



Medinfo 2007
Tutorial S009
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The Unified Medical Language System
What is it and how to use it?



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Outline

- ◆ What is the UMLS?
 - Introduction
 - Overview through an example
 - The three UMLS Knowledge Sources
- ◆ How to use the UMLS?
 - Obtaining a license
 - Remote access
 - Local installation and customization
 - A UMLS-based algorithm
 - Benefits and limitations




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Part I

What is the UMLS?

Outline

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



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Part I

What is the UMLS?


(1) Introduction

What does UMLS stand for?

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



UMLS®
Unified Medical Language System®
UMLS Metathesaurus®



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Motivation

- ◆ Started in 1986
- ◆ National Library of Medicine
- ◆ “Long-term R&D project”
- ◆ Complementary to IAIMS (Integrated Academic Information Management Systems)

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

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



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)
 - RRF browser (browsing subsets)



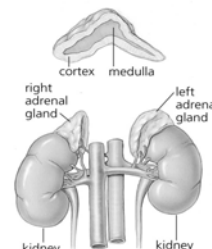
The UMLS is *not* an end-user application

Part I What is the UMLS?

(2) Overview through an example

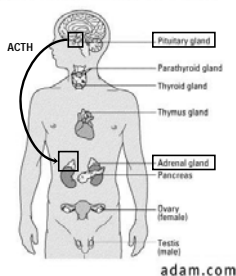
Addison's disease

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



Adrenal insufficiency Clinical variants

- ◆ Primary / Secondary
 - Primary: lesion of the adrenal glands themselves
 - Secondary: inadequate secretion of ACTH by the pituitary gland
- ◆ Acute / Chronic
- ◆ Isolated / Polyendocrine deficiency syndrome




Addison's disease: Symptoms

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



AD in medical vocabularies

- ◆ Synonyms: different terms
 - Addisonian syndrome } eponym
 - Bronzed disease } symptoms
 - Melasma addisonii } symptoms
 - Asthenia pigmentosa } symptoms
 - Primary adrenal deficiency } clinical variants
 - Primary adrenal insufficiency } clinical variants
 - Primary adrenocortical insufficiency } clinical variants
 - Chronic adrenocortical insufficiency } clinical variants
- ◆ Contexts: different hierarchies



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Organize terms

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

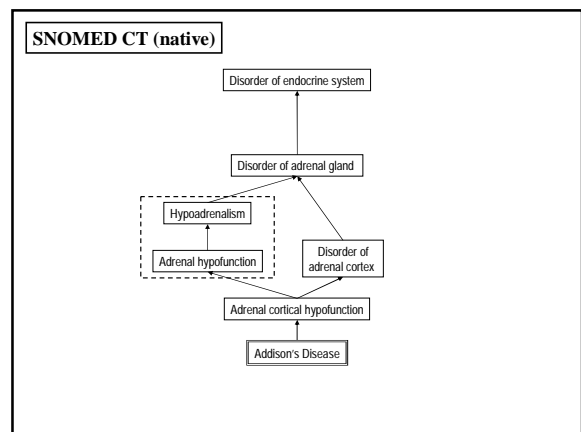
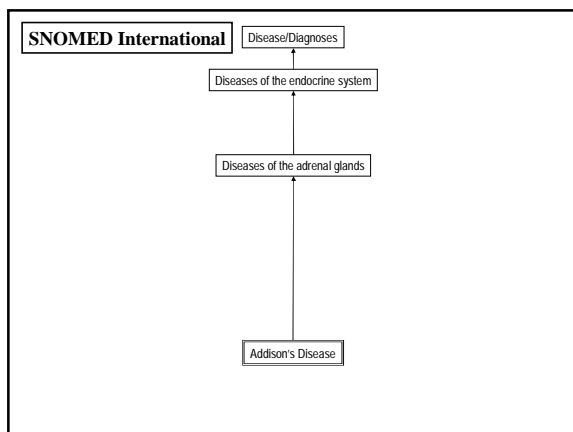
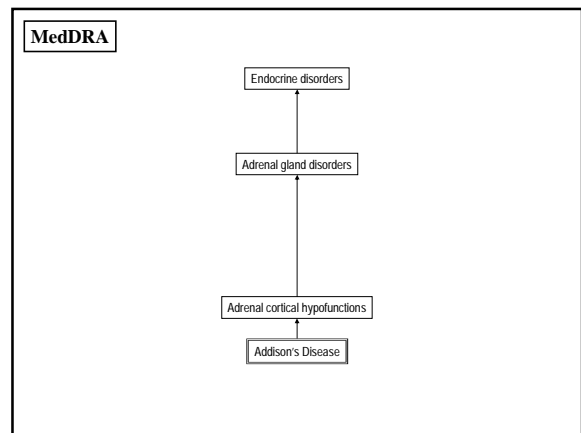
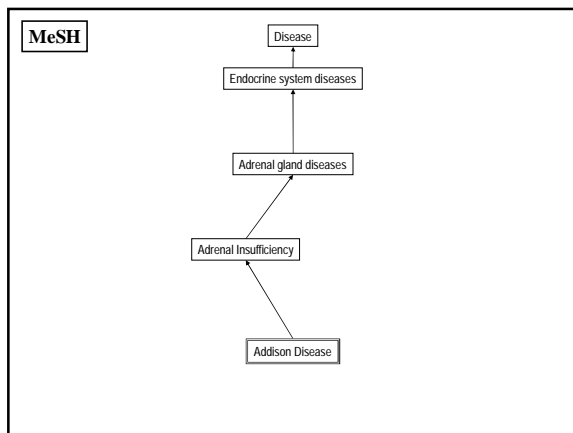
Addison Disease	MeSH	D000224
Primary hypoadrenalism	MedDRA	10036696
Primary adrenocortical insufficiency	ICD-10	E27.1
Addison's disease (disorder)	SNOMED CT	363732003

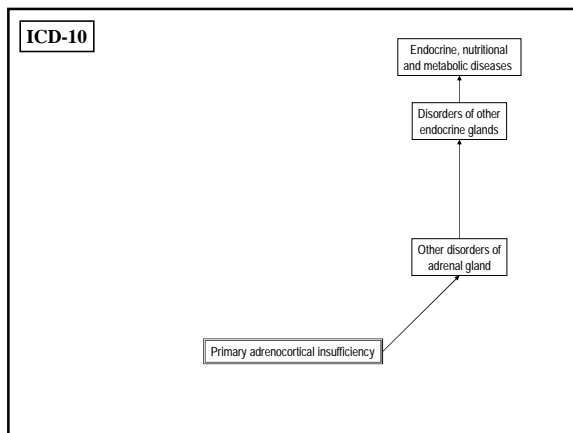
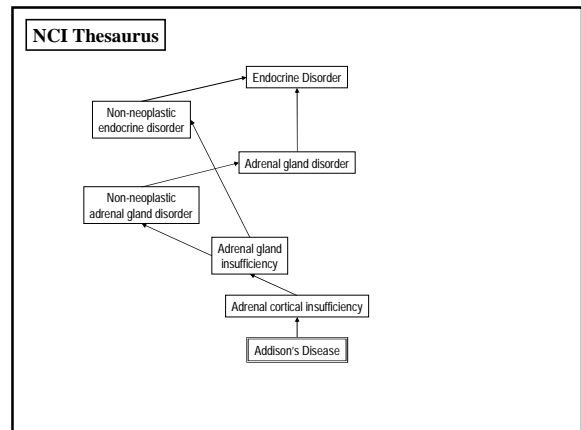
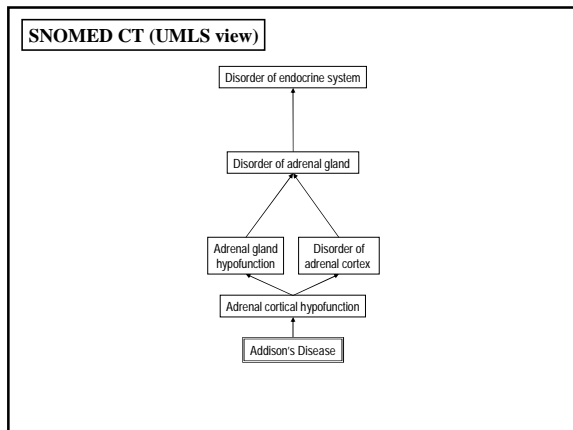
C0001403

Addison's disease



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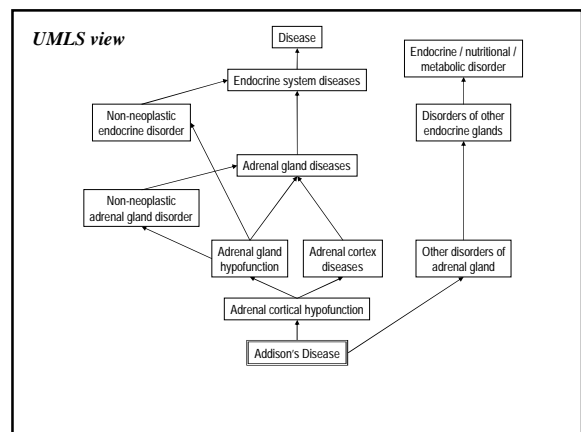
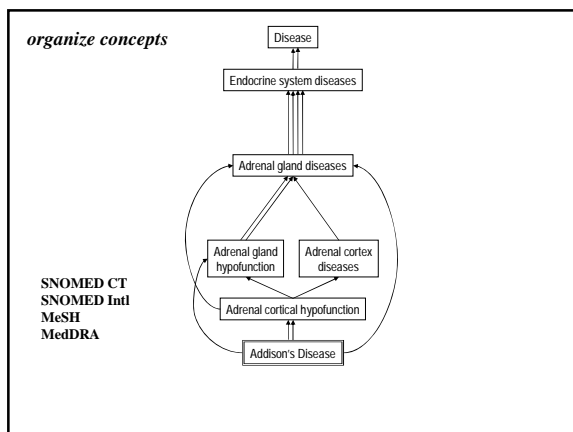


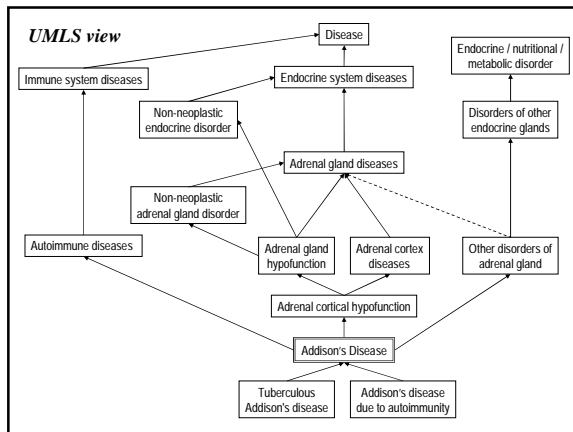
Organize concepts

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

The diagram shows three separate source vocabularies at the top, each with a root node (A, C, B) and its own set of child nodes (B, D, E, H; E, F, H; D, E; G, H). These are then integrated into a single graph below where node A is the root, and nodes B, C, D, E, F, G, H are connected to it through multiple paths, illustrating multiple inheritance and redundancy.

NLM 22





Relate to other concepts

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

NLM 26

Categorize concepts

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

NLM 27

How do they do that?

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

NLM 28

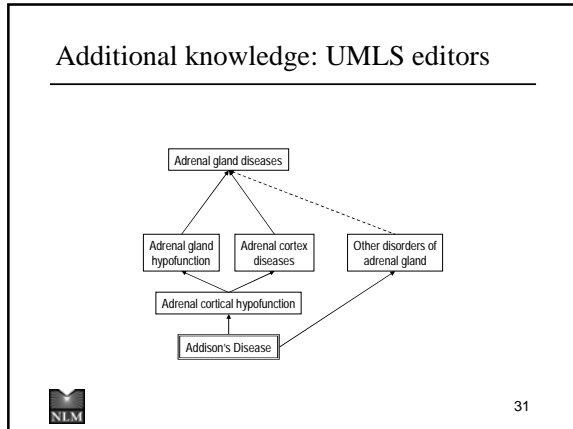
Lexical knowledge

NLM 29

Semantic pre-processing

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

NLM 30




- ### UMLS Summary
- ◆ Synonymous terms clustered into concepts
 - ◆ Unique identifier

 - ◆ Finer granularity
 - ◆ Broader scope
 - ◆ Additional hierarchical relationships
 - ◆ Semantic categorization
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Part I
What is the UMLS?

(3) UMLS Knowledge Sources


- ### Unified Medical Language System
- 
- ◆ SPECIALIST Lexicon
 - 360,000 lexical items
 - Part of speech and variant information
 - ◆ Metathesaurus
 - 6M names from over 100 terminologies
 - 1.5M concepts
 - 8M relations
 - ◆ Semantic Network
 - 135 high-level categories
 - 7000 relations among them
- Lexical resources
 Terminological resources
 Ontological resources
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UMLS Metathesaurus

- ### Metathesaurus Basic organization
- ◆ Concepts
 - Synonymous terms are clustered into a concept
 - Properties are attached to concepts, e.g.,
 - Unique identifier
 - Definition
 - ◆ Relations
 - Concepts are related to other concepts
 - Properties are attached to relations, e.g.,
 - Type of relationship
 - Source
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Source Vocabularies (2007AB)


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



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Biomedical terminologies

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




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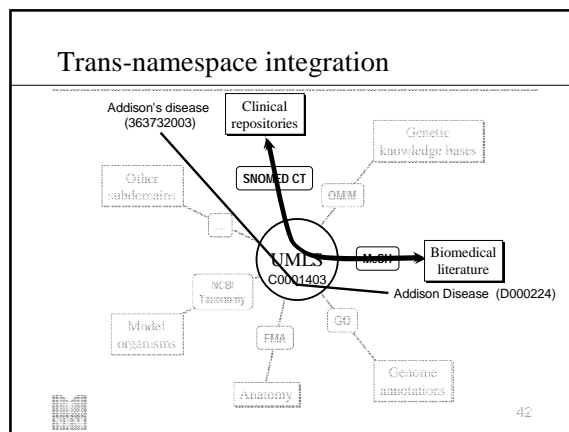
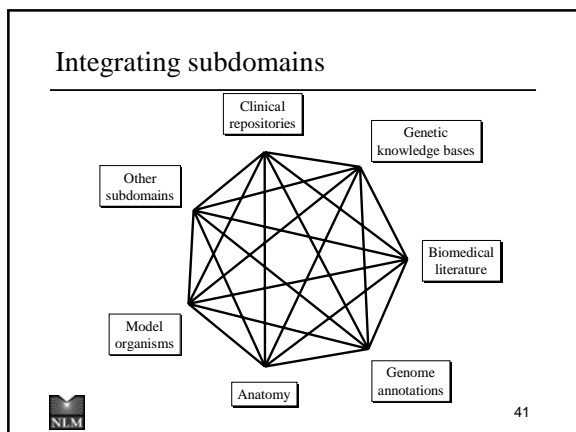
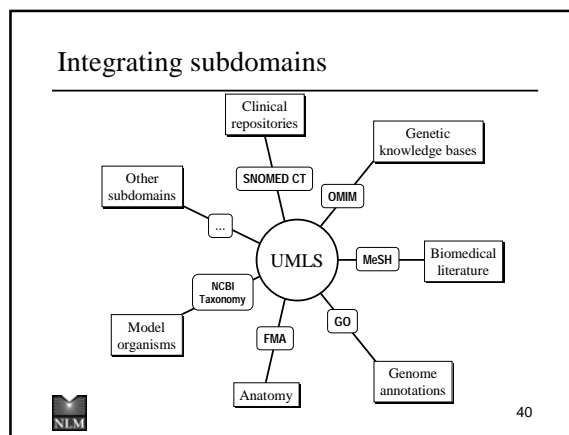
Biomedical terminologies (cont'd)

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

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



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Addison's Disease: Concept

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Metathesaurus Concepts (2007AB)

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

A0066000	Headache (MeSH)
A0065992	Headache (ICD-10)
S0046854	
A0066007	Headaches (MedDRA)
A12003304	Headaches (OMIM)
S0046855	
L0018681	
A0540936	Cephalodynia (MeSH)
S0475647	
L0380797	
C0018681	

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Cluster of synonymous terms

Term L0001403	80354372 Addison's disease 80010784 Addison's Disease 80010792 Addison Disease 80010786 Addisons Disease 50023558 Disease, Addison 50469271 Addison's disease, NOS	[...]
Term L0494940	S590736 Primary Adrenocortical Insufficiency S591678 Insufficiencies, Primary Adrenocortical	[...]
Term L0494851	S590734 Primary Adrenal Insufficiency S5924573 Adrenal Insufficiency, Primary	[...]
Term L0505243	S590743 Primary Hypoadrenalism 50718109 Primary hypoadrenalism	[...]
Term L3541031	S4115514 primary; hypoadrenocorticism S4090095 hypoadrenocorticism; primary	[...]
Term L1229627	S1471573 Addison-Krankheit	GER
Term L5345155	S6107160 Maladie d'Addison	FRE [...]

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Beyond concepts Descriptor level

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

MeSH Heading	Hydrocortisone	C0020268
Tree Number	D04.808.745.745.654.600	
Tree Number	D06.472.040.585.353.476	
Tree Number	D06.472.040.585.478.392	
Scope Note	The main glucocorticoid secreted by the ADRENAL CORTEX. Its synthetic counterpart is used, either as an injection or in inflammation, allergy, collagen diseases, arthritis, adrenocortical deficiency, shock, and some neoplastic conditions.	
Entry Term	11-Epicortisol	C0887247
Entry Term	Cortifair	C0699401
Entry Term	Cortisol	C0020268
Entry Term	Cortil	C0699402
Entry Term	Epicortisol	C0887247
Entry Term	Hydrocortisone, (11 alpha)-Isomer	C0887247
Entry Term	Hydrocortisone, (9 beta,10 alpha,11 alpha)-Isomer	C0887246

Beyond concepts Descriptor level

A0070119	Hydrocortisone	(MeSH)
A0043102	Cortisol	(MeSH)
C0020268		
A0066000	Epicortisol	(MeSH)
A0016625	11-Epicortisol	(MeSH)
A0055118	Hydrocortisone, (11 alpha)-Isomer	(MeSH)
C0887247		
A7801724	Hydrocortisone, (9 beta,10 alpha,11 alpha)-Isomer	(MeSH)
C0887246		
A7757592	Cortifair	(MeSH)
C0699401		
A7757595	Cortil	(MeSH)
C0699402		
D006854		

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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

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Metathesaurus Relationships

- ◆ Symbolic relations: ~8 M pairs of concepts
- ◆ Statistical relations : ~6 M pairs of concepts (co-occurring concepts)
- ◆ Mapping relations: ~150,000

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




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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



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Symbolic relationships Type


- ◆ Hierarchical
 - Parent / Child **PAR/CHD**
 - Broader / Narrower than **RB/RN**
- ◆ Derived from hierarchies
 - Siblings (children of parents) **SIB**
- ◆ Associative
 - Other **RO**
- ◆ Various flavors of near-synonymy
 - Similar **RL**
 - Source asserted synonymy **SY**
 - Possible synonymy **RQ**

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Symbolic relationships Attribute

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




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Mapping relations

- ◆ Simple mappings
 - <atom 1> mapped_to <atom 2>
 - e.g.,
 - SNOMED CT to ICD-9-CM
- ◆ Complex mappings
 - <atom 1> mapped_to <boolean expression>
 - e.g.,
 - ICD-9-CM to MeSH (search strategies)


NB: partially redundant with relations in MRREL



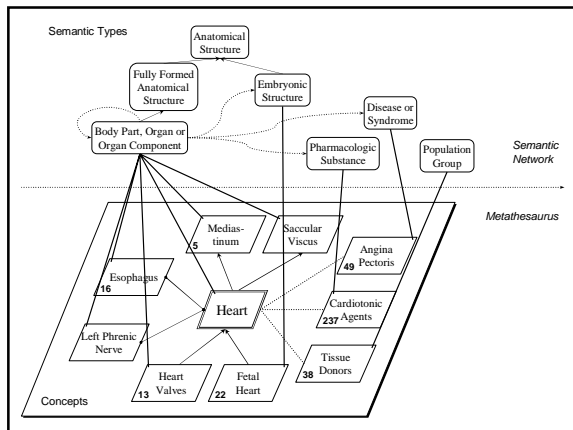
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Everything else

- ◆ Co-occurrence information (MRCOC)
 - Co- occurrence of MeSH descriptors in MEDLINE for the most part
- ◆ Source-specific attributes (MRSAT)
 - Legacy identifiers, external cross-references
 - SNOMED International legacy codes (SNOMED CT)
 - RxNorm to NDC
 - Concept status in a particular source (SNOMED CT)
 - Frequency of occurrence in MEDLINE (MeSH)
 - MedlinePlus URL (MeSH)
 - ...



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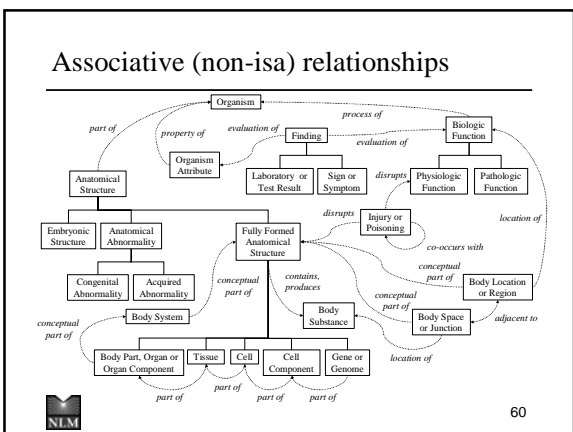
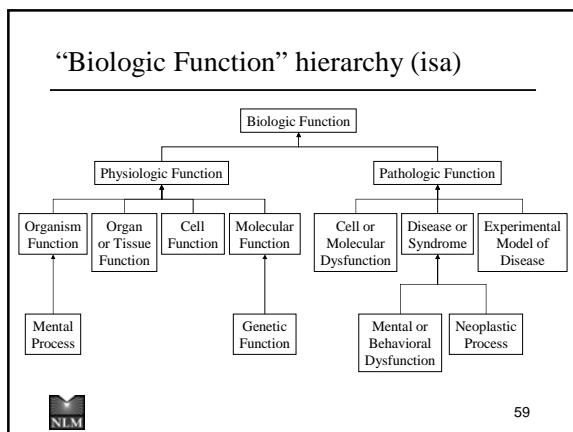
UMLS Semantic Network

Semantic Network

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

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



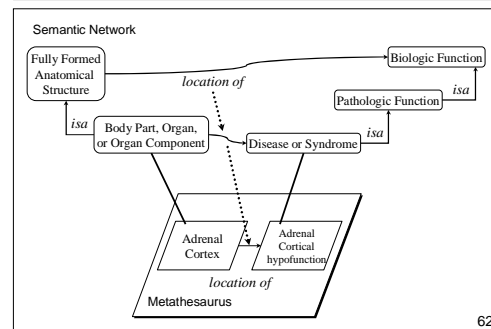
Why a semantic network?

- ◆ Semantic Types serve as high level categories assigned to Metathesaurus concepts, *independently of their position in a hierarchy*
- ◆ A relationship between 2 Semantic Types (ST) is a possible link between 2 concepts that have been assigned to those STs
 - The relationship may or may not hold at the concept level
 - Other relationships may apply at the concept level



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Relationships can inherit semantics



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SPECIALIST Lexicon and lexical tools

SPECIALIST Lexicon

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



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Morphology

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



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Orthography


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



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Syntax


- ◆ **Complementation**
 - verbs
 - intransitive I'll treat.
 - transitive He treated the patient.
 - ditransitive He treated the patient with a drug.
 - nouns
 - prepositional phrase
Valve of coronary sinus
- ◆ **Position for adjectives**



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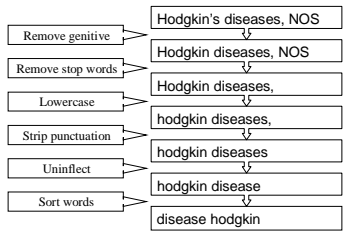

Lexical tools

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



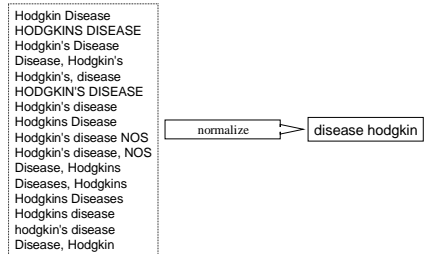

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Normalization

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Normalization: Example

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Normalization Applications


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



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Indexes


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



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Lexical Variant Generation program

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




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Part II

How to use the UMLS?

Outline

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







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Part II

How to use the UMLS?

(1) Obtaining a license

First step License agreement

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



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



License Agreement for Use of the UMLS® Metathesaurus®

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WHEREAS, the NLM was established by statute in order to assist the advancement of medical and related sciences, and to aid the dissemination and exchange of scientific and other information important to the progress of medicine and to the public health, (section 465 of the Public Health Service Act, as amended (42 U.S.C. section 285) and to carry out this purpose has been authorized to develop the Unified Medical Language System® (UMLS) to facilitate the retrieval and integration of machine-readable biomedical information from disparate sources;

WHEREAS, the NLM's UMLS project has produced the UMLS Metathesaurus, a machine-readable vocabulary knowledge source, that is useful in a variety of settings;

WHEREAS, the LICENSEE is willing to use the UMLS Metathesaurus at its sole risk and at no expense to NLM, which will result in information useful to NLM, may provide immediate improvements in biomedical information transfer to segments of the biomedical community, and is consistent with NLM's statutory functions;

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Accept & continue **Accept and continue** **Not accept**

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@Printer - Randy Verzon

APPENDIX A.1
Appendix to the License Agreement for Use of the UMLS® Metathesaurus
UMLS METATHESAUROS® SOURCE VOCABULARIES - 2007AB 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 use 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 editions of most of these vocabulary sources.

AIR93 - AIR93HDM Bethesda, (MD): National Library of Medicine, Letter Hill Center, 1993.
Contact: May Cheh, Letter Hill National Center for Biomedical Communications, National Library of Medicine, Building 38A, Room 9E902, 8600 Rockville Pike, Bethesda, MD 20894, e-mail: cheh@nlm.nih.gov

ALT2006 - Alternative Billing Concepts (AltBills) 7th Version, Las Cruces, NM, 2006.
CATEGORY 3 RESTRICTIONS APPLY **source restriction level**

Contact: Alternative Lask LLC, 6121 Indian School Road NE, Suite 131, Albuquerque, NM 87110, Phone: (505) 875-0001, Toll Free: (877)621-5465, Fax: (505) 875-0002, e-mail: alask@alternatelask.com

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WHOFRE_1997 - WHO Adverse Drug Reaction Terminology (WHOART) French Translation. Uppsala (Sweden) WHO Collaborating Centre for International Drug Monitoring, 1997.

CATEGORY 2 RESTRICTIONS APPLY

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

WHOGER_1997 - WHO Adverse Drug Reaction Terminology (WHOART) German Translation. Uppsala (Sweden) WHO Collaborating Centre for International Drug Monitoring, 1997.

CATEGORY 2 RESTRICTIONS APPLY

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

WHOPOR_1997 - WHO Adverse Drug Reaction Terminology (WHOART) Portuguese Translation. Uppsala (Sweden) WHO Collaborating Centre for International Drug Monitoring, 1997.

CATEGORY 2 RESTRICTIONS APPLY

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

WHOSPA_1997 - WHO Adverse Drug Reaction Terminology (WHOART) Spanish Translation. Uppsala (Sweden) WHO Collaborating Centre for International Drug Monitoring, 1997.

CATEGORY 2 RESTRICTIONS APPLY

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

Printer - Swedish Version

APPENDIX 2
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This is a Licence Agreement between (1) **The International Health Terminology Standards Development Organisation** (forming med-
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License Restriction Levels 0-4 (2007AB)

- ◆ Level 0 (79.3%)
 - *unrestricted*
- ◆ Level 1 (5.0%)
 - *negotiate to translate*
- ◆ Level 2 (0.5%)
 - *negotiate to use in health data creation*
- ◆ Level 3 (25.4%)
 - *negotiate to use in production*
 - *explicitly prohibited to provide Internet access*
- ◆ Level 4 (21.8%)
 - *SNOMED CT (unrestricted in member countries)*

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

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Part II


How to use the UMLS?

(2) Remote access

Remote Access

- ◆ UMLS Knowledge Source Server:
<http://umlsks.nlm.nih.gov>
- ◆ Web search interface
- ◆ Application Programming Interface (API)
- ◆ Coming soon: web services

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Knowledge Source Server

Web search interface

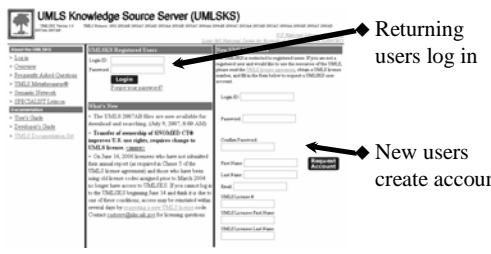
UMLSKS Web search interface

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




91

UMLSKS Web search interface log in





- ◆ Returning users log in
- ◆ New users create account



92

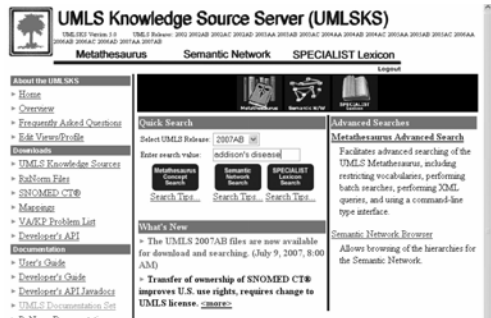

UMLS Knowledge Source Server Home Page

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

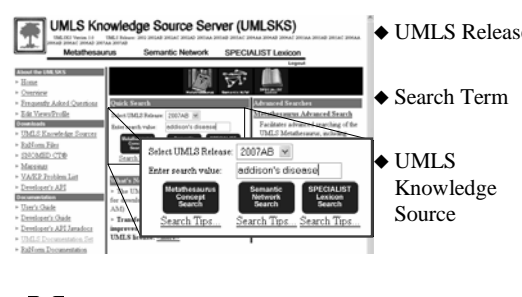
91

UMLS Knowledge Source Server Home Page





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Metathesaurus Basic Search Addison's disease

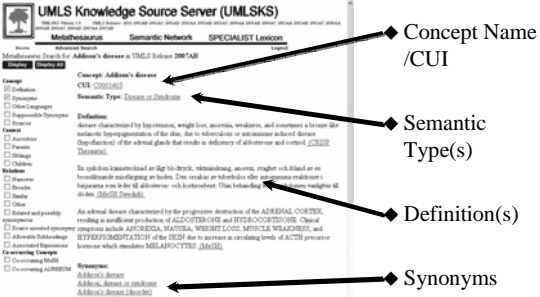


- ◆ UMLS Release
- ◆ Search Term
- ◆ UMLS Knowledge Source




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Concept Report Addison's disease



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



96

Display All

UMLS Knowledge Source Server (UMLS)

Metathesaurus Semantic Network SPECIALIST Lexicon

Metathesaurus Search for Adrenal gland insufficiency in UMLS Release 2004AB

Display Display All

“Display” shows results for selected options

“Display All” shows results for all available options

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Metathesaurus Basic Search

Adrenal gland insufficiency

UMLS Knowledge Source Server (UMLS)

Metathesaurus Semantic Network SPECIALIST Lexicon

Metathesaurus Search

Adrenal gland insufficiency

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

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Basic Concept Report

Adrenal gland insufficiency

UMLS Knowledge Source Server (UMLS)

Metathesaurus Semantic Network SPECIALIST Lexicon

Metathesaurus Search for Adrenal gland insufficiency in UMLS Release 2004AB

Display All

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Concept Report Display All

Adrenal Gland Insufficiency

UMLS Knowledge Source Server (UMLS)

Metathesaurus Semantic Network SPECIALIST Lexicon

Metathesaurus Search for Adrenal Gland Insufficiency in UMLS Release 2004AB

Display All

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

100

Concept Report Display All (continued)

Synonyms

Sources

101

Concept Report Display All (continued)

Hierarchies

102

Concept Report Display All (continued)

Narrower Concepts:
 Addison's disease (National Drug File - Reference Terminology) [Relation: isa]
 Addison's disease (Metathesaurus Names) [Relation: Terminology] [Relation: isa]
 Hypoadosteronism (National Drug File - Reference Terminology) [Relation: isa]
 Other adrenal hypofunction NOS (Metathesaurus Names) [Relation: Terminology]
 Adrenocorticosteroids (National Drug File - Reference Terminology) [Relation: isa]

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

Relations

Concept Report Display All (continued)

Co-occurrence data

Associated Expressions: None found.
 Co-occurring MeSH Terms:
 [64] Corticotropin
 [57] Hydrocortisone
 [38] Glucocorticoids
 [31] Endogenous oestrogen
 [30] Dog Diseases
 [28] Adrenal Glands

Metathesaurus Advanced Search Options

- ◆ Focused Search
- ◆ Raw Relational Records

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Metathesaurus Advanced Search Feature Focused Search

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

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Restricted Source Concept Report Addison's Disease

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

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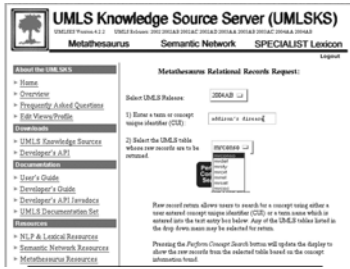
Addison's disease in SNOMED CT Preferred Term and Code

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

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Metathesaurus Advanced Search Feature Relational Record Request

- ◆ UMLS Release
- ◆ Search Term
- ◆ UMLS Relational Table



Relational Records MRCONSO.RRF



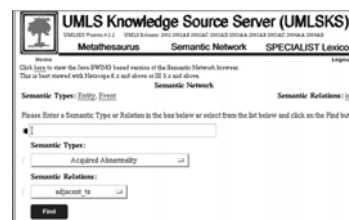
Semantic Network Searching

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



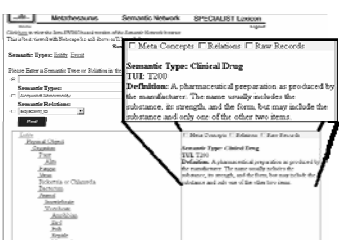
Semantic Network Search

- ◆ Enter search string
- or-
- ◆ Select semantic type
- or-
- ◆ Select semantic relation



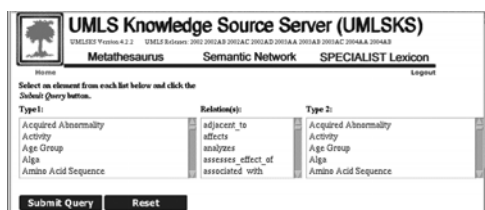
Semantic Type Clinical Drug

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



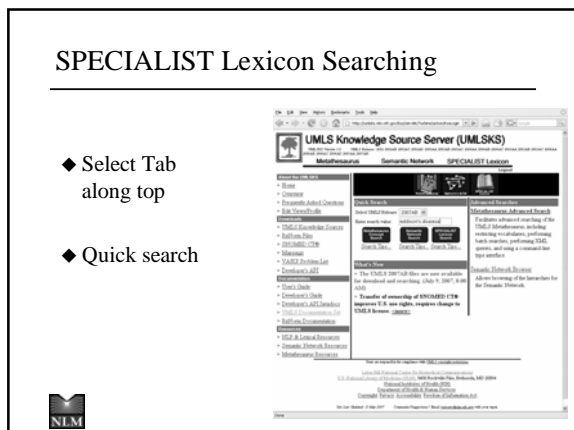
Show Relations Between Types

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

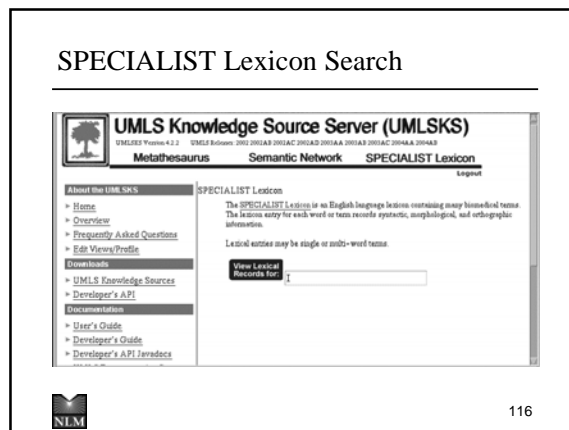


SPECIALIST Lexicon Searching

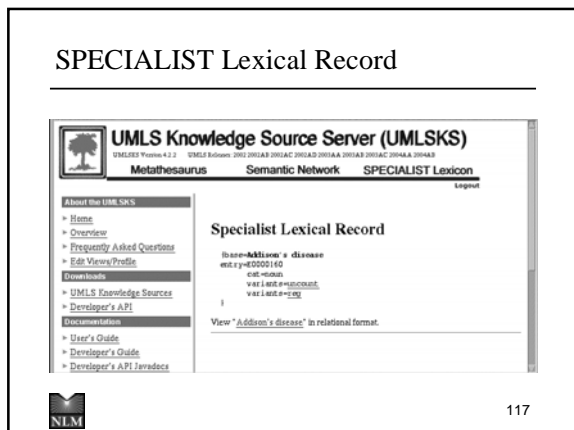
- ◆ Select Tab along top
- ◆ Quick search



SPECIALIST Lexicon Search

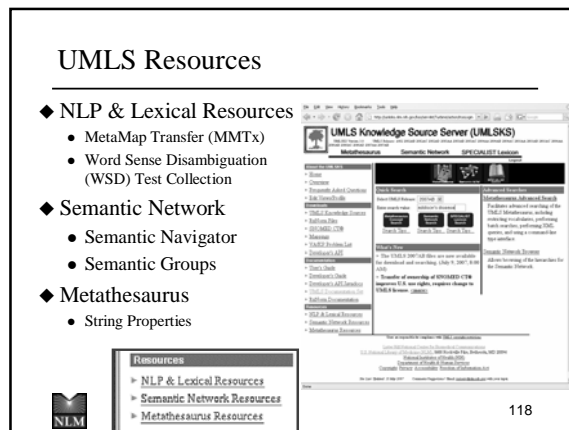


SPECIALIST Lexical Record

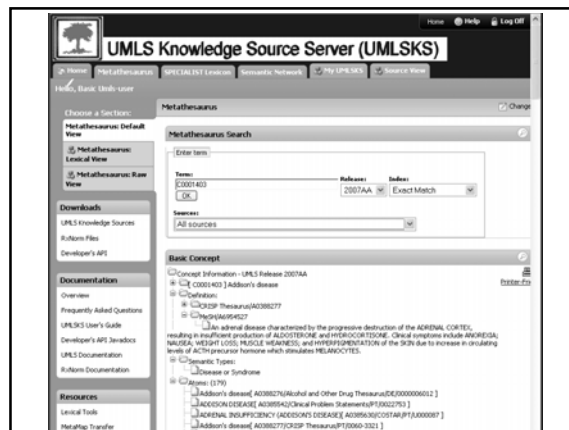
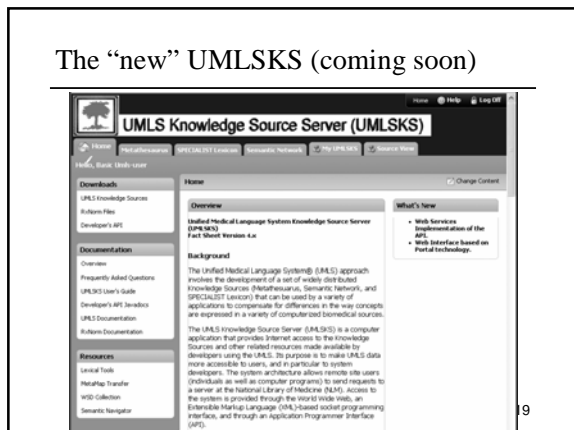


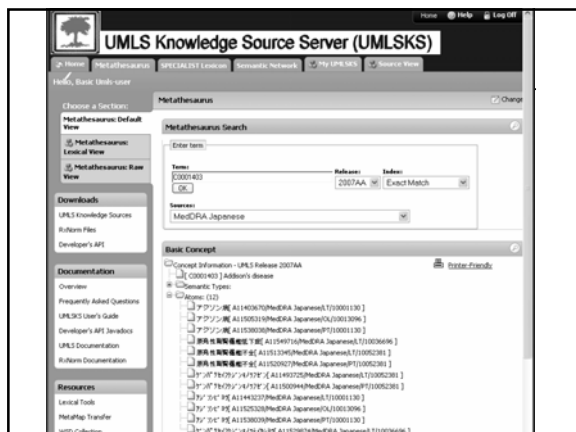
UMLS Resources

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



The "new" UMLS (coming soon)



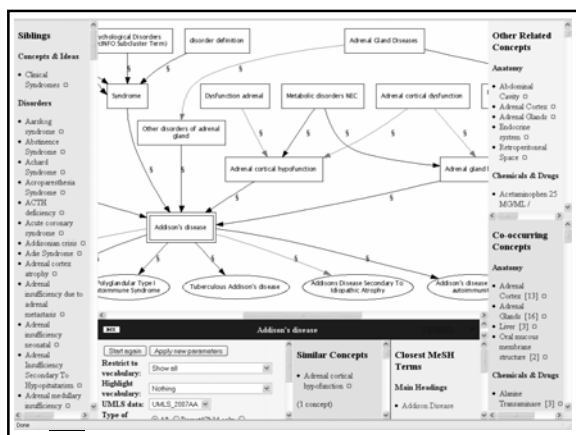


UMLS Semantic Navigator

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



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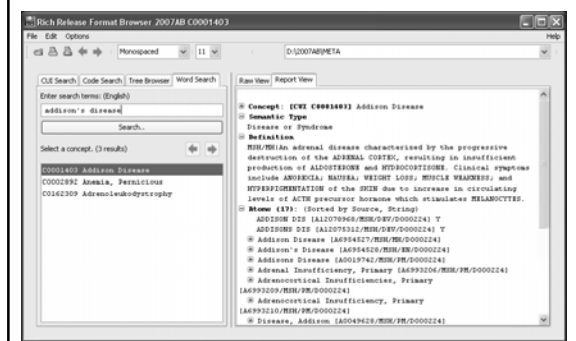
RRF Browser

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



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RRF Browser



Knowledge Source Server Application Programming Interface

UMLS SKS API basics

- ◆ Remote server at NLM
- ◆ Local application connected through

<h4>Java RMI</h4> <ul style="list-style-type: none"> ◆ Java-based applications ◆ Developer's Guide: Chapter 3 ◆ Set of Java classes (part of the UMLS SKS API download) ◆ Detailed <i>Javadoc</i> documentation online and with API download 	<h4>TCP/IP socket</h4> <ul style="list-style-type: none"> ◆ XML-based queries ◆ Developer's Guide: Chapter 5 ◆ XML schema ◆ Socket server <ul style="list-style-type: none"> ● Host: umlsks.nlm.nih.gov ● Port: 8042
--	---

NLM 127

Developer's Guide

The screenshot shows the 'Developer's Guide' page with a navigation bar containing 'Home', 'Table of Contents', and 'About This Guide'. Below the navigation bar is a section titled 'About This Guide' which describes the guide's purpose and audience. A 'Table of Contents' section lists chapters 1 through 5, each with a brief description of its content.

NLM 127

Documentation Java API

The screenshot shows the 'UMLS Knowledge Source Server (UMLS SKS)' website. The 'Developer's Guide' section is highlighted, showing a list of links for downloading the UMLS SKS API, including 'UMLS SKS API Download' and 'UMLS SKS API Documentation Set'. The page also includes a 'Table of Contents' and 'About This Guide' section.

NLM 129

Documentation Javadocs

The screenshot shows the 'UMLS SKS API Javadoc' page. It features a 'Packages' table listing various API packages such as 'gov.nlm.nih.gov.umlsks.api', 'gov.nlm.nih.gov.umlsks.api.exceptions', and 'gov.nlm.nih.gov.umlsks.api.messages'. The page also includes a 'Classes' section and a 'Index' section.

NLM 130

Sample XML query (1) Current version

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

⇒

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

NLM 131

Sample XML query (2) Concepts by string

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

⇒

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

NLM 132

Sample XML query (3) Concepts properties

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

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

NLM 133

Sample XML query (4) Relationships

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

```
<?xml version="1.0"?>
<RelationCollection version="1.0">
[...
<relation>
<cu>A3188910</cu>
<sab>SNOMEDCT</sab>
<relSource>
<cu>C0007112</cu>
<cn>Adenocarcinoma of prostate</cn>
<cu>A3318222</cu>
<rel>RO</rel>
<cu>R54806623</cu>
<rel>has_finding_site</rel>
</relSource>
</relation>
[...]
```

NLM

Sample XML query (5) All semantic type IDs

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

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

NLM 135

Performing XML queries from UMLS

NLM 136

Performing XML queries from UMLS

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

NLM 137

Part II

How to use the UMLS?

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

What is MetamorphoSys?

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



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Using MetamorphoSys

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



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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



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Why use MetamorphoSys?

Changing Default Settings

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



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Customization is Critical

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

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



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Machine Requirements

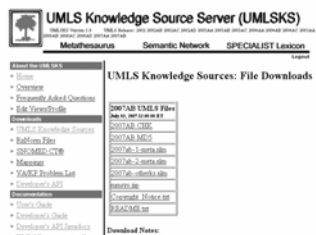
- ◆ A fast CPU – 1 GHz or higher
- ◆ 1 GB RAM recommended (512 MB min.)
- ◆ 6x (or better) DVD drive
- ◆ 22 GB minimum free disk space
- ◆ Runs on Sun Solaris 8 & 9, Windows XP, NT, and 2000, Linux, and Mac
- ◆ 1-10 hours run time on platforms tested



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Download from UMLSKS ...

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



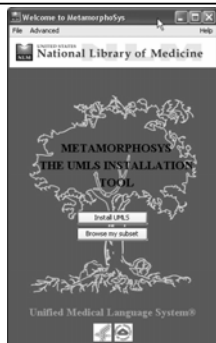
145

...or DVD?

- ◆ Order at: umls_support@nlm.nih.gov
- ◆ Include your license number
- ◆ Run MetamorphoSys from DVD
 - Windows
 - Autorun; or go to root directory and click on "windows_mmsys.bat"
 - Linux, Solaris, Macintosh
 - open a terminal window, change to the root directory and type appropriate command: `./linux_mmsys.sh`, `./solaris_mmsys.sh`, `./macintosh_mmsys.sh`

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Welcome Screen



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Install UMLS

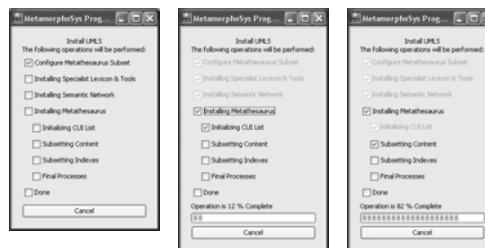


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UMLS License Notice



Installation progress monitor



150

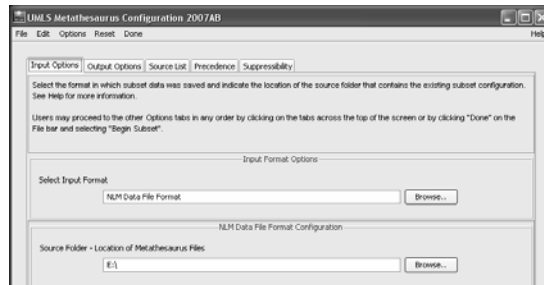
Select a default subset



- Level 0 → no separate additional license agreements
- Level 0 + SNOMEDCT → Users from non-IHTSDO member countries must have separate license agreements

151

Input Options Tab



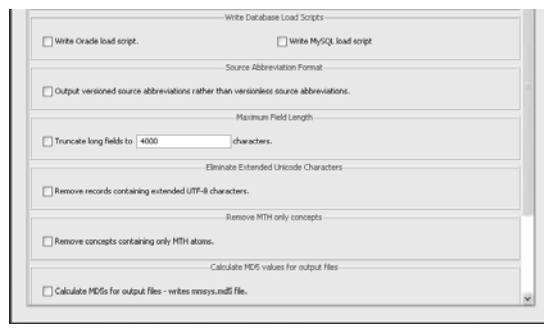
152

Output Options Tab

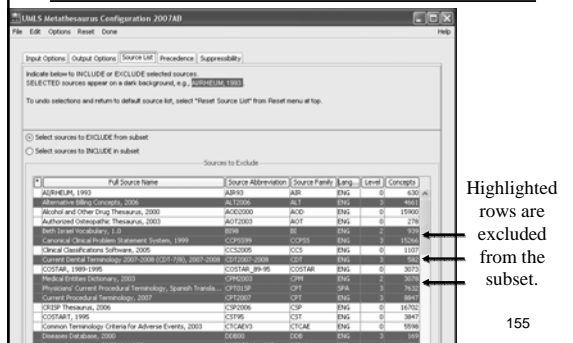


153

Output Options Tab



Source List Tab



Highlighted rows are excluded from the subset.

155

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

156

Suppressibility Tab

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

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

Select One or More Suppressible Term Types

Source	Source Abbreviation	Term Type
Logical Observation Identifier Names and Codes, 219	LNCT219	LN
Logical Observation Identifier Names and Codes, 219	LNCT219	LO
Logical Observation Identifier Names and Codes, 219	LNCT219	LPON
Logical Observation Identifier Names and Codes, 219	LNCT219	LPRN
Logical Observation Identifier Names and Codes, 219	LNCT219	LS
Logical Observation Identifier Names and Codes, 219	LNCT219	LSX
Logical Observation Identifier Names and Codes, 219	LNCT219	OSN
Logical Observation Identifier Names and Codes, 219	LNCT219	OSX
Logical Observation Identifier Names and Codes, 219	LNCT219	SN
Logical Observation Identifier Names and Codes, 219	LNCT219	SX
Logical Observation Identifier Names and Codes, 219	LNCT219	SXN
McMaster University Epidemiology Terms, 1962	MC219	PT
McMaster University Epidemiology Terms, 1962	MC219	PTX
MediWiki Health Topics_2004_08_14_20040814	MEDWIKELIS_20040814	ET
MediWiki Health Topics_2004_08_14_20040814	MEDWIKELIS_20040814	PT
MediWiki Health Topics_2004_08_14_20040814	MEDWIKELIS_20040814	SX
Medical Subject Headings, 2007_05_05	MSH2007_05_05	ST
Medical Subject Headings, 2007_05_05	MSH2007_05_05	STX

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File menu

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Edit menu

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Options menu

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Reset menu

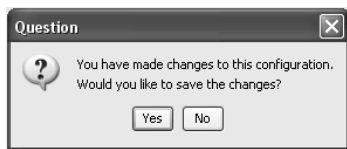
◆ Returns all filters to default selections
 ◆ Default selections in "mmsys.prop.default file" in config folder
 ◆ mmsys.prop.default contains properties in last run

161

Done – Begin Subset

162

Save configuration for next installation



163

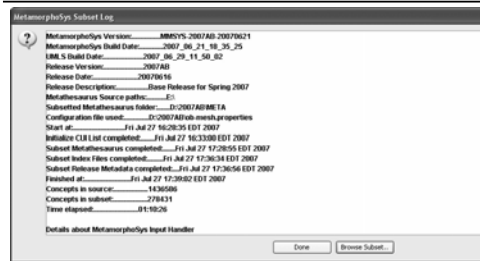
How MetamorphoSys Works

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



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MetamorphoSys log



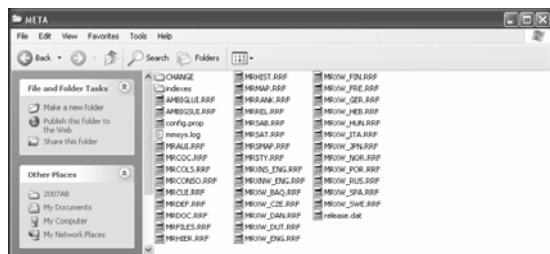
165

MetamorphoSys log



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Output directory contents



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Part II
How to use the UMLS?
(4) A UMLS-based algorithm

Indexing Initiative [Aronson & al., AMIA, 2000]

- ◆ For noun phrases extracted from medical texts, map to UMLS concepts
- ◆ Then, select from the MeSH vocabulary the concepts that are the most closely related to the original concepts

The diagram illustrates the indexing process. It starts with a box labeled 'Medical text' which points to a box labeled 'Noun phrase'. An arrow then points from 'Noun phrase' to a circular graph labeled 'UMLS'. From the 'UMLS' graph, an arrow points to a box labeled 'MeSH descriptor'.

NLM 169

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

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

NLM 170

Restrict to MeSH Synonymy

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

NLM 171

Restrict to MeSH Assoc. expressions

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

The diagram shows a text box on the left: 'Endoscopic removal of intraluminal foreign body from oesophagus without incision'. A double-headed arrow points to a tree structure on the right. The tree has a root node 'AND' which branches into three nodes: 'MH/SH', 'Esophagus', and 'Foreign Bodies'. The 'MH/SH' node further branches into 'surgery' and 'Foreign Bodies'.

NLM 172

Restrict to MeSH Ancestors

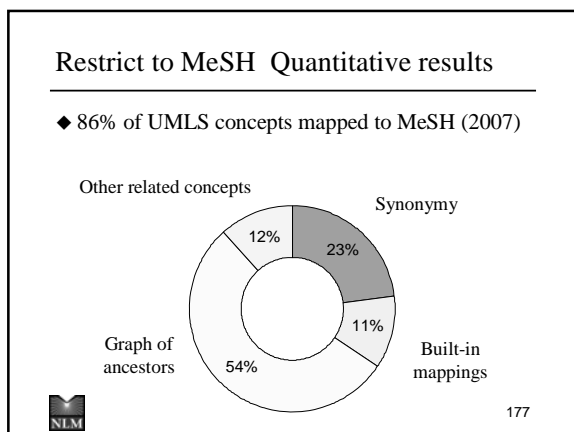
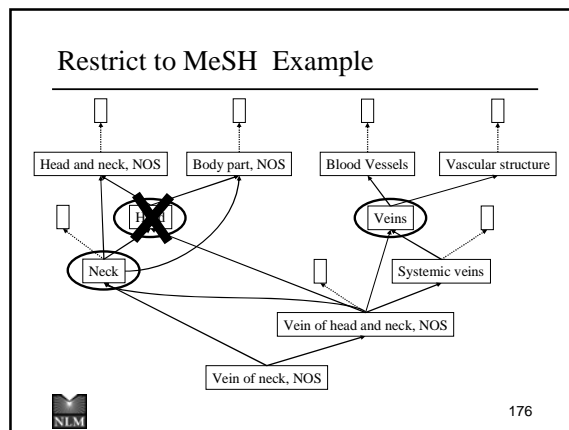
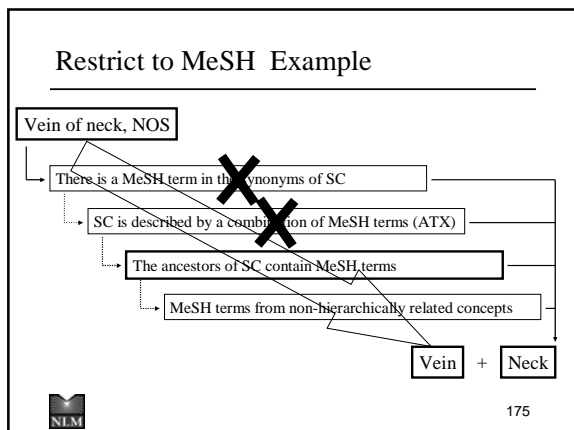
- ◆ If not, let us build the graph of the ancestors of this concept
 - using parents and broader concepts (MRREL)
 - all the way to the top
 - excluding ancestors whose semantic types are not compatible with those of the source concept (MRSTY)
- ◆ From the graph, select the concepts that come from MeSH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

NLM 173

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 (MRCONSO)

NLM 174




- ### 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
- 178

Part II
How to use the UMLS?
(5) Benefits and Limitations

Benefits


UMLS compared to individual vocabularies

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

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Direct benefits

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

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UMLS as an enabling resource


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

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Limitations

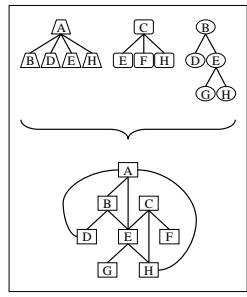
Limitations [Cimino, JAMA, 1998]


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

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Structural inconsistency From trees to graph

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



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Structural inconsistency Cycles in the UMLS graph

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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]

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Acyclicity

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

Reflexive
13,000

Direct
1800

Indirect
120

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Semantic inconsistency A two-level structure

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Semantic inconsistency A limited study

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

ICR and SNR not compatible

ICR = SNR or ICR descendant of SNR

Violation 13%

Validated 29%

Inferred 36%

Ambiguity 22%

ICR not specified and SNR compatible and multiple

ICR not specified and SNR compatible and unique

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

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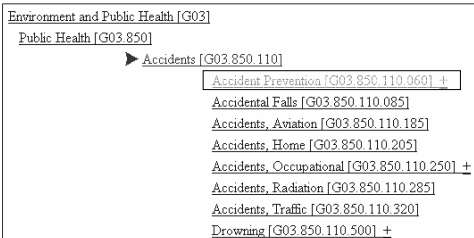
Semantic inconsistency Issues

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

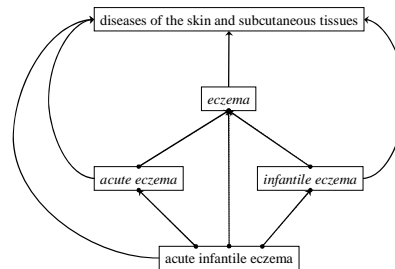
192

Underspecified relationships

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

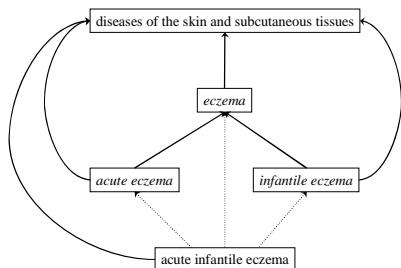


Missing relations Example



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Missing relations Example



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Missing relations A limited study

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

[Bodenreider & al., TIA, 2001]



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Compensation mechanisms

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



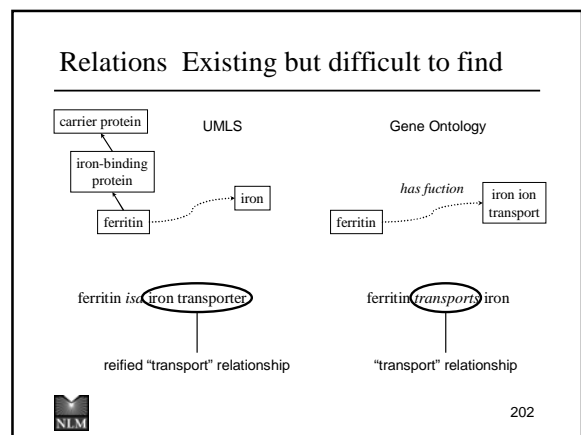
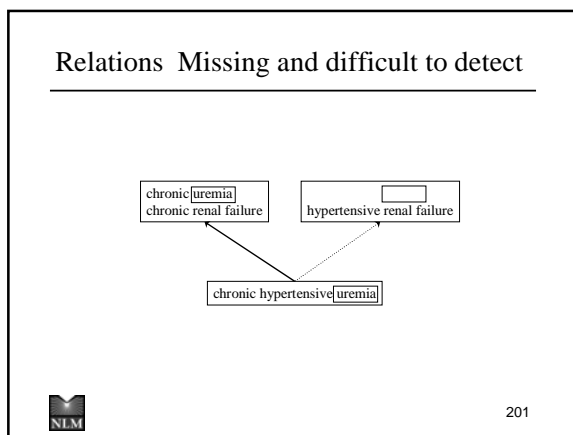
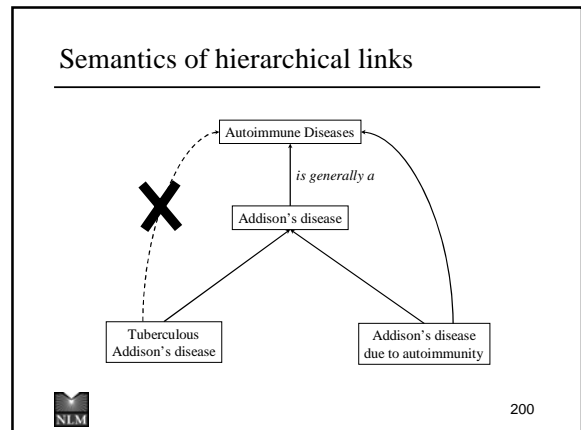
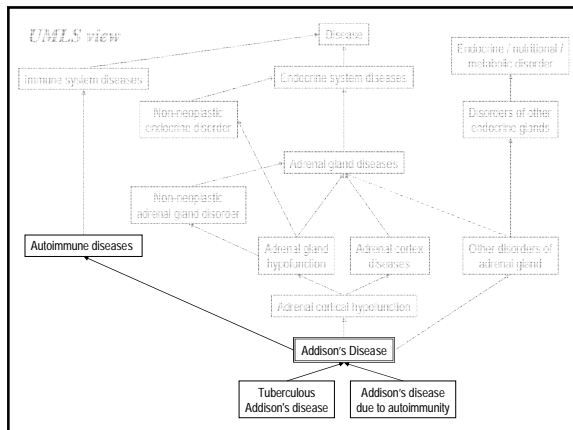
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More limitations

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



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- How to address these limitations?
- ◆ Description logics
 - ◆ Natural Language Processing (semantic interpretation of the terms)
 - ◆ Comparing knowledge sources (alignment, inference)

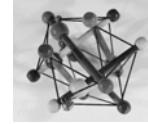
Summary

UMLS Summary

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



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Medical Ontology Research

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 Web: mor.nlm.nih.gov



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Lister Hill National Center
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 Bethesda, Maryland - USA

Bibliography

References: UMLS home page

- ◆ UMLS home page
 - [http:// www.nlm.nih.gov/research/umls/](http://www.nlm.nih.gov/research/umls/)
- ◆ UMLS documentation
 - Formerly know as the “Green Book”
 - Now online documentation
 - <http://www.nlm.nih.gov/research/umls/UMLSDOC.HTML>



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References

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



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References

- ◆ Technical papers
 - McCray, A. T., & Nelson, S. J. (1995). The representation of meaning in the UMLS. *Methods Inf Med*, 34(1-2), 193-201.
- ◆ Comprehensive bibliography 1986-96
 - <http://www.nlm.nih.gov/pubs/cbm/umlsbcm.html>




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Documentation and Support

UMLS documentation and support

- ◆ UMLS homepage
 - links to various UMLS resources
 - <http://www.nlm.nih.gov/research/umls/>
- ◆ UMLSKS homepage
 - links to the User's and Developer's guides
 - <http://umlsks.nlm.nih.gov/>
- ◆ UMLS mailing list
 - UMLSUSERS-L@LIST.NIH.GOV
- ◆ Email address for support
 - custserv@nlm.nih.gov



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Appendix

UMLS files in Rich Release Format

MRCONSO (sample rows 1..5) (2004AB)

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

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




Appendix - Metathesaurus relational files (RRF) 214

MRCONSO (sample rows 6..10) (2004AB)

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

12	13	14	15	16	17	18
SAB	ALI	CODE	STR	TRS	SUPPRESS	CVE
1	WHOFRE	IT	0410	MALADIE D'ADDISON	2	N
2	MSHGER	MH	D000224	Addison-Krankheit	3	N
3	MSHGER	SY	D000224	Bronzehautkrankheit	3	N
4	MSHJPN	MH	D000224	Addison病	3	N
5	MSHPJN	SY	D000224	副腎性黒皮症	3	N




Appendix - Metathesaurus relational files (RRF) 215

MRCONSO (sample rows 11-13) (2004AB)

1	2	3	4	5	6	7	8	9	10	11
CUI	LAT	SL	LUI	LLS	SUI	IRRES	AUI	SAUI	SCUI	SDUI
11	C0001403	POR	P	L3302998	PF	S3831123	N	A6382080		
12	C0001403	RUS	P	L336992	PF	S3864473	Y	A4157629		
13	C0001403	SPA	P	L1226877	PF	S1468823	Y	A1419475		

12	13	14	15	16	17	18
SAB	ALI	CODE	STR	TRS	SUPPRESS	CVE
11	MDRPPOR	LT	1001130	Doença de Addison	3	N
12	MSHRUS	MH	D000224	АДДИСОНОВА БОЛЕЗНЬ	3	N
13	WHOSPA	IT	0410	ADDISON, ENFERMEDAD	3	N



Appendix - Metathesaurus relational files (RRF) 216

MRHIER (sample rows) (2004AB)

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

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

Appendix - Metathesaurus relational files (RRF) 217

MRREL (sample rows) (2004AB)

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

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

Appendix - Metathesaurus relational files (RRF) 218

MRDEF (2004AB)

CUI	AUI	ATUI	SATUI	SAB	DEF	SUPPRESS	CVF
C0001403	A0019740	AT15061584		MSH	A disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin. It is due to tuberculosis- or autoimmune-induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol. In the absence of replacement therapy, it is usually fatal.	N	

Appendix - Metathesaurus relational files (RRF) 219

MRSAT (sample rows) (2004AB)

1	2	3	4	5	6
CUI	LUI	SUI	METAUI	STYPE	CODE
1 C0001403	L0001403	S0010792	A0019738	AUI	D000224
2 C0001403	L0001403	S0010794	A6326321	SCUI	C712
3 C0001403	L0001403	S0354372	A2922421	SAUI	363732003
4 C0001403			R15742591	SRUI	
5 C0001403				CUI	

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

Appendix - Metathesaurus relational files (RRF) 220

MRSTY (2004AB)

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

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MRHIST (sample rows) (2004AB)

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

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

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