The Unified Medical Language System

*What is it and how to use it?*

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Jan Willis
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Acknowledgements

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Outline

◆ What is the UMLS?
◆ How to use the UMLS?
  ● Obtaining a license
  ● Remote access
    ■ Knowledge Source Server (UMLSKS)
    ■ UMLSKS Application programming interface (API)
  ● Local installation and customization
    (MetamorphoSys)
◆ Questions
Part I
What is the UMLS?
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
Motivation

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

«[...] 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.»

[Lindberg & al., Methods, 1993]
[Humphreys & al., JAMIA, 1998]
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>Term</th>
<th>Source</th>
<th>Identifier</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adrenal gland diseases</td>
<td>MeSH</td>
<td>D000307</td>
</tr>
<tr>
<td>Adrenal disorder</td>
<td>AOD</td>
<td>0000005418</td>
</tr>
<tr>
<td>Disorder of adrenal gland</td>
<td>Read</td>
<td>C15z.</td>
</tr>
<tr>
<td>Diseases of the adrenal glands</td>
<td>SNOMED</td>
<td>DB-70000</td>
</tr>
</tbody>
</table>

C0001621

Adrenal Gland Diseases
Diseases of the endocrine system

Diseases of the Adrenal Glands

Addison’s Disease
Endocrine Diseases

Adrenal Gland Diseases

Adrenal Gland Hypofunction

Addison’s Disease

Diseases

MeSH
Endocrine disorder

Adrenal disorder

Adrenal cortical disorder

Adrenal cortical hypofunction

Addison’s Disease

Addison’s Disease
Endocrine disorder

Disorder of adrenal gland

Hypoadrenalism

Adrenal Hypofunction

Corticoadrenal insufficiency

Addison’s Disease
Primary adrenocortical insufficiency

Other disorders of adrenal gland

Disorders of other endocrine gland

ICD-10
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

organize concepts

SNOMED
MeSH
AOD
Read Codes

UMLS
Relate to other concepts

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

Adrenal Gland Diseases

Adrenal Cortex Diseases

Hypoadrenalism

Adrenal Gland Hypofunction

Adrenal cortical hypofunction

Secondary hypocortisolism

Addison’s Disease

Addison’s disease due to autoimmunity

Diseases

Endocrine Diseases

Adrenal Dysfunction

Disorders of other endocrine gland

Other disorders of adrenal gland

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

![Diagram]

- 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
UMLS Summary

- Synonymous terms clustered into concepts
- Unique identifier
- Finer granularity
- Broader scope
- Additional hierarchical relationships
- Semantic categorization
Part I
What is the UMLS?

(3) UMLS Knowledge Sources
UMLS 3 components

- Metathesaurus
  - Concepts
  - Inter-concept relationships

- Semantic Network
  - Semantic types
  - Semantic network relationships

- Lexical resources
  - SPECIALIST Lexicon
  - Lexical tools
UMLS Metathesaurus
Metathesaurus  Basic organization

◆ Concepts
  ● Synonymous terms are clustered into a concept
  ● Properties are attached to concepts, e.g.,
    ■ Unique identifier
    ■ Definition

◆ Relations
  ● Concepts are related to other concepts
  ● Properties are attached to relations, e.g.,
    ■ Type of relationship
    ■ Source
Source Vocabularies

- 134 source vocabularies
  - 126 contributing concept names
- 73 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.
**Metathesaurus Concepts**

- **Concept** (> 1M) CUI
  - Set of synonymous concept names
- **Term** (> 3.8 M) LUI
  - Set of normalized names
- **String** (> 4.3M) SUI
  - Distinct concept name
- **Atom** (> 5.1M) AUI
  - Concept name in a given source

Examples:
- **A0000001** headache (source 1)
- **A0000002** headache (source 2)
- **A0000003** Headache (source 1)
- **A0000004** Headache (source 2)
- **A0000005** Cephalgia (source 1)
- **S0000001**
- **S0000002**
- **S0000003**
- **L0000001**
- **L0000002**
- **C0000001**
Cluster of synonymous terms

<table>
<thead>
<tr>
<th>Concept</th>
<th>C0001621</th>
</tr>
</thead>
<tbody>
<tr>
<td>Term</td>
<td>L0001621</td>
</tr>
<tr>
<td>S0011232</td>
<td>Adrenal Gland Diseases</td>
</tr>
<tr>
<td>S0011231</td>
<td>Adrenal Gland Disease</td>
</tr>
<tr>
<td>S0000441</td>
<td>Disease of adrenal gland</td>
</tr>
<tr>
<td>S0481705</td>
<td>Disease of adrenal gland, NOS</td>
</tr>
<tr>
<td>S0220090</td>
<td>Disease, adrenal gland</td>
</tr>
<tr>
<td>S0044801</td>
<td>Gland Disease, Adrenal</td>
</tr>
</tbody>
</table>

| 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, NOS
Metathesaurus Relationships

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

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

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

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

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

<table>
<thead>
<tr>
<th>Type</th>
<th>Symbol</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hierarchical</td>
<td></td>
</tr>
<tr>
<td>Parent / Child</td>
<td>PAR/CHD</td>
</tr>
<tr>
<td>Broader / Narrower than</td>
<td>RB/RN</td>
</tr>
<tr>
<td>Derived from hierarchies</td>
<td></td>
</tr>
<tr>
<td>Siblings (children of parents)</td>
<td>SIB</td>
</tr>
<tr>
<td>Associative</td>
<td></td>
</tr>
<tr>
<td>Other</td>
<td>RO</td>
</tr>
<tr>
<td>Various flavors of near-synonymy</td>
<td></td>
</tr>
<tr>
<td>Similar</td>
<td>RL</td>
</tr>
<tr>
<td>Source asserted synonymy</td>
<td>SY</td>
</tr>
<tr>
<td>Possible synonymy</td>
<td>RQ</td>
</tr>
</tbody>
</table>
Symbolic relationships

- 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

Phi 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
  - Mental Process
- Organ or Tissue Function
- Cell Function
  - Genetic Function
- Molecular Function

Pathologic Function
- Cell or Molecular Dysfunction
- Disease or Syndrome
  - Mental or Behavioral Dysfunction
- Experimental Model of Disease
  - Neoplastic Process
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

location of

Biologic Function

isa

Pathologic Function

isa

Body Part, Organ, or Organ Component

Disease or Syndrome

location of

Adrenal Cortex

Adrenal Cortical hypofunction

location of

Metathesaurus
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
- ae/e
- ise/ize
- genitive mark

oesophagus - esophagus
anaemia - anemia
cauterise - cauterize
Addison's disease
Addison disease
Addisons disease
Syntax

◆ Complementation

  ● verbs
    ■ intransitive  I'll treat.
    ■ transitive    He treated the patient.
    ■ ditransitive He treated the patient with a drug.

  ● nouns
    ■ prepositional phrase

  Valve of coronary sinus

◆ Position for adjectives
Lexical tools

- To manage lexical variation in biomedical terminologies

- Major tools
  - Normalization
  - Indexes
  - Lexical Variant Generation program (lvg)

- Based on the SPECIALIST Lexicon

- Used by noun phrase extractors, search engines
Normalization

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

disease hodgkin
Normalization: Example

Hodgkin Disease
HODGKINS DISEASE
Hodgkin's Disease
Disease, Hodgkin's
Hodgkin's, disease
HODGKIN'S DISEASE
Hodgkin's disease
Hodkins Disease
Hodkin's disease NOS
Hodgkin's disease, NOS
Disease, Hodgkins
Diseases, Hodgkins
Hodgkins Diseases
Hodgkins disease
hodkin'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)
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


CATEGORY 3 RESTRICTIONS APPLY

Contact: Alternative Link LLC; 6212 Indian School Road NE, Suite 131; Albuquerque, NM 87110; phone: 877-621-5465; http://www.alternativelink.com; e-mail: mail@alternativelink.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 (WHOGER_1997), Portuguese (WHOPOR_1997), and Spanish (WHOESP_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

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  - 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 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 hormones associated with a secondary failure due to a primary adrenocorticotropic failure (MeSH) (Harrison’s Principles of Internal Medicine).

**Synonyms:**
Adrenal gland hypofunction
## Adrenal Gland Insufficiency

<table>
<thead>
<tr>
<th>Concept Name/CUI</th>
<th>Semantic Type(s)</th>
<th>Definition(s)</th>
<th>Synonyms, including foreign languages</th>
<th>Relations (broader, narrower, etc.)</th>
<th>Co-occurrence data</th>
</tr>
</thead>
<tbody>
<tr>
<td>Adrenal gland hypofunction</td>
<td>Disease or Syndrome</td>
<td>Adrenocortical hypofunction includes all conditions in which adrenal steroid hormone secretion falls below the requirements of the body. Adrenal insufficiency may be divided into two general categories: (1) those associated with primary inability of the adrenal to elaborate sufficient quantities of hormone and (2) those associated with a secondary failure due to a primary failure in the elaboration of adrenocorticotropic. (Harrison’s Principles of Internal Medicine, 13th ed, p1970) (Mesh)</td>
<td>Adrenal gland hypofunction, Adrenal failure, Adrenal hypofunction, Adrenal hypofunction (disorder), Adrenal insufficiency, Hypofunctions, Adrenal Gland</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Concept Report Display All (continued)

**Synonyms**

- Japanese: 副腎機能低下
- Portuguese: Hipofunção das Glândulas Supra-Renais
- Russian: НАДПОЧЕЧНИКОВ ГИПОФУНКЦИЯ
- Spanish: Hipofunción suprarrenal

**Sources**

- Beth Israel Problem List
- Clinical Problem Statements
- COSTAR
- CRISP Thesaurus
- COSTART

Suppressible Synonyms:
- adrenal insufficiency <2>
Concept Report Display All (continued)

Hierarchies

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

MedDRA
- Metabolism and Nutrition [C01]
- Endocrine System [C19]
- Adrenal Gland Diseases [C19.053]
- Adrenal Gland Hypofunction [C19.053.264]

NCI Thesaurus
- Diseases, Disorders and Findings []
- Diseases and Disorders []
- Disorder by Site []
- Endocrine Disorder []
- Adrenal Gland Disorder []
- Non-Neoplastic Adrenal Gland Disorder []
- Adrenal Gland Insufficiency []
Concept Report Display All (continued)

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]
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
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
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 Search for: *Addison’s disease* in UMLS Release 2004AB

- Term Name: Addison’s disease
- CUI: C0001403
- Term UI: L0001403

- **Source**: SNOMED Clinical Terms
- **TTY**: PT
- **ID**: 363732003

Users are responsible for compliance with UMLS copyright restrictions.
Metathesaurus Advanced Search Feature
Relational Record Request

- **UMLS Release**
- **Search Term**
- **UMLS Relational Table**

---

**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
  - Developer’s API Javadocs
  - UMLS Documentation Set
- Resources
  - NLP & Lexical Resources
  - Semantic Network Resources
  - Metathesaurus Resources

**Metathesaurus Relational Records Request:**

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.
**Relational Records**  MRCONSO.RRF

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<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
- Select semantic type
- 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

<table>
<thead>
<tr>
<th>Type 1:</th>
<th>Relation(s):</th>
<th>Type 2:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acquired Abnormality</td>
<td>adjacent_to</td>
<td>Acquired Abnormality</td>
</tr>
<tr>
<td>Activity</td>
<td>affects</td>
<td>Activity</td>
</tr>
<tr>
<td>Age Group</td>
<td>analyzes</td>
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</tr>
<tr>
<td>Alga</td>
<td>assesses_effect_of</td>
<td>Alga</td>
</tr>
<tr>
<td>Amino Acid Sequence</td>
<td>associated_with</td>
<td>Amino Acid Sequence</td>
</tr>
</tbody>
</table>

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

- Select Tab along top
- Quick search
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.
Specialist Lexical Record

<table>
<thead>
<tr>
<th>base</th>
<th>Addison's disease</th>
</tr>
</thead>
<tbody>
<tr>
<td>entry</td>
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</tr>
<tr>
<td>cat</td>
<td>noun</td>
</tr>
<tr>
<td>variants</td>
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</tr>
<tr>
<td>variants</td>
<td>reg</td>
</tr>
</tbody>
</table>

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>Java RMI</th>
<th>TCP/IP socket</th>
</tr>
</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</td>
<td>◆ XML schema</td>
</tr>
<tr>
<td>download)</td>
<td>◆ Socket server</td>
</tr>
<tr>
<td>◆ Detailed <em>Javadoc</em> documentation online and</td>
<td>● Host: umlsks.nlm.nih.gov</td>
</tr>
<tr>
<td>with API download</td>
<td>● Port: 8042</td>
</tr>
<tr>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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

This guide describes the installation of the UMLSKS 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., kx - meta - c aids) that are to be executed by the server with the results sent back to the client.
UMLS Knowledge Source Server (UMLSKS)

Developer's 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

Download 

Documentation Java API
### Packages

- `gov.nih.nlm.kss.api`
- `gov.nih.nlm.kss.example`
- `gov.nih.nlm.kss.models`
- `gov.nih.nlm.kss.models.models`
- `gov.nih.nlm.kss.models.models.meta`
- `gov.nih.nlm.kss.models.models.meta.attribute`
- `gov.nih.nlm.kss.models.models.meta.concept`
- `gov.nih.nlm.kss.models.models.meta.cooccurrence`
- `gov.nih.nlm.kss.models.models.meta.dataset`
- `gov.nih.nlm.kss.models.models.meta.deltas`
- `gov.nih.nlm.kss.models.models.meta.locator`
- `gov.nih.nlm.kss.models.models.meta.meshentry`
- `gov.nih.nlm.kss.models.models.meta.relation`
- `gov.nih.nlm.kss.models.models.sem`
- `gov.nih.nlm.kss.models.models.sem.rels`
- `gov.nih.nlm.kss.models.models.sem.units`
- `gov.nih.nlm.kss.query`
- `gov.nih.nlm.kss.query.query`
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>

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

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

<?xml version="1.0"?>
<SemanticTypeCollection version="1.0">
  <release>2004AB</release>
  <cui>C0033572</cui>
  <cn>Prostate</cn>
  <semanticType>
    <tui>T023</tui>
    <sty>Body Part, Organ,
         or Organ Component</sty>
  </semanticType>
</SemanticTypeCollection>
```
Sample XML query (4) Relationships

```xml
<?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>
  [...]
</RelationCollection>
```
Sample XML query (5) All semantic type IDs

```xml
<?xml version="1.0"?>
<listSemTypeIds version="1.0">
  <semnetId>
    <name>Acquired Abnormality</name>
    <ui>T020</ui>
  </semnetId>
  <semnetId>
    <name>Activity</name>
    <ui>T052</ui>
  </semnetId>
</listSemTypeIds>

[...]
```
Performing XML queries from UMLSKS

UMLS Knowledge Source Server (UMLSKS)

Metathesaurus
Semantic Network
SPECIALIST Lexicon

Documentation
- User's Guide
- Developer's Guide
- Developer's API Invadotes
- UMLS Documentation Set

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

Perform XML Queries

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.

Request AXML Relational Records

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

```
<?xml version="1.0"?>
<getRelations>
  <cui>C0033572</cui>
  <rel>RO</rel>
</getRelations>
```
Part II
How to use the UMLS?

(3) Installing the UMLS locally and Customizing the Metathesaurus using MetamorphoSys
What is MetamorphoSys?

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

◆ Simple to use

◆ Screens and tabs lead you through process

◆ Installs NLM data format files to local storage
Why use MetamorphoSys?

Customize the Metathesaurus

- To remove terminology that is unhelpful, or even harmful, to your needs and purposes

- To comply with terms of license agreement
Why use MetamorphoSys?

*Changing Default Settings*

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

- Requires a clear understanding of:
  - Characteristics of source vocabularies
  - License arrangements
  - User’s functional requirements
  - User’s purpose and perspective

- Technical expertise

... and requires a multidisciplinary technical team
Machine Requirements

◆ A fast CPU – 1 GHz or higher
◆ 1 GB RAM recommended (512 MB min.)
◆ 6x (or better) DVD drive
◆ 22 GB minimum free disk space

◆ Runs on Sun Solaris 8 & 9, Windows XP, NT, and 2000, Linux, and Mac
◆ 1-10 hours run time on platforms tested
Download from UMLSKS …

- High speed Internet connection required

- Read the README file for the release

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

Please README!
...or DVD?

Order at: umls_support@nlm.nih.gov

Include your license number

Run MetamorphoSys from DVD

- Windows
  - Autorun; or go to root directory and click on “windows_mmsys.bat”

- Linux, Solaris, Macintosh
  - open a terminal window, change to the root directory and type appropriate command: ./linux_mmsys.sh, ./solaris_mmsys.sh, ./macintosh_mmsys.sh
Be patient! A lot of software must load.
Welcome Screen
Install UMLS

Select one or all of these options
Install UMLS Advanced Options

Advanced Options

- Copy NLM data format files (.nlsm) 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 12 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

Operation is 17% Complete
Select a default subset

Level 0 → no separate additional license agreements
Level 0 + SNOMEDCT → Non-U.S. users must have separate license agreements
RxNorm → no separate additional license agreements
Input Options Tab

Customize the input of UMLS data. See Help for more information.

Input Format Options

Select Input Format

- NLM Data File Format

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.

Hold down the <Ctrl> key to select multiple rows.

To reset to the default Source List, click on Reset on the menu bar, and select Reset Source List.

<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>AIRHEUM, 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>3</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>B198</td>
<td>BI</td>
<td>ENG</td>
<td>2</td>
</tr>
<tr>
<td>Canonical Clinical Problem Statement System, 1999</td>
<td>CCPSS39</td>
<td>CCPSS</td>
<td>ENG</td>
<td>3</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>3</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
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</th>
<th>Key</th>
<th>Code</th>
</tr>
</thead>
<tbody>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>CP</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>CS</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>CX</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>HT</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>PC</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>PS</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>PT</td>
</tr>
<tr>
<td>International Classification of Primary Care, 1993</td>
<td>ICPC93</td>
<td>PX</td>
</tr>
<tr>
<td>ICPC, Basque Translation, 1993</td>
<td>ICPCBA0_1993</td>
<td>CP</td>
</tr>
</tbody>
</table>

Highlighted source term types will be marked as suppressible.
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

Input Format Options

Select Input Format:

- NLM Data File Format

NLM Data File Format

Source Folder - Location of Metathesaurus Files:

C:\Documents and Settings\tilley\Desktop\MINIMAGE
File menu

- Enable/Disable Filter
- Import Filter
- New Configuration
- Open Configuration
- Save Configuration
- Exit
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.

Select Input Format

NLM Data File Format

Source Folder - Location of Metathesaurus Files

D:\

Help for more information.
Edit menu

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

The UMLS Metathesaurus Configuration 2004AB options menu allows users to select various options for customizing the Metathesaurus. The menu includes options for MetamorphoSys, Advanced Input, Advanced Output, Advanced Source List, and Advanced Supressibility. Users can select the desired options and browse for input formats, such as NLM Data File Format. The source folder for the Metathesaurus files can also be specified.
Options menu

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

After you have customized your subset, 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

<table>
<thead>
<tr>
<th>File</th>
<th>Edit</th>
<th>Format</th>
<th>View</th>
<th>Help</th>
<th>mmsys.log - Notepad</th>
</tr>
</thead>
<tbody>
<tr>
<td>MetamorphoSys Version:..................</td>
<td>5.21</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>MetamorphoSys Build Date:.............</td>
<td>2004_08_30_14_47_11</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>UMLS Build Date:......................</td>
<td>2004_07_12_09_57_26</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Release Version:......................</td>
<td>2004AB</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Release Date:..........................</td>
<td>20040720</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Release Description:...................</td>
<td>July 2004 Release</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Metathesaurus Source paths:..........</td>
<td>C:\UMLS\DVDIMAGE</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Subsetted Metathesaurus folder:......</td>
<td>C:\UMLS\DVDIMAGE\2004AB\META</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Start at:............................</td>
<td>Wed Sep 01 13:04:18 EDT 2004</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Initialize CUI List completed:......</td>
<td>Wed Sep 01 13:06:20 EDT 2004</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Subset Metathesaurus completed.......</td>
<td>Wed Sep 01 14:13:02 EDT 2004</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Subset Index Files completed:.......</td>
<td>Wed Sep 01 14:21:11 EDT 2004</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Subset Release Metadata completed...</td>
<td>Wed Sep 01 14:21:17 EDT 2004</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Finished at:.........................</td>
<td>Wed Sep 01 14:22:48 EDT 2004</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Concepts in source:..................</td>
<td>1078246</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Concepts in subset:..................</td>
<td>1078246</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Time elapsed:.......................</td>
<td>01:18:29</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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
  Alternative Billing Concepts
  Alcohol and Other Drug Thesaurus, 2000
  Beth Israel Vocabulary, 1.0
  Canonical Clinical Problem Statement System, 1999
  Clinical Classifications Software, 2003
  Current Dental Terminology (CDT), 4
  COSTAR, 1989-1995
  Medical Entities Dictionary, 2003
  Physicians' Current Procedural Terminology, Spanish Translation,...
  CRISP Thesaurus, 2004
  COSTART, 1995
  Diseases Database, 2000
  German translation of ICD10, 1995
  German translation of UMDNS, 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>MR.COC.RRF</td>
<td>809,207 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.COLS.RRF</td>
<td>21 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.CONSO.RRF</td>
<td>596,528 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.CUI.RRF</td>
<td>9,221 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.CXT.RRF</td>
<td>9,391,778 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.DEF.RRF</td>
<td>17,172 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.DOC.RRF</td>
<td>88 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.FILES.RRF</td>
<td>4 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.HIER.RRF</td>
<td>899,786 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.HIST.RRF</td>
<td>70,843 KB</td>
<td>RRF File</td>
</tr>
<tr>
<td>MR.MAP.RRF</td>
<td>9,362 KB</td>
<td>RRF File</td>
</tr>
</tbody>
</table>
Summary
UMLS Overview

- UMLS = 3 Knowledge Sources
  - Metathesaurus
  - Semantic Network
  - SPECIALIST Lexicon and Lexical Tools

- MetamorphoSys
  - installs
  - customizes

- UMLSKS
  - remote access
  - resources and documentation
Questions
Documentation and Support
UMLS documentation and support

  - with links to all other UMLS information

  - 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)

<table>
<thead>
<tr>
<th>CUI</th>
<th>LAT</th>
<th>I1</th>
<th>LUI</th>
<th>LJS</th>
<th>SUI</th>
<th>ISPREF</th>
<th>AUI</th>
<th>SAUI</th>
<th>SCUI</th>
<th>SDUI</th>
</tr>
</thead>
<tbody>
<tr>
<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>
<td></td>
</tr>
<tr>
<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>
<td></td>
</tr>
<tr>
<td>C0001403</td>
<td>ENG</td>
<td>P</td>
<td>L0001403</td>
<td>VC</td>
<td>S0010794</td>
<td>Y</td>
<td>A0019740</td>
<td>M0000346</td>
<td>D000224</td>
<td></td>
</tr>
<tr>
<td>C0001403</td>
<td>ENG</td>
<td>S</td>
<td>L0494851</td>
<td>PF</td>
<td>S2164152</td>
<td>N</td>
<td>A2018589</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>C0001403</td>
<td>FRE</td>
<td>P</td>
<td>L3246333</td>
<td>PF</td>
<td>S3773545</td>
<td>Y</td>
<td>A3996251</td>
<td>D000224</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

### SAB (sample rows 1..5)

<table>
<thead>
<tr>
<th>SAB</th>
<th>I1</th>
<th>CODE</th>
<th>STR</th>
<th>SRL</th>
<th>SUPPRESS</th>
<th>CVF</th>
</tr>
</thead>
<tbody>
<tr>
<td>MTH</td>
<td>PN</td>
<td>NOCODE</td>
<td>Addison's disease</td>
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**Appendix - Metathesaurus relational files (RRF)**
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Appendix - Metathesaurus relational files (RRF)
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Appendix - Metathesaurus relational files (ORF)
### Appendix - Metathesaurus relational files (ORF)

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[...]

(2003AA)
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)
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[...]

Appendix - Metathesaurus relational files (ORF)
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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

(* = C0001403 | S0010794 | MSH)

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MRSAT  Simple concept attributes  (2003AA)

Appendix - Metathesaurus relational files (ORF)
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Appendix - Metathesaurus relational files (ORF)
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[...]

Appendix - Metathesaurus relational files (ORF)
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[...]
# MRCUI Concept history

(2003AA)

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Appendix - Metathesaurus relational files (ORF) (2003AA)
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**physically_related_to**

- Related by virtue of some physical attribute or characteristic.

**part_of**

- 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.

**isa**

- 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.
SRSTR Structure

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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)