
Remote Access

- UMLS Knowledge Source Server:

- 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
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: Adrenal gland hypofunction
CUI: C0001623

Semantic Type: Disease or Syndrome

Definition:
Adrenocortical hypofunction includes all conditions where adrenal hormone secretion falls below the requirements of the body. It may be divided into two general categories: (1) the inability of the adrenal to elaborate sufficient quantities of aldosterone associated with a secondary failure due to a primary adrenocorticotropic (ACTH) insufficiency, and (2) adrenal failure associated with a primary ACTH insufficiency.

Synonyms:
Adrenal gland hypofunction, adrenal insufficiency, adrenal failure, Addison disease (MeSH)
Adrenal Gland Insufficiency

- **Concept Name/CUI**: adrenal gland hypofunction (C0001623)
- **Semantic Type(s)**: Disease or Syndrome
- **Definition**: Adrenocortical hypofunction includes all conditions in which adrenal steroid hormone secretion falls below the requirements of the body. Adrenal insufficiency may be divided into two general categories: (1) those associated with primary insufficiency of the adrenal to elaborate sufficient quantities of hormone and (2) those associated with a secondary failure due to a primary failure in the elaboration of adrenocorticotropic. (Harrison’s Principles of Internal Medicine, 15th ed, p19710) (MeSH)
- **Synonyms**, including foreign languages: Adrenal gland hypofunction, Adrenal failure, Adrenal Insufficiency
- **Relations (broader, narrower, etc.)**: Hypofunctions, Adrenal Gland
- **Co-occurrence data**

[Image of UMLS Knowledge Source Server (UMLSKS) with search for adrenal gland insufficiency in UMLS Release 2004AB]
Concept Report Display All (continued)

Hierarchies

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

Narrower Concepts:

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

Concept Report Display All (continued)
Co-occurrence data

Associated Expressions: None found.

Co-occurring MeSH Terms:

- [64] Corticotropin
- [57] Hydrocortisone
- [38] Glucocorticoids
- [31] Esophageal achalasia
- [30] Dog Diseases
- [28] Adrenal Glands

93
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
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

Metathesaurus Relational Records Request:

Select UMLS Release:

1) Enter a term or concept unique identifier (CUI): addison's disease

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

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

Pressing the Perform Concept Search button will update the display to show the row records from the selected table based on the concept information found.
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<th>SUI</th>
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<th>AUI</th>
<th>SAUI</th>
<th>SCUI</th>
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</table>
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

About the UMLSKS
- Home
- Overview
- Frequently Asked Questions
- Edit Views/Profile

Downloads
- UMLS Knowledge Sources
- Developer’s API
- Documentation
- User’s Guide
- Developer’s Guide
- Developer’s API Javadocs

SPECIALIST Lexicon

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

Lexical entries may be single or multi-word terms.

View Lexical Records for: I
Specialist Lexical Record

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

View "Addison's disease" in relational format.
UMLS Resources

- **NLP & Lexical Resources**
  - MetaMap Transfer (MMTx)
  - Word Sense Disambiguation (WSD) Test Collection
- **Semantic Network**
  - Semantic Navigator
  - Semantic Groups
- **Metathesaurus**
  - String Properties
Knowledge Source Server
Application Programming Interface
# UMLSKS API basics

- Remote server at NLM
- Local application connected through

<table>
<thead>
<tr>
<th><strong>Java RMI</strong></th>
<th><strong>TCP/IP socket</strong></th>
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</thead>
<tbody>
<tr>
<td>- Java-based applications</td>
<td>- XML-based queries</td>
</tr>
<tr>
<td>- Developer’s Guide: Chapter 3</td>
<td>- Developer’s Guide: Chapter 5</td>
</tr>
<tr>
<td>- Set of Java classes (part of the UMLSKS API download)</td>
<td>- XML schema</td>
</tr>
<tr>
<td>- Detailed <em>Javadoc</em> documentation online and with API download</td>
<td>- Socket server</td>
</tr>
<tr>
<td></td>
<td>- Host: umlsks.nlm.nih.gov</td>
</tr>
<tr>
<td></td>
<td>- Port: 8042</td>
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</tbody>
</table>
Developer’s Guide

UMLSKS Knowledge Sources

About the UMLSKS
- Home
- Overview
- Frequently Asked Questions
- Edit Views/Profile

Documentation
- 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
- User’s Guide
- UMLSKS Documentation Set

Resources
- NLP & Lexical Resources
- Semantic Network Resources
- On-the-scene Resources

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

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

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

How to Use This Guide
This manual contains the following chapters:
- Chapter 1 - Introduction describes the basic features and architecture of the UMLSKS.
- Chapter 2 - Installing the UMLSKS provides administrators instructions on installing and tailoring a UMLSKS installation.
- Chapter 3 - Building UMLSKS Software Applications describes the functions available to developers wanting to interface to the UMLSKS through another Java program.
- Chapter 4 - Using the XML Query Facility describes how to use the querying facility of the UMLSKS wherein users build XML queries to be executed.
- Chapter 5 - Using the UMLSKS Socket Server describes how to use the socket server to pass XML formatted commands or command-line type queries (e.g. ks -meta -c aids) that are to be executed by the server with data where appropriate.
Documentation  Java API

UMLS Knowledge Source Server (UMLSKS)

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

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
### Packages

<table>
<thead>
<tr>
<th>Package</th>
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</thead>
<tbody>
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<tr>
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<td>.gov.nlm.kss.query.lex</td>
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Sample XML query  (1) Current version

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

```xml
<?xml version="1.0"?>
<CurrentUMLSYear version="1.0">
  2004AB
</CurrentUMLSYear>
```
Sample XML query (2) Concepts by string

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

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<ConceptIdCollection version="1.0"> 
  <release>2004AB</release> 
  <conceptId> 
    <cui>C0003611</cui> 
    <cn>Appendectomy</cn> 
  </conceptId> 
</ConceptIdCollection>
```
Sample XML query (3) Concepts properties

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

<?xml version="1.0"?>
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  <cn>Prostate</cn>
  <semanticType>
    <tui>T023</tui>
    <sty>Body Part, Organ,
       or Organ Component</sty>
  </semanticType>
</SemanticTypeCollection>
```
Sample XML query  (4) Relationships

```xml
<?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
<?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>
  </semnetId>
  <semnetId>
    <name>Activity</name>
    <ui>T052</ui>
  </semnetId>
  ...
</SemNetIdCollection>
```
Performing XML queries from UMLSKS

The XML query facility allows users to specify a query using the eXtensible Markup Language (XML) dialect created for the UMLSKS. Users of this searching feature must be familiar with the internal database structures and how table data are related to each other.

The data tables used to populate the backend Oracle database can be returned to the user that matches a user’s input term or concept unique identifier.
Performing XML queries from UMLSKS

```xml
<?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

Advanced Options

- Copy NLM data format files (.nlm) to hard drive
- Copy MetamorphoSys to hard drive
- Run MD5 validation

[Done] [Cancel]
Notice:

The Metathesaurus contains source vocabularies produced by many different copyright holders. The majority of the content of the Metathesaurus is available for use under the basic (and quite open) terms described in the Metathesaurus license [http://www.nlm.nih.gov/research/umls/license.html](http://www.nlm.nih.gov/research/umls/license.html).

However, some vocabulary producers place ADDITIONAL RESTRICTIONS ON THE USE OF THEIR CONTENT AS DISTRIBUTED WITHIN THE METATHESAURUS.

The various levels of additional restrictions are described in Section 1.2 of the license. The level that applies to individual vocabularies is recorded in the Appendix to the license. If a UMLS user already has a separate license for use of one of the source vocabularies, the user’s existing license also applies to that source as distributed within the Metathesaurus. In some cases, UMLS users may have to request permission or negotiate a separate license with a vocabulary producer in order to use that vocabulary in a production system. There may be a charge associated with these separate permissions or license agreements.

Please select “Accept” or “Do Not Accept” below after reviewing the license agreement at the URL above.
Installation progress monitor

Install UMLS
The following operations will be performed:

- Installing Specialist Lexicon & Tools
- Installing Semantic Network
- Configure Metathesaurus Subset
- Installing Metathesaurus
  - Initializing CUI List
  - Subsetting Content
  - Subsetting Indexes
  - Final Processes
- Done

Cancel

Operation is 17% Complete

Cancel
Select a default subset

<table>
<thead>
<tr>
<th>Name</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Level 0</td>
<td>No separate additional license agreements</td>
</tr>
<tr>
<td>Level 0 + SNOMEDCT</td>
<td>Non-U.S. users must have separate license agreements</td>
</tr>
<tr>
<td>RxNorm Subset</td>
<td>No separate additional license agreements</td>
</tr>
</tbody>
</table>

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

Customize the input of UMLS data. 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.

Input Format Options

Select Input Format

NLM Data File Format

Browse..

NLM Data File Format

Source Folder - Location of Metathesaurus Files

D:\

Browse..
Output Options Tab

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

Include or exclude source vocabularies for your Metathesaurus subset. See Help for more information.

<table>
<thead>
<tr>
<th>Full Source Name</th>
<th>Source Abbreviation</th>
<th>Source Family</th>
<th>Language</th>
<th>Level</th>
</tr>
</thead>
<tbody>
<tr>
<td>AIRHELM, 1993</td>
<td>AIR93</td>
<td>AIR</td>
<td>ENG</td>
<td>0</td>
</tr>
<tr>
<td>Alternative Billing Concepts</td>
<td>ALT2003</td>
<td>ALT</td>
<td>ENG</td>
<td>5</td>
</tr>
<tr>
<td>Alcohol and Other Drug Thesaurus, 2000</td>
<td>AOD2000</td>
<td>AOD</td>
<td>ENG</td>
<td>0</td>
</tr>
<tr>
<td>Beth Israel Vocabulary, 1.0</td>
<td>BI98</td>
<td>BI</td>
<td>ENG</td>
<td>2</td>
</tr>
<tr>
<td>Canonical Clinical Problem Statement System, 1999</td>
<td>CCPSS99</td>
<td>CCPSS</td>
<td>ENG</td>
<td>5</td>
</tr>
<tr>
<td>Clinical Classifications Software, 2003</td>
<td>CCS2003</td>
<td>CCS</td>
<td>ENG</td>
<td>0</td>
</tr>
<tr>
<td>Current Dental Terminology (CDT), 4</td>
<td>CDT4</td>
<td>CDT</td>
<td>ENG</td>
<td>5</td>
</tr>
</tbody>
</table>

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

Cut and paste rows to alter the preferred name
# Suppressibility Tab

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.

<table>
<thead>
<tr>
<th>Source Term Type</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
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<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
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<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICCPC33</td>
</tr>
</tbody>
</table>

Highlighted source term types will be marked as suppressible.
File menu

Image of a computer interface for UMLS Metathesaurus Configuration 2004AB.
File menu

- Enable/Disable Filter, Ctrl+O
- Import Filter, Ctrl+I
- New Configuration, Ctrl+N
- Open Configuration, Ctrl+O
- Save Configuration, Ctrl+S
- Exit, Ctrl+Q
Edit menu

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.
Edit menu

- Undo Suppressibility action: Ctrl+Z
- Redo: Ctrl+Y
- Increase Font: Ctrl+]
- Decrease Font: Ctrl+[
Options menu

[Image of a software interface showing the Options menu with options for MetamorphoSys, Advanced Options, and Advanced Source List Options.]
Options menu

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

![Image of UMLS Metathesaurus Configuration 2004AB with reset options]

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

For more information, 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.
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

Customize the input of UMLS data. 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.

Select Input Format

NLM Data File Format

Source Folder - Location of Metathesaurus Files

D:\

Browse..
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

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\DV DIMAGE
Subsetted Metathesaurus folder:.........C:\UMLS\DV DIMAGE\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

-----------------------------
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
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

<table>
<thead>
<tr>
<th>Name</th>
<th>Size</th>
<th>Type</th>
</tr>
</thead>
<tbody>
<tr>
<td>CHANGE</td>
<td></td>
<td>File Folder</td>
</tr>
<tr>
<td>indexes</td>
<td></td>
<td>File Folder</td>
</tr>
<tr>
<td>release.dat</td>
<td>1 KB</td>
<td>DAT File</td>
</tr>
<tr>
<td>config.prop</td>
<td>8 KB</td>
<td>PROP File</td>
</tr>
<tr>
<td>AMBIGLUI.RRF</td>
<td>1,225 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>AMBIGSUI.RRF</td>
<td>955 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRCOC.RRF</td>
<td>809,207 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRCOLS.RRF</td>
<td>21 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRCONSO.RRF</td>
<td>596,528 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRCUI.RRF</td>
<td>9,221 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRCXT.RRF</td>
<td>9,391,778 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRDEF.RRF</td>
<td>17,172 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRDOC.RRF</td>
<td>88 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRF FILES.RRF</td>
<td>4 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRHIER.RRF</td>
<td>899,786 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRHIST.RRF</td>
<td>70,843 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MRRMAP.RRF</td>
<td>9,362 KB</td>
<td>RRF File</td>
</tr>
</tbody>
</table>
Part II
How to use the UMLS?

(4) A UMLS-based algorithm
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

- 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

[Bodenreider & al., AMIA, 1998]
Restrict to MeSH  Synonymy

- Term mapped to Source concept
- For this concept, is there a synonym term that comes from MeSH? (MRSO)
If not,

Is there an associated expression (ATX) that describes this concept using a combination of MeSH descriptors? (MRATX)
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)
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

- Head and neck, NOS
- Body part, NOS
- Blood Vessels
- Vascular structure

- Head
- Neck
- Veins
- Systemic veins
- Vein of head and neck, NOS
- Vein of neck, NOS
Restrict to MeSH  Quantitative results

82.5% of UMLS concepts mapped to MeSH

- Other related concepts
- Synonymy
- Graph of ancestors
- Associated expressions
**Restrict to MeSH Qualitative results**

- **Qualitative evaluation**
  - 1,036 concepts extracted from 200 MEDLINE citations
  - Manual review of every mapping or failure
- **61% Relevant**
  - Subtotal Gastrectomy ➔ Gastrectomy
  - Encephalopathy, NOS ➔ Brain Diseases
- **28% More or less relevant**
  - Vitamin A measurement ➔ Laboratory Procedure
  - Swelling, NOS ➔ Symptoms
- **11% Non relevant**
Part II
How to use the UMLS?

(5) Benefits and Limitations
Benefits
UMLS compared to individual vocabularies

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

- Concept categorization
- Information retrieval
  - Synonyms
  - Cross-language features
- Information extraction
  - MetaMap
  - Normalization
- Information visualization
  - Knowledge Source Server
  - Semantic Navigator
UMLA as an enabling resource

◆ Examples
  ● Mapping across vocabularies
  ● Semantics of statistical associations
  ● Redundancy in hierarchical relations
Limitations
Limitations

- **Structural inconsistency**
  - Cycles in the graph of hierarchical relations

- **Semantic inconsistency**
  - Between Metathesaurus and Semantic Network

- **Missing relations**
  - Synonymy
  - Hierarchical relations (missing or underspecified)

[Cimino, *JAMIA*, 1998]
Structural inconsistency  From trees to graph

- Multiple tree structures combined into a graph structure
- Directed acyclic graph (DAG)
Structural inconsistency  There are some cycles

- Anti-infective Agents
  - Disinfectants and Cleansers
    - Disinfectants
      - Disinfectant soap
        - Germicidal soap
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 Network

- Fully Formed Anatomical Structure
- Biologic Function
- Pathologic Function
- Disease or Syndrome
- Body Part, Organ, or Organ Component
- Adrenal Cortex
- Adrenal Cortical hypofunction
- Metathesaurus

location of

isa
Semantic inconsistency  A limited study

◆ 6894 interconcept relationships
  • among the 3764 concepts in the semantic neighborhood of “Heart”

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

diseases of the skin and subcutaneous tissues

eczema

acute eczema

infantile eczema

acute infantile eczema
Missing relations  Example

- diseases of the skin and subcutaneous tissues
- eczema
- acute eczema
- infantile eczema
- acute infantile eczema
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*

- Autoimmune Diseases
  - Addison’s disease
    - Tuberculous Addison’s disease
    - Addison’s disease due to autoimmunity

*is generally a*
Relations Missing and difficult to detect

- Chronic uremia
- Chronic renal failure
- Hypertensive renal failure
- Chronic hypertensive uremia
Relations  Existing but difficult to find

**Carrier Protein**

- ferritin
- iron-binding protein
- iron

**UMLS**

- ferritin
- iron

**Gene Ontology**

- ferritin
- iron ion transport

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

◆ Short presentation
References

◆ Technical papers


◆ Comprehensive bibliography 1986-96

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)

<table>
<thead>
<tr>
<th></th>
<th>CUI</th>
<th>LAT</th>
<th>7^</th>
<th>LUI</th>
<th>SUI</th>
<th>ISPREF</th>
<th>AUI</th>
<th>SAUI</th>
<th>SCUI</th>
<th>SDUI</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>C0001403</td>
<td>ENG</td>
<td>P</td>
<td>L0001403</td>
<td>PF</td>
<td>S0354372</td>
<td>Y</td>
<td>A4367951</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td>C0001403</td>
<td>ENG</td>
<td>P</td>
<td>L0001403</td>
<td>PF</td>
<td>S0354372</td>
<td>N</td>
<td>A2922421</td>
<td>485624014</td>
<td>363732003</td>
</tr>
<tr>
<td>3</td>
<td>C0001403</td>
<td>ENG</td>
<td>P</td>
<td>L0001403</td>
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Appendix 2

UMLS files in
Original Release Format
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Appendix - Metathesaurus relational files (ORF)
MRDEF  Definitions

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. | [...]

Appendix - Metathesaurus relational files (ORF)
## MRSTY Semantic Types

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**Closed fracture of malar and maxillary bones, NOS**

**Unilateral congenital dislocation of hip**

**Suture of bladder**

**Corneal abrasion**

**CORRECTIVE LENS PROBLEM**

**Chronic cough**

**Cyst and pseudocyst of pancreas**

**Cystitis**
MRCXT  Contexts (2003AA)

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* 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)  
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Appendix - Metathesaurus relational files (ORF)
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Appendix - Metathesaurus relational files (ORF)
## MRCOC Co-occurrences

### Appendix - Metathesaurus relational files (ORF)

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## MRCUI Concept history

(2003AA)

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Appendix - Metathesaurus relational files (ORF)
Appendix - Metathesaurus relational files (ORF)
## Basic information

### Organism

| T001 | Generally, a living individual, including all plants and animals. | Homozygote; Radiation Chimera; Sporocyst |

### Plant

| T002 | 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 |

### Alga

| T003 | A chiefly aquatic plant that contains chlorophyll, but does not form embryos during development and lacks vascular tissue. | Chlorella; Laminaria; Seaweed |

### Fungus

| T004 | A eukaryotic organism characterized by the absence of chlorophyll and the presence of a rigid cell wall. Included are both slime molds and true fungi such as yeasts, molds, mildews, and mushrooms. | Aspergillus clavatus; Blastomyces; Helminthosporium; Neurospora |

### Appendix - Semantic Network relational files (ORF)
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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 |

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from Biologic Function \text{affects} Organism

Appendix - Semantic Network relational files (ORF)