Unified Medical Language System® (UMLS®) Basics

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Instructors:
Rachel Kleinsorge
Jan Willis

National Library of Medicine
National Institutes of Health
U.S. Department of Health & Human Services

Schedule

8:30 - 9:30	**Welcome and Introduction**
9:30 - 10:15	**Metathesaurus**
10:15 - 10:30	**BREAK**
10:30 - 11:45	**Metathesaurus, continued**
11:45 - 12:45	**LUNCH**
12:45 - 2:15	**MetamorphoSys & UMLS Tools**
2:15 - 2:30	**BREAK**
2:30 - 3:00	**SPECIALIST Lexicon & Lexical Tools** **Semantic Network**
3:00 – 3:30	**NLM, the UMLS, and Health Data Standards**
3:30 – 4:00	**Questions, Help, and Evaluation**
4:00 - 5:00	**Open Lab Time**

Acknowledgements

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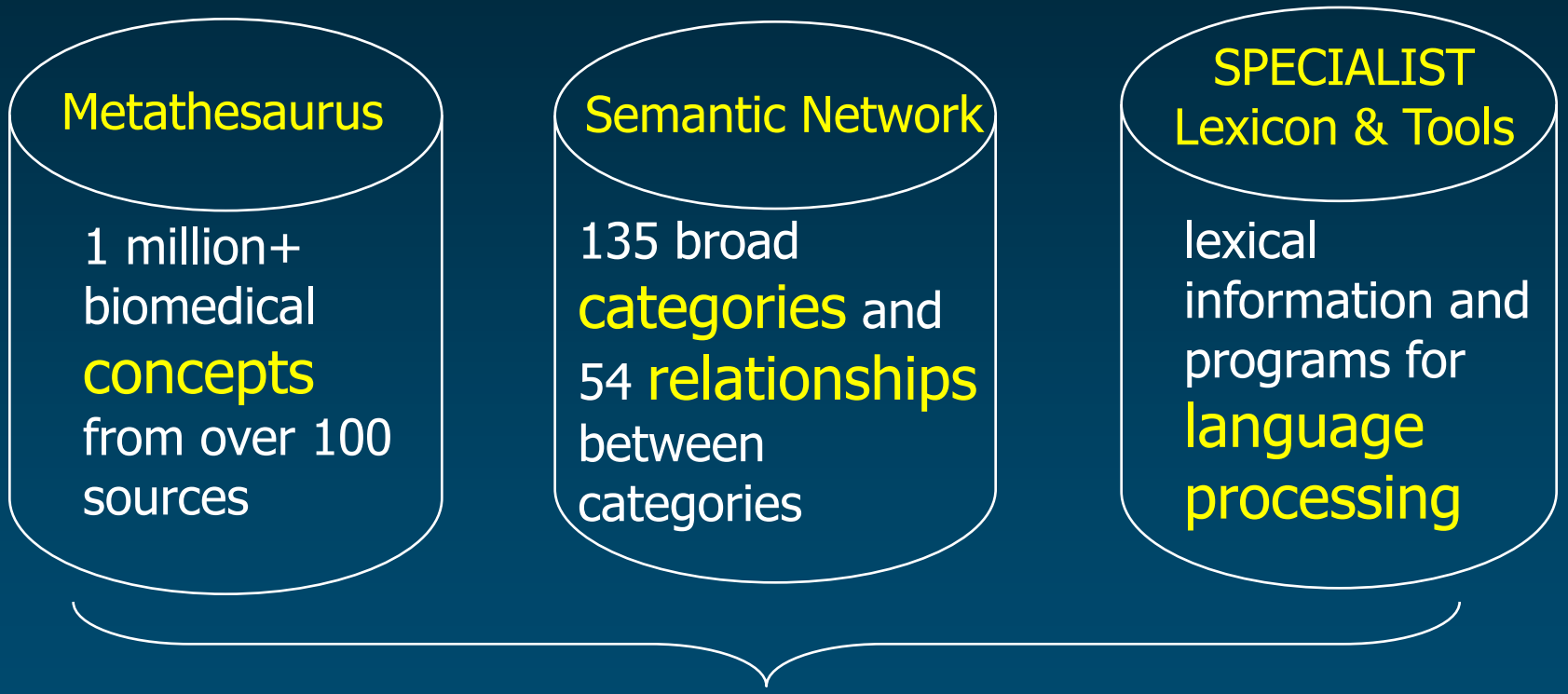
UMLS Video

http://www.nlm.nih.gov/research/umls/UMLS300kbps.swf

The UMLS consists of



3 Knowledge Sources

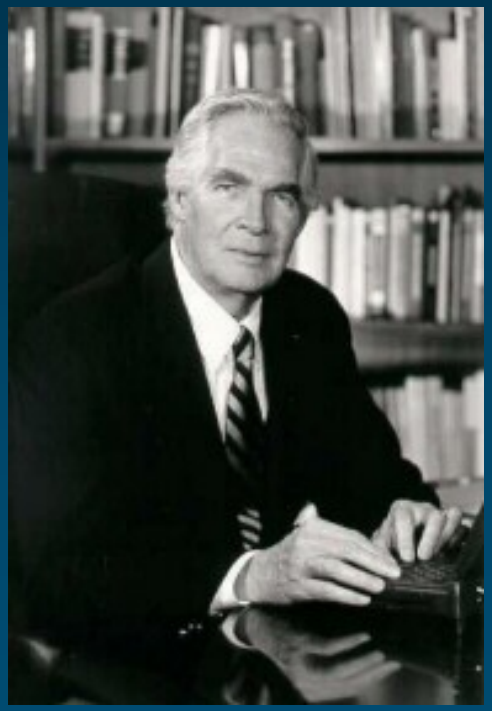
used separately or together

History of the UMLS

[Lindberg & al., *Methods*, 1993]
[Humphreys & al., *JAMIA*, 1998]

- Started at National Library of Medicine, 1986
- "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.»

UMLS Objectives

- Intellectual "middleware"
- A set of multi-purpose tools for system developers
- Knowledge Sources used to overcome:
  - disparities in language format
    Ex: atrial fibrillation, auricular fibrillation, af
  - disparities in granularity and perspective
    Ex: Contusions, hematoma, bruise
    Ex: Instruct patient to promptly report nosebleeds and excessive bruising (NIC), Epistaxis (MeSH)

UMLS Made available as:

- Data files
  - 3 separate sets of relational files
- Tools
  - MetamorphoSys (installation and customization)
  - RRF Subset Browser
  - lvg (lexical programs)
- Distributed on DVD; downloaded from UMLSKS

The UMLS is *not* an end-user application

UMLS Access

- UMLS Knowledge Source Server
  - http://umlsks.nlm.nih.gov/
  - Browser, Navigators
  - APIs
  - Download data files and programs
  - Documentation and other resources
- Local storage and customization (Metathesaurus)
  - MetamorphoSys
  - RRF Browser

UMLS Uses

- Information retrieval
- Thesaurus construction
- Natural language processing
- Automated indexing
- Electronic health records (EHR)
- Distribution mechanism for HIPAA, CHI, PHIN regulatory standards

2008AB UMLS: November 2008

- Metathesaurus:
  - 148 sources
  - 1,885,896 concepts
  - 17 languages
- Semantic Network:
  - 135 Semantic Types
  - 54 Semantic Relationships
- SPECIALIST Lexicon:
  - Over 330K records (over 557K inflected forms)

A guided tour of UMLS Resources

- http://www.nlm.nih.gov/research/umls/
- http://www.nlm.nih.gov/research/umls/documentation.html
- http://www.nlm.nih.gov/research/umls/listserv_info.html
- https://list.nih.gov/archives/umlsusers-l.html
- http://www.nlm.nih.gov/research/umls/new_users.html

What is the UMLS?

UMLS 3 Knowledge Sources

- Metathesaurus
  - Source vocabularies
  - Concepts
  - Relationships, Attributes
- Semantic Network
  - Semantic Types (categories)
  - Semantic Relationships
- Lexical resources
  - SPECIALIST Lexicon
  - Lexical tools

Metathesaurus: clusters terms by meaning

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

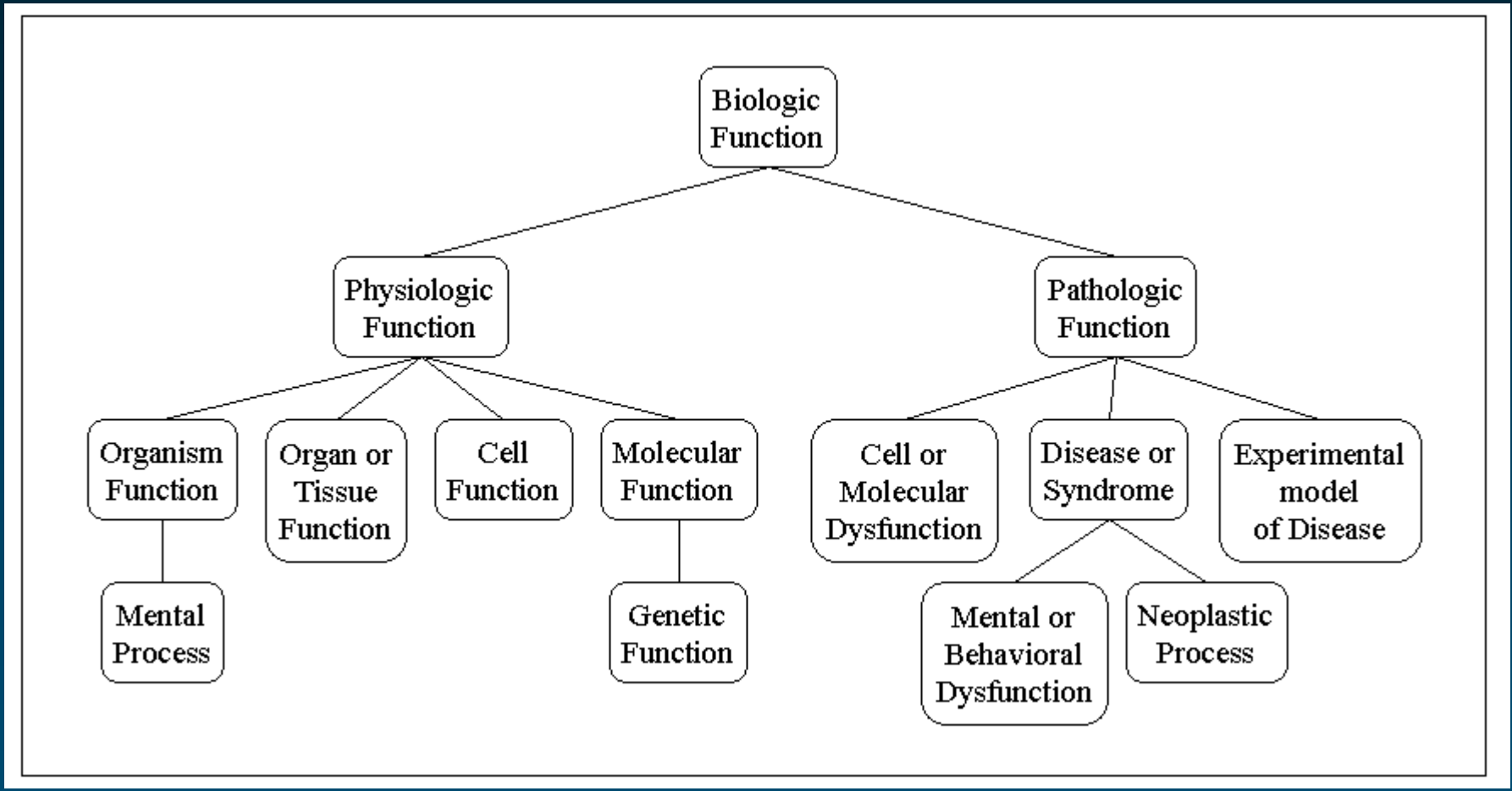
Addison's disease	Metathesaurus	PN	
Addison's disease	SNOMED CT	PT	363732003
Addison's Disease	MedlinePlus	PT	T1233
Addison Disease	MeSH	PT	D000224
Bronzed disease	SNOMED Intl 1998	SY	DB-70620
Deficiency; corticorenal, primary	ICPC2-ICD10 Thesaurus	PT	MTHU021575
Primary Adrenal Insufficiency	MeSH	EN	D000224
Primary hypoadreanlism syndrome, Addison	MedDRA	LT	10036696

C0001403 Addison's disease

Semantic Network

- 135 Semantic Types
  - Broad subject categories (Clinical Drug, Virus)
  - Ex:
    - **Addison's Disease**
    - **Semantic Type: Disease or Syndrome**
- 54 Semantic Relationships
  - Links between categories (isa, causes, treats)
  - Ex:
    - Virus **causes** Disease or Syndrome
- Types + Relationships
  - Form the structure of the semantic network
  - Broadly categorize the biomedical domain

Semantic Network: Biologic Function



SPECIALIST Lexicon & Lexical Tools

- General English lexicon of common words, biomedical terms
- Used by SPECIALIST Natural Language Processing System
- Lexical records contain encoded information:
  - syntactic (how words are put together),
  - morphological (inflection, derivation, and compounding), and
  - orthographic (spelling) information
- Lexical tools process terms
- Used in Metathesaurus production

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

Lexical Tools

- Manage lexical variation in biomedical terminologies and text
- Used separately or with SPECIALIST Lexicon
- Perform transformations selected and ordered by users
- 3 primary programs: normalizer, word index generator, lexical variant generator

Summary

Metathesaurus:

- clusters terms into concepts – assigns unique identifier

Semantic Network

- defines relationships between concepts, organizes concepts into categories

Lexicon and Lexical Tools:

- processes terms for entry into the Metathesaurus

UMLS Bookmark

NATIONAL LIBRARY OF MEDICINE®

Unified Medical Language System® (UMLS®)

SPECIALIST LEXICON

Metathesaurus®

SEMANTIC NETWORK

www.nlm.nih.gov/research/umls

The UMLS is a collection of tools and resources that enable **systems** to link biomedical information from different sources, such as patient record systems, bibliographic databases, factual databases, decision support systems and other biomedical and health applications.

The UMLS also improves the ability of computer programs to understand biomedical meaning in user inquiries, and to use that understanding to retrieve and integrate relevant information for users.

The UMLS consists of data files and programs organized into three Knowledge Sources. Those sources are:

METATHESAURUS — Contains information about biomedical concepts and terms from over 100 controlled vocabularies and classification, including U.S. clinical and administrative standards. Primarily in English, with some foreign language translations. Accompanied by MetamorphoSys, a program that can be used to produce customized versions for particular applications.

SEMANTIC NETWORK — Defines broad subject categories to which Metathesaurus concepts are assigned, as well as the useful relationships that exist between categories.

SPECIALIST LEXICON — Includes commonly occurring English and biomedical words, as well as information necessary for natural language processing, such as word form, structure, and spelling variants. Accompanied by a suite of lexical programs.

Together these Knowledge Sources and programs provide useful tools to overcome differences in biomedical terminologies that often create barriers to effective retrieval and integration of biomedical information.

The UMLS Knowledge Source Server (umlsks.nlm.nih.gov) provides Web access to the component parts of the UMLS as well as the ability to download UMLS files and programs for local use

The UMLS does not include end-user applications beyond the Knowledge Source Server. Using the UMLS requires appropriate computing resources and technical expertise.

License Agreement — The UMLS is free of charge. Users complete a license agreement that protects the rights of the providers of component vocabularies. A separate license agreement is necessary with some sources.

ADDITIONAL INFORMATION

umlsinfo.nlm.nih.gov

- Fact Sheets
- Online Documentation
- License Agreement
- Knowledge Source Server
- Other UMLS Information, e.g., FAQs, Learning Resources, UMLS Users Listserv

custserv@nlm.nih.gov — NLM's e-mail address for questions on the UMLS and all other NLM resources

NATIONAL INSTITUTES OF HEALTH

NATIONAL LIBRARY OF MEDICINE

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES

4/2004 CB158

WWW.NLM.NIH.GOV

1-888-346-3656 • 8600 ROCKVILLE PIKE, BETHESDA, MD 20894

Metathesaurus

Metathesaurus

- The Metathesaurus is a
  - very large
  - multi-purpose
  - multi-lingual biomedical vocabulary
- It contains information about
  - biomedical and health related concepts
  - their various names
  - the relationships among them

Metathesaurus Sources

- Over 100 controlled vocabularies, code sets, and mappings
- Determine scope of Metathesaurus
- Source meanings, hierarchies preserved
- Relationships, Semantic Types added

Metathesaurus Sources 2

- 126 vocabularies in 17 languages
- Broad coverage of general, specialized biomedicine
- Varying purposes, structures, properties
- Sets of valid values:
  - Thesauri, e.g., MeSH, CRISP, NCI
  - Statistical classifications, e.g., ICD-9-CM
  - Billing codes, e.g., CPT, ABC Codes
  - Clinical coding systems, e.g., SNOMED CT
- Complete source lists in Appendixes A.1, B.4

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

Metathesaurus Sources 3

- Sources reflect regular and irregular update schedules
- NLM resources determine currency of sources in the Metathesaurus
- Priority given to maintaining critical standards vocabularies

Why Not Combine Them Into One?

- One size does not fit all
- NLM supports coordination when possible
- Growing awareness of benefits of standardization

"The UMLS approach assumes continuing diversity in the formats and vocabularies of different information sources and in the language employed by different elements of the biomedical community. It is not an attempt to build a single standard biomedical vocabulary."

Humphreys, BL and PL Schuyler, The Unified Medical Language System: Moving beyond the vocabulary of bibliographic retrieval. In: Broering NC, ed. High- Performance Medical Libraries: advanced information management for the virtual era. Westport (CT): Meckler; 1993, p. 33.

Metathesaurus Source Evaluation 1

1. Who does the vocabulary serve?
2. Is the vocabulary authoritative?
3. Is the thesaurus available in a well-structured, computable electronic form?
4. Have copyright ownership and license restriction levels been considered?

Metathesaurus Source Evaluation 2

5. What thesaurus characteristics and principles are used?

6. How fully descriptive are the terms when seen alone, rather than in the associated context?

7. Will there be many cases where the 'face' meaning of your terms are different from the meaning of the same term in other areas of biomedicine?

8. Who would be available to assist in editing and review?

Cimino's Desiderata

- Dr. James J. Cimino, M.D.
- http://www.dbmi.columbia.edu/cimino/
- Cimino JJ. Desiderata for Controlled Medical Vocabularies in the Twenty-First Century. *Methods Inform Med* 1998; 37: 394-403.
- Content
- Concept Orientation
- Concept Permanence
- Meaningless Concept Identifiers
- [….]

Source data in Metathesaurus files

Names, Synonyms Terms, Codes	→	MRCONSO
Relationships	→	MRREL
Hierarchies	→	MRHIER
Mappings	→	MRREL, MRMAP, MRSMAP
Attributes	→	MRSAT
Definitions	→	MRDEF

CUIs links concept data across files

Metathesaurus files

- Concepts (MRCONSO)
  - Synonymous and near-synonymous terms
  - Codes
  - Concept Unique Identifier (CUI), Semantic Type added
- Relationships and mappings (MRREL, MRHIER, MRMAP, MRSMAP)
  - Concepts are related to other concepts
  - Source hierarchies are represented
  - Relationship attributes specify type, source of relationship
  - Simple and complex maps
- Attributes (MRSAT), (MRDEF)

Concept cluster of synonymous terms

Concept C0001621

Term
adrenal disease gland
L0001621

S0011232 ***Adrenal Gland Diseases***
S0011231 **Adrenal Gland Disease**
S0000441 **Disease of adrenal gland**
S0481705 **Disease of adrenal gland, NOS**
S0220090 **Disease, adrenal gland**
S0044801 **Gland Disease, Adrenal**

Term
adrenal disorder gland unspecified
L0041793

S0860744 ***Disorder of adrenal gland, unspecified***
S0217833 **Unspecified disorder of adrenal glands**

Term
adrenal disorder
L0161347

S0225481 ***ADRENAL DISORDER***
S0627685 **DISORDER ADRENAL (NOS)**

Term
adrenal disorder gland
L0181041

S0632950 ***Disorder of adrenal gland***
S0354509 **Adrenal Gland Disorders**

Term
L0162317

S0226798 ***SURRENALE, MALADIES*** FRE

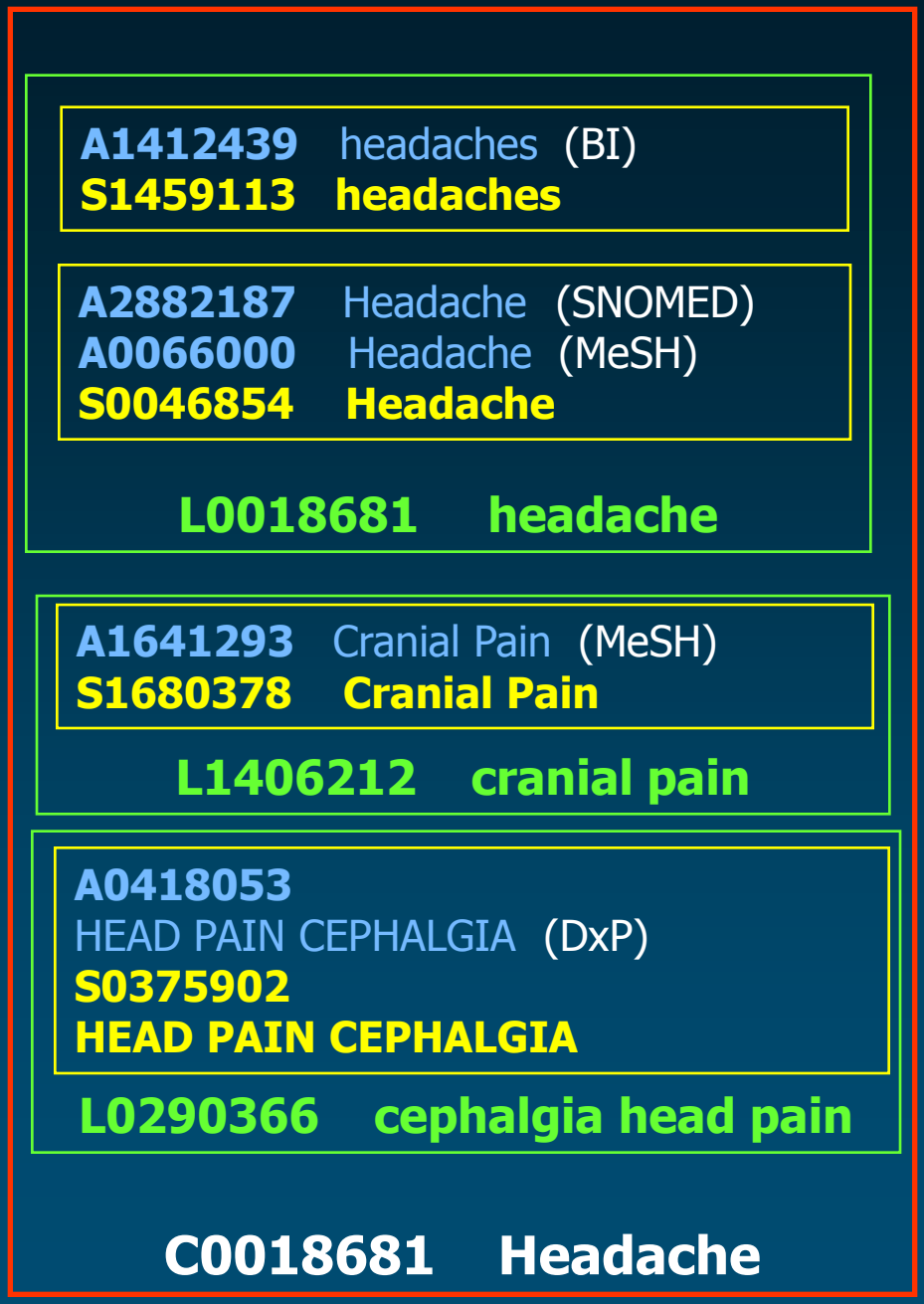
Concept structure

Concept (1.3 M) CUI
set of synonymous names

Term (> 4.6 M) LUI
set of normalized names

String (> 5.1 M) SUI
distinct concept name

Atom (> 6.2 M) AUI
concept name in source



Metathesaurus metadata, history and indexes

- Metadata files
  - MRSAB sources
  - MRFILES files
  - MRCOLS data elements
  - MRDOC finite values of selected data
  - MRRANK source/term type precedence
- Change files deleted, merged CUI, LUI
- MRCUI.RRF retired CUIs
- Concept name indexes

Representing ambiguity multiple meanings

- Same string, different meanings
- Cold (L0009264)
  - cold temperature (C0009264)
  - common cold (C0009443)
  - chronic obstructive lung disease (C0024117)
  - Cold sensation (C0234192)
- AMBIGLUI.RRF (AMBIG.LUI)

  L0009264|C0009443|
  L0009264|C0024117|
  L0009264|C0234192|
- AMBIGSUI.RRF (AMBIG.SUI)

  S0026353|C0009443|
  S0026353|C0234192|

Metathesaurus Distribution Formats

- Pipe-delimited text files
- UTF-8 character encoding

- Original Release Format (ORF) or Rich Release Format (RRF)

- Original Release Format (ORF)
  - Metathesaurus-Concept-Centric view
  - Explicit concept-based connection between terms in different sources
  - Most information represented at concept (CUI) level

Rich Release Format (RRF)

- Source-Centric View supports source transparency
- Represents original source information at atom (AUI) level
- Atom = unit of meaning (name) in a source
- New data fields and files to capture source specific identifiers, mappings

Metathesaurus highlights

- Concept based
- Represents the meaning in each source
- Represents data in common format
- Adheres to principle of “source transparency”

- Tags source information
- Adds context-free unique identifiers

- Includes normalized word and string indexes produced using UMLS lexical tools

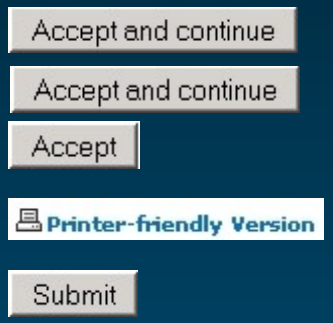
Accessing the Metathesaurus

- UMLSKS http://umlsks.nlm.nih.gov/
  - Simple and advanced searches
  - View concept reports and raw relational records

- RRF Browser in MetamorphoSys
  - Generate and view customized subset
  - Search and browse
  - View concept reports and raw relational records

Metathesaurus License Agreement 1

- Online Web-based license:
  http://www.nlm.nih.gov/research/umls/license.html
  - Read license
  - Read appendix 1
  - Read appendix 2
  - Print a copy for your records
  - Complete the Web form



- Verification and turnaround:
  - Receive e-mail from NLM and respond within 72 hours
  - NLM official countersigns, license added to database
  - Receive 2nd e-mail from NLM with new license number

Metathesaurus License Agreement 2

- 2. No charges, usage fees or royalties will be paid to NLM.
- 5. Within 30 days of the end of any calendar year … provide NLM with a brief report
- 11.c. required to include … identifiers from … the original source vocabularies
- 12. For material … from some sources additional restrictions … may apply.
- Special restrictions for SNOMED CT in light of the International Agreement

See list of current sources in Appendix A.1.

License Restriction Levels 0-4

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

67%

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

Metathesaurus Data Files

MRCONSO (sample rows 6..10) ⑩(2008AA)

	1 CUI	2 LAT	3 TS	4 LUI	5 STT	6 SUI	7 ISPREF	8 AUI	9 SAUI	10 SCUI	11 SDUI
6	C0001403	FRE	S	L1272481	PF	S1514427	Y	A1464383			
7	C0001403	GER	P	L1229627	PF	S1471573	Y	A7481335			D000224
8	C0001403	GER	S	L1239271	PF	S1481217	Y	A7485281			D000224
9	C0001403	JPN	P	L3437833	PF	S3965327	Y	A7888016			D000224
10	C0001403	JPN	S	L3442091	PF	S3969585	Y	A7890846			D000224

	12 SAB	13 TTY	14 CODE	15 STR	16 SRL	17 SUPPRESS	18 CVF
6	WHOFRE	IT	0410	MALADIE D'ADDISON	2	N	
7	MSHGER	MH	D000224	Addison-Krankheit	3	N	
8	MSHGER	EP	D000224	Bronzehautkrankheit	3	N	
9	MSHJPN	MH	D000224	Addison病	3	N	
10	MSHPJN	SY	D000224	副腎性黒皮症	3	N	

MRHIER (sample rows)

(2008AA)

	1	2	3	4	5	6
	CUI	AUI	CXN	PAUI	SAB	RELA
1	C0001403	A6954527	1	A6993207	MSH	
2	C0001403	A6954527	2	A0028022	MSH	
3	C0001403	A7559242	1	A7559213	PSY	
4	C0001403	A2922421	10	A3307650	SNOMEDCT	isa
5	C0001403	A2922421	11	A3307650	SNOMEDCT	isa

	7	8	9
	PTR	HCD	CVF
1	A0434168.A2367943.A2366890.A0135391.A6967433.A0020267.A6993207	C19.053.500.263	
2	A0434168.A2367943.A2366890.A0135391.A6970450.A0028022	C20.111.163	
3	A0449751.A7559213		
4	A3684559.A3886745.A3456474.A3456963.A3459284.A3473498.A6938225.A6919956.A6938229.A3307650		
5	A3684559.A3886745.A3456474.A3456963.A3459284.A6938487.A6938225.A6919956.A2933400.A2989549.A3307650		

MRSAT (sample rows)

(2008AA)

	1 CUI	2 LUI	3 SUI	4 METAUI	5 STYPE	CODE
1	C0001403	L0001403	S0010792	A6954527	SDUI	D000224
2	C0001403	L0001403	S0010794	A6326321	SCUI	C712
3	C0001403	L0001403	S0354372	A2922421	SAUI	363732003
4	C0001403	L0001403	S0354372	A2922421	SAUI	363732003
5	C0001403				CUI	

	7 ATUI	8 SATUI	9 ATN	10 SAB	11 ATV	14 SUPPRESS	15 CVF
1	AT38111026		DC	MSH	1	N	
2	AT33411754		MESH_UI	NDFRT	D000224	N	
3	AT24166602		DESCRIPTIONSTATUS	SNOMEDCT	0	N	
4	AT24529925		LANGUAGECODE	SNOMEDCT	en	N	
5	AT44908841		ST	MTH	R	N	

MRSTY

⑩(2008AA)

CUI	TUI	STN	STY	ATUI	CVF
C0001403	T047	B2.2.1.2.1	Disease or Syndrome	AT32681465	1792

MRDEF

⑩(2008AA)

CUI	AUI	ATUI	SATUI	SAB	DEF	SUPPRESS	CVF
C0001403	A0388277	AT51219985		CSP	Disease characterized by hypotension, weight loss, anorexia, weakness, and sometimes a bronze-like melanotic hyperpigmentation of the skin; due to tuberculosis or autoimmune induced disease (hypofunction) of the adrenal glands that results in deficiency of aldosterone and cortisol.	N	
C0001403	A6954527	AT43116350		MSH	An adrenal disease characterized by the progressive destruction of the ADRENAL CORTEX, resulting in insufficient production of ALDOSTERONE and HYDROCORTISONE. Clinical symptoms include ANOREXIA; NAUSEA; WEIGHT LOSS; MUSCLE WEAKNESS; and HYPERPIGMENTATION of the SKIN due to increase in circulating levels of ACTH precursor hormone which stimulates MELANOCYTES.	N	

MetamorphoSys

MetamorphoSys

- Multi-platform Java software
- Included in each UMLS release
- Unzips native Metathesaurus compressed files
- Installs Knowledge Sources to local storage
- Customizes a local Metathesaurus

Download from UMLSKS …

- High speed Internet connection required
- Files must be stored in the same folder
- 2006AD UMLS Files
  - **mmsys.zip (zipped MetamorphoSys application)**
  - 2006AD-1-meta.nlm (compressed Metathesaurus data)
  - 2006AD-2-meta.nlm (compressed Metathesaurus data)
  - 2006AD-otherks.nlm (compressed Semantic Network and SPECIALIST Lexicon)
  - 2006AD.CHK
  - 2006AD.MD5
  - Copyright_Notice.txt
  - **README.txt ```**

… or request DVD

- umls_support@nlm.nih.gov
- Include your license number
- Run MetamorphoSys from DVD

Machine Requirements

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

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

Customize the Metathesaurus

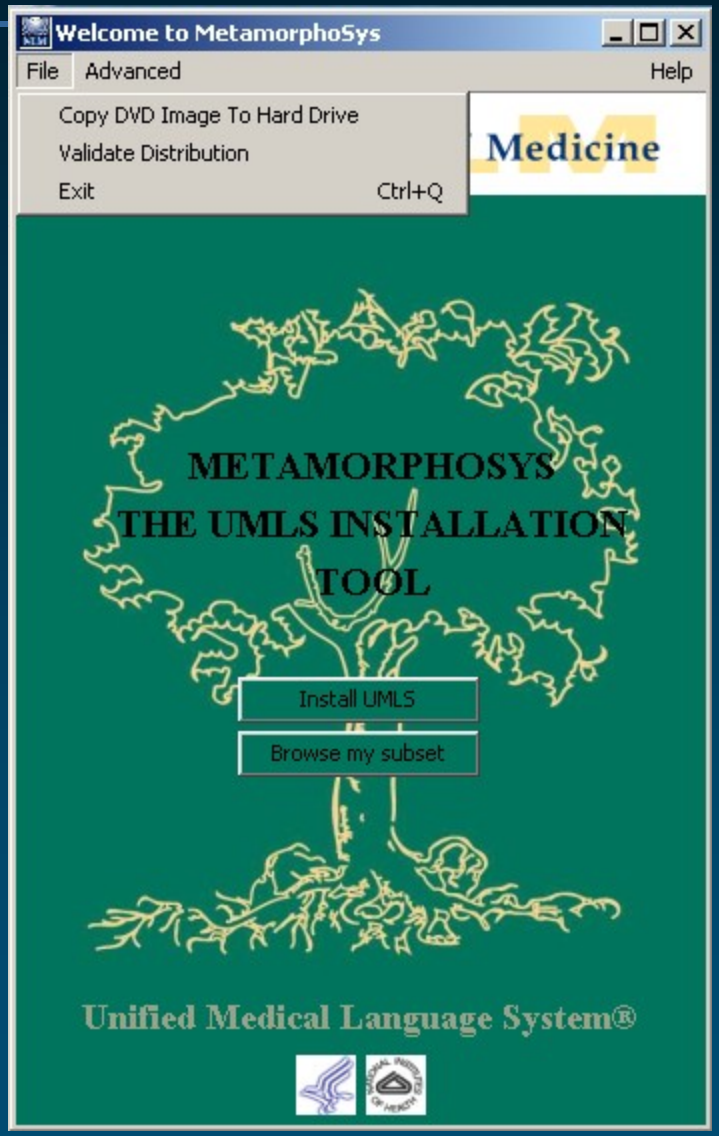
- Use MetamorphoSys
  - To comply with terms of license agreement
  - To remove unhelpful or harmful content
  - To change default settings (precedence, output)
- Customization is critical and requires understanding of:
  - Selected vocabularies
  - Functional requirements, purpose and perspective
- Technical expertise requires multidisciplinary team

How MetamorphoSys Works

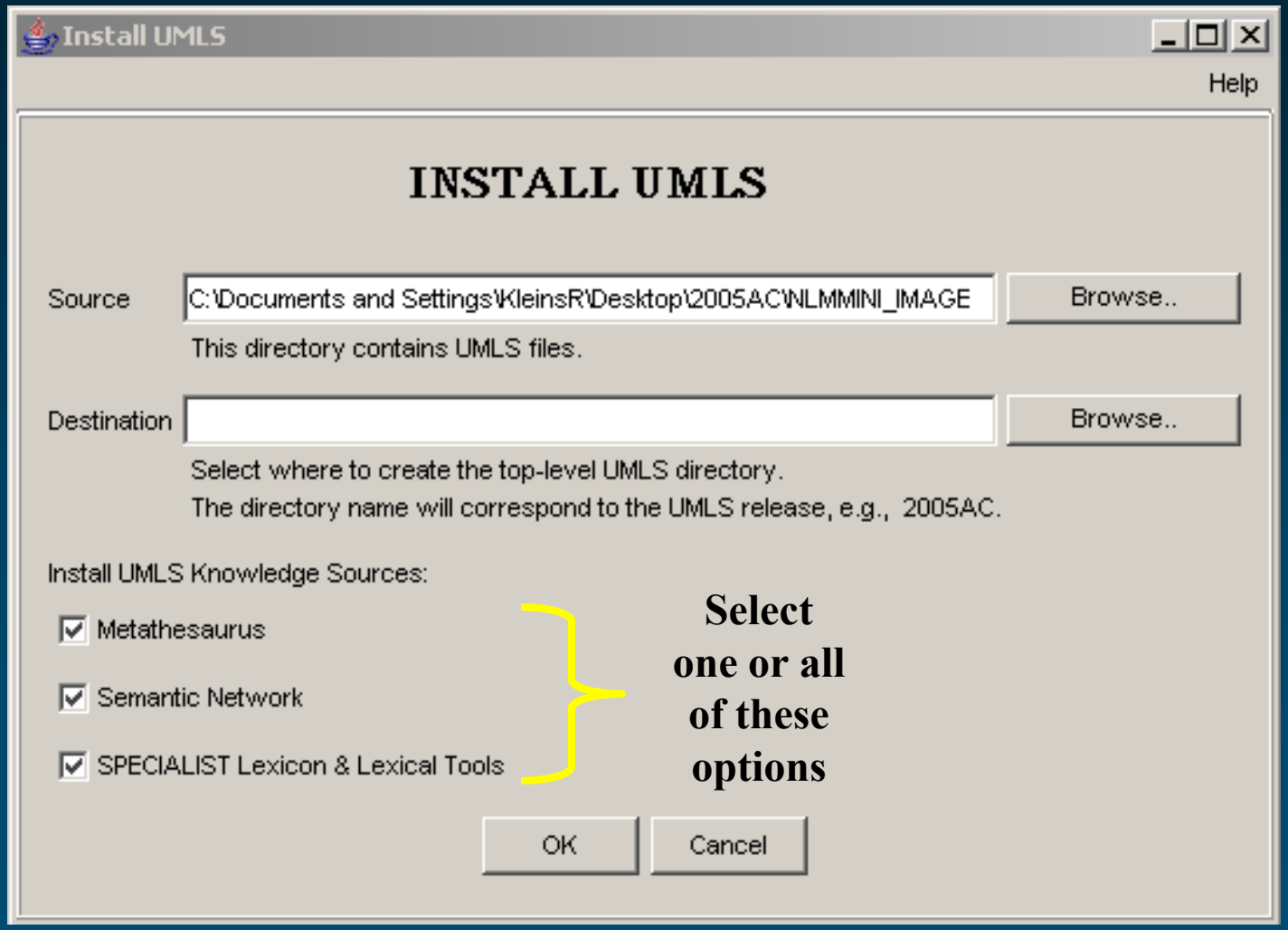
- Removes all information from all relational files from excluded vocabularies
  - atoms, strings, relations, hips, attributes, mappings
- Applies additional options selected by user
  - Changes to suppressibility or precedence
- Produces custom set of Metathesaurus relational files reflecting selected criteria
- Log file records subset details
- Output directory: set of Metathesaurus files

MetamorphoSys Welcome Screen

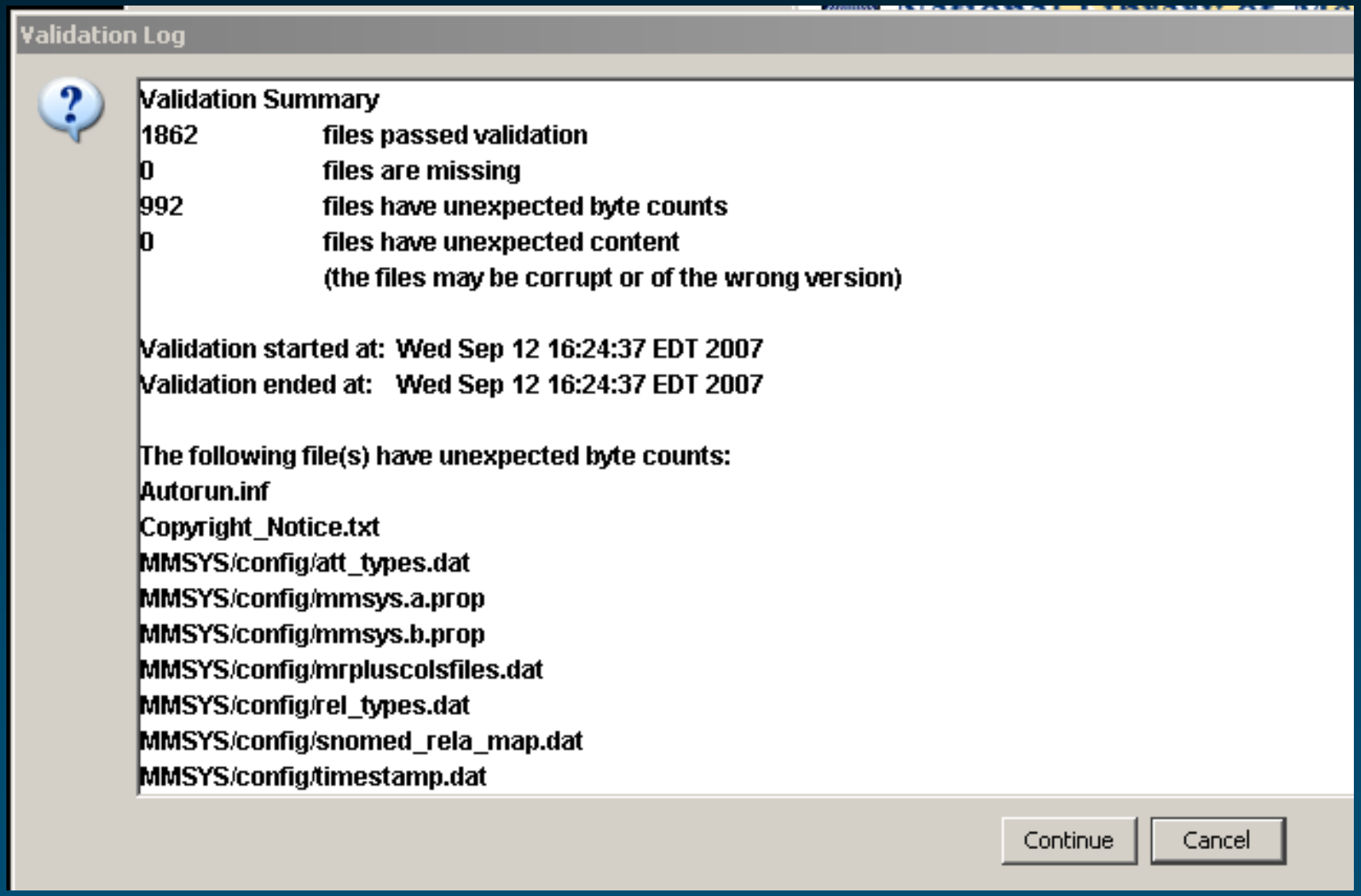
- Validate Distribution
  - Ensures that all files were downloaded
  - Process takes @ 30 minutes
  - Writes validation.log file
- Copy DVD to hard drive
  - Copies all files to local storage
  - Allows multiple people to use one DVD
  - May improve run time



Install UMLS



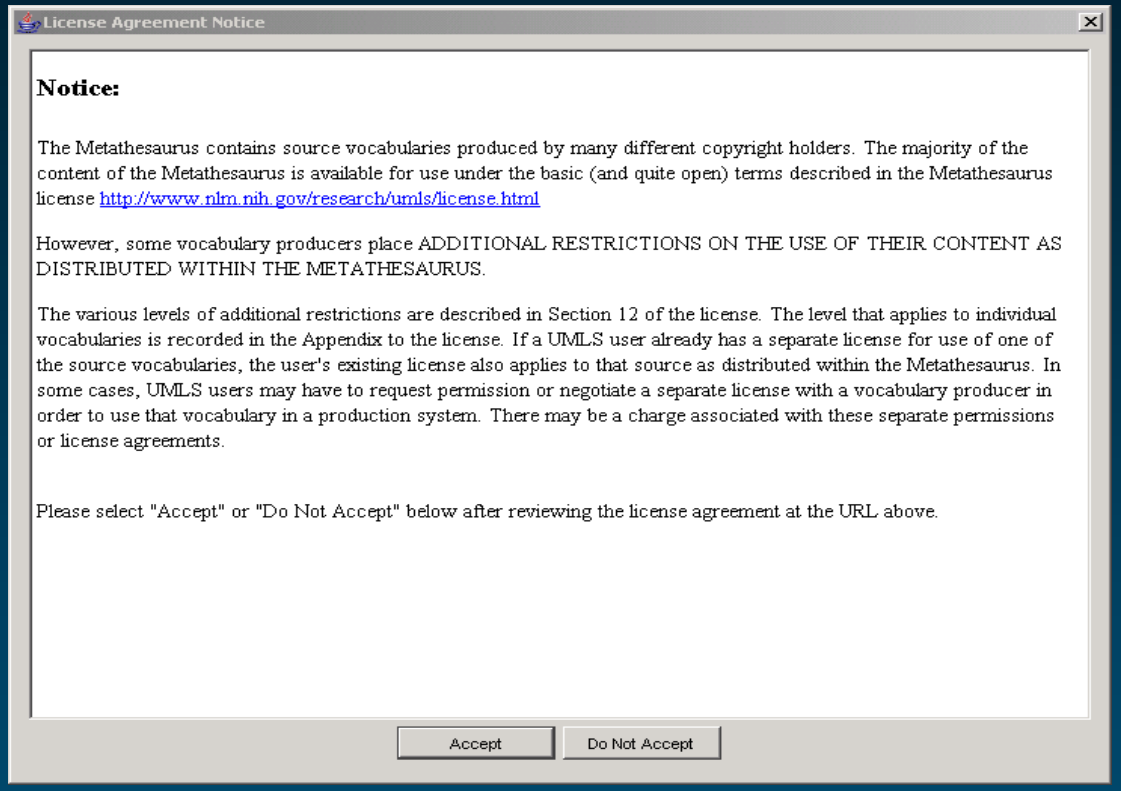
Validation Summary

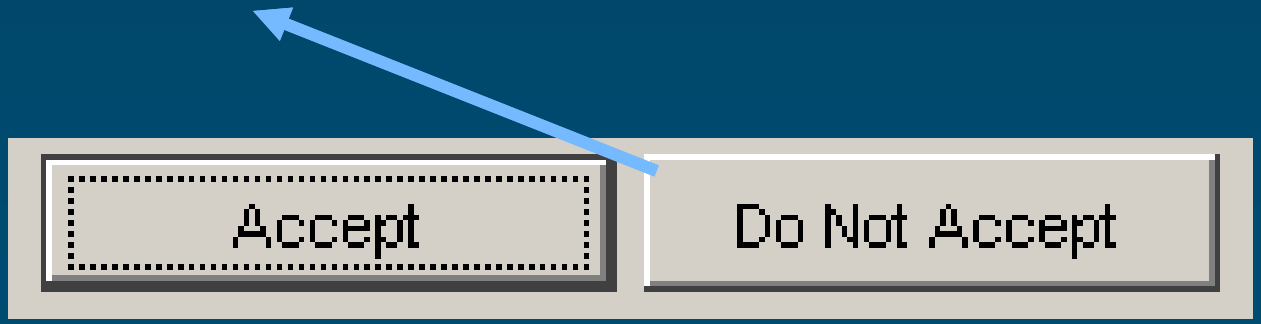


Configuration Files

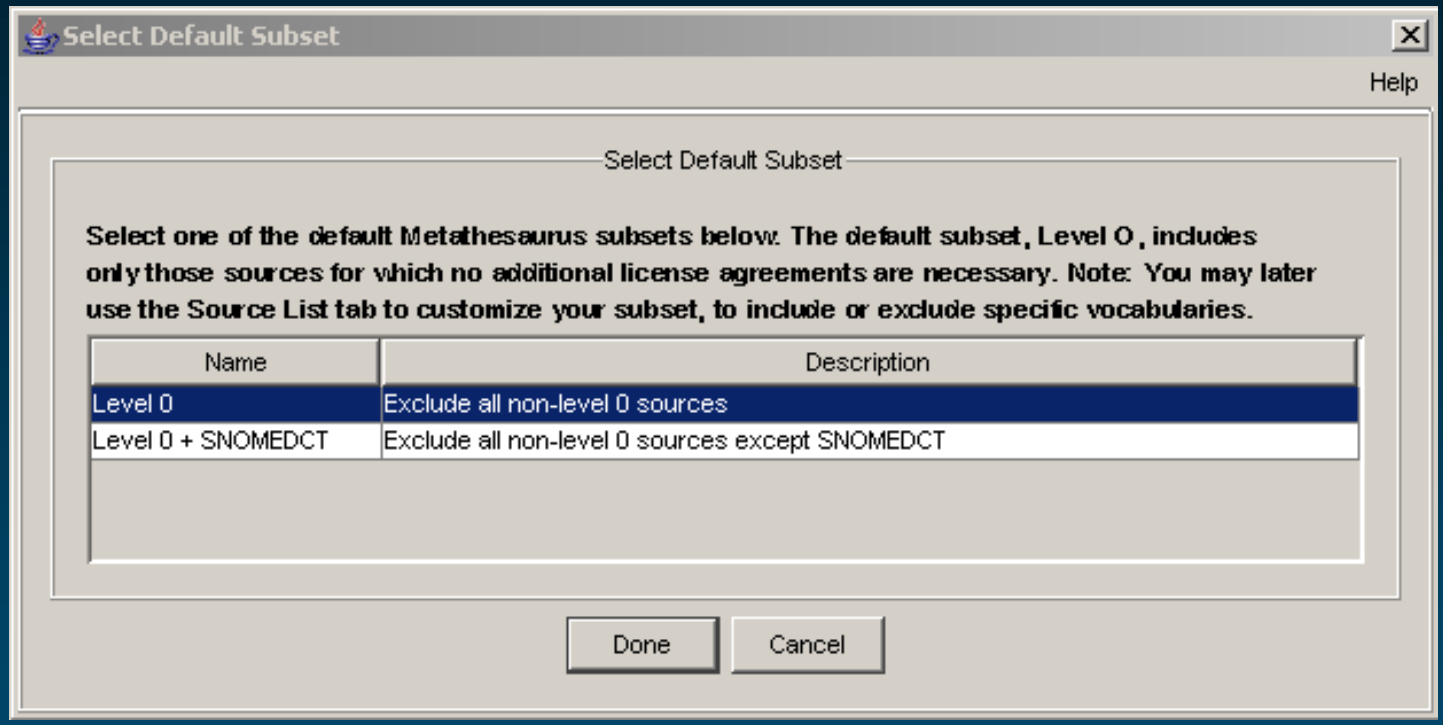
- New users must select “New Configuration”
- Returning users may select “Open Configuration” to open a previously saved MetamorphoSys Configuration
- This screen only appears if you install the Metathesaurus

UMLS License Notice





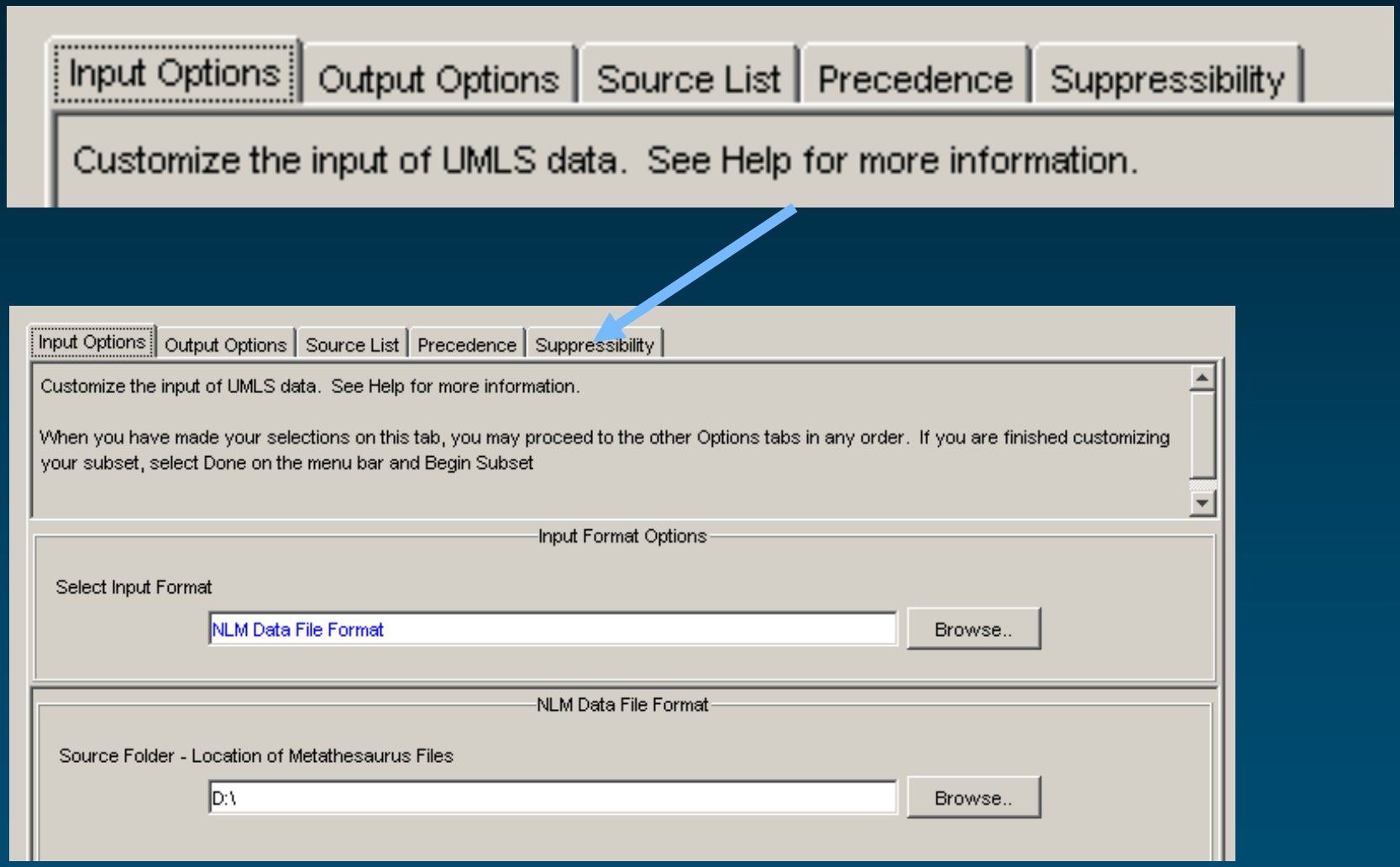
Select a default subset



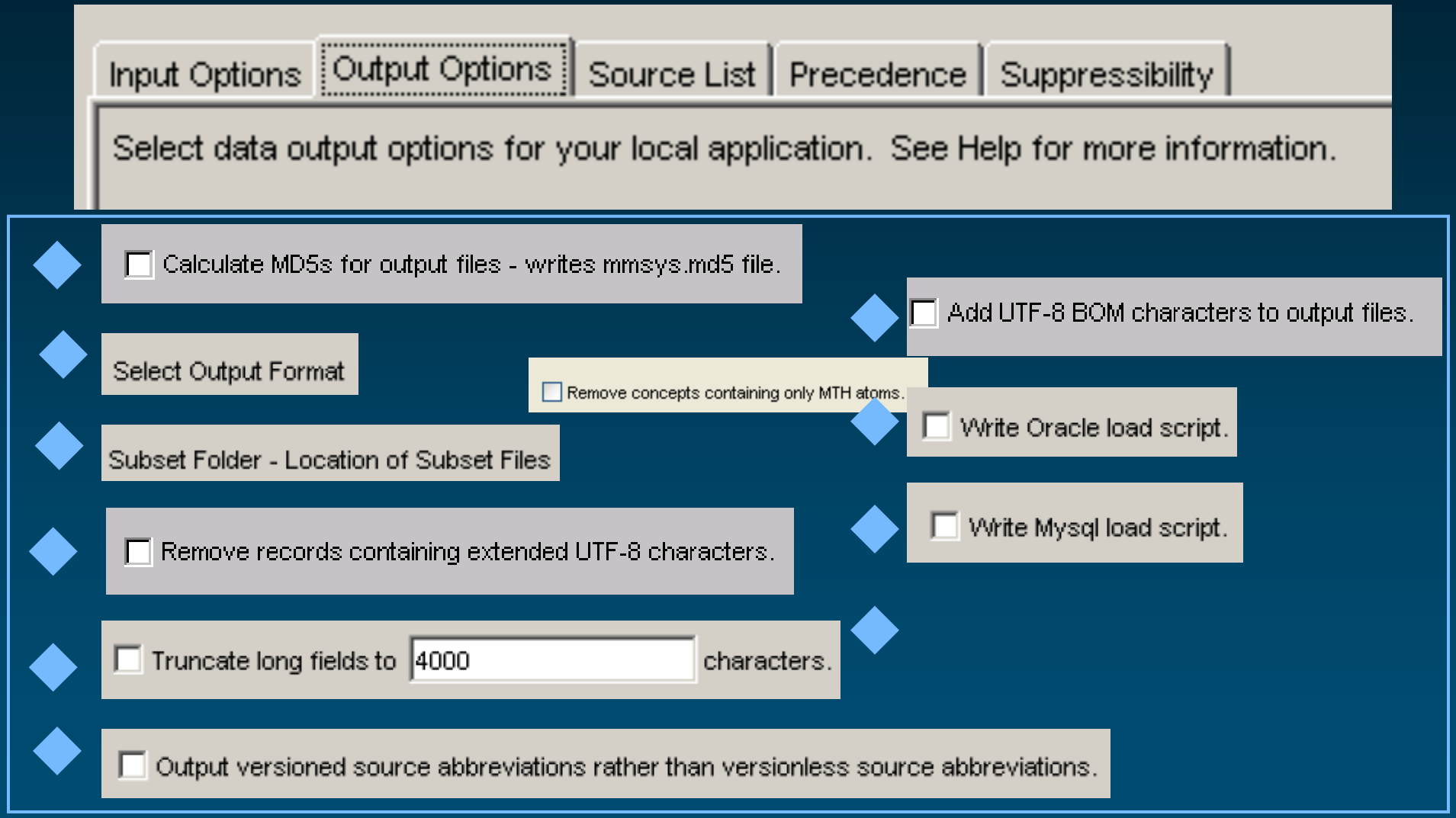
Level 0 → no separate additional license agreements

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

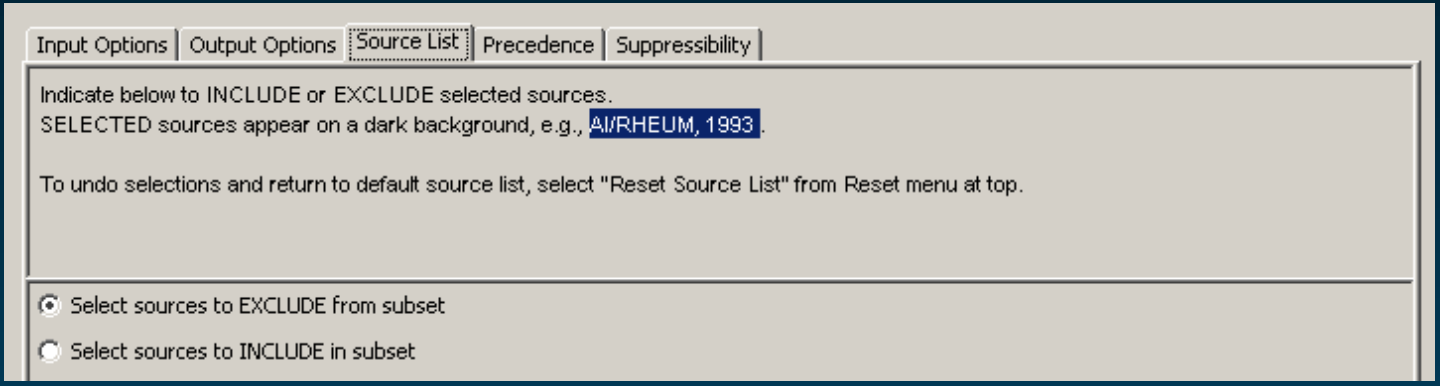
Input Options Tab



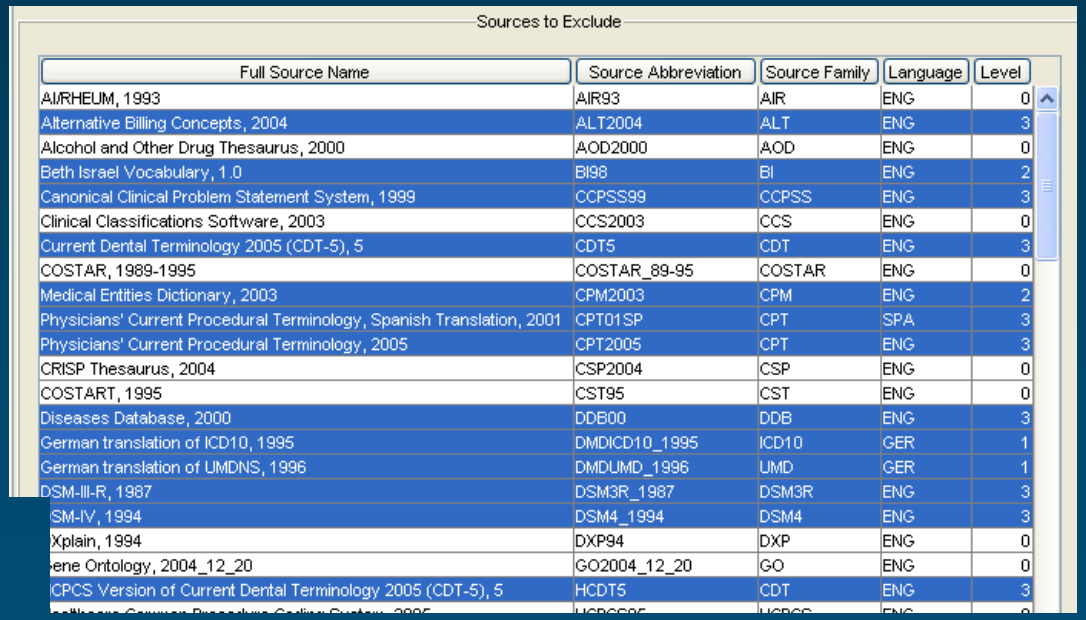
Output Options Tab



Source List Tab



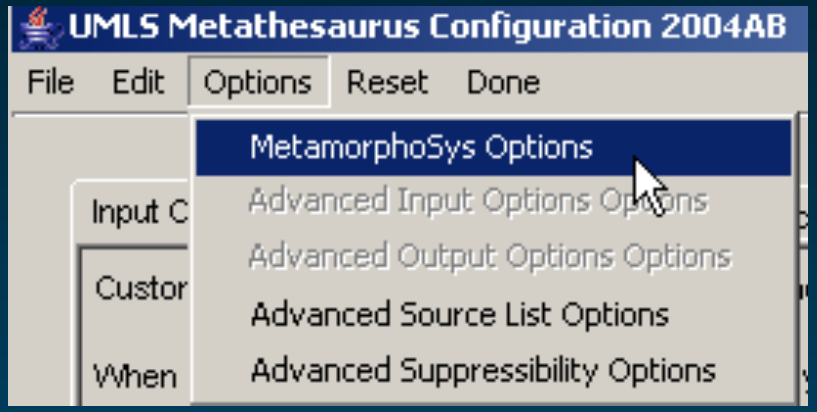
Exclude or Include



Full Source Name	Source Abbreviation	Source Family	Language	Level
AI/RHEUM, 1993	AIR93	AIR	ENG	0
Alternative Billing Concepts, 2004	ALT2004	ALT	ENG	3
Alcohol and Other Drug Thesaurus, 2000	AOD2000	AOD	ENG	0
Beth Israel Vocabulary, 1.0	BI98	BI	ENG	2
Canonical Clinical Problem Statement System, 1999	CCPSS99	CCPSS	ENG	3
Clinical Classifications Software, 2003	CCS2003	CCS	ENG	0
Current Dental Terminology 2005 (CDT-5), 5	CDT5	CDT	ENG	3
COSTAR, 1989-1995	COSTAR_89-95	COSTAR	ENG	0
Medical Entities Dictionary, 2003	CPM2003	CPM	ENG	2
Physicians' Current Procedural Terminology, Spanish Translation, 2001	CPT01SP	CPT	SPA	3
Physicians' Current Procedural Terminology, 2005	CPT2005	CPT	ENG	3
CRISP Thesaurus, 2004	CSP2004	CSP	ENG	0
COSTART, 1995	CST95	CST	ENG	0
Diseases Database, 2000	DDB00	DDB	ENG	3
German translation of ICD10, 1995	DMDICD10_1995	ICD10	GER	1
German translation of UMDNS, 1996	DMDUMD_1996	UMD	GER	1
DSM-III-R, 1987	DSM3R_1987	DSM3R	ENG	3
SM-IV, 1994	DSM4_1994	DSM4	ENG	3
Xplain, 1994	DXP94	DXP	ENG	0
ene Ontology, 2004_12_20	GO2004_12_20	GO	ENG	0
CPCS Version of Current Dental Terminology 2005 (CDT-5), 5	HCDT5	CDT	ENG	3

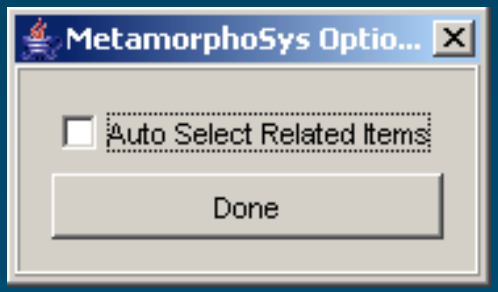
Highlighted rows are excluded from the subset.

MetamorphoSys Option Tab

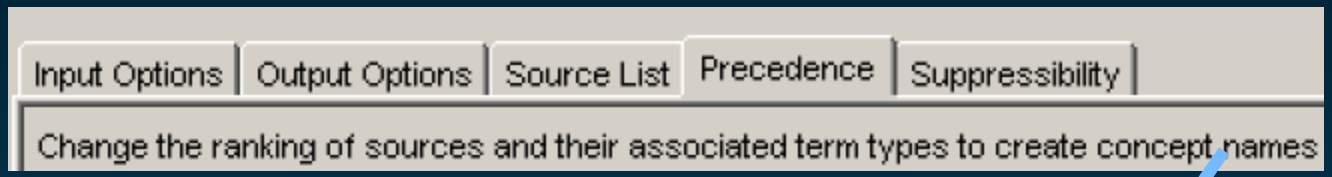


Source list behavior can be changed using the MetamorphoSys Option Tab

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

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

Change the ranking of sources and their associated term types to create concept names that are more useful in your local application.
Concept names are determined by the term with highest ranking source/term type.

To move rows, either cut and paste rows, or drag and drop.

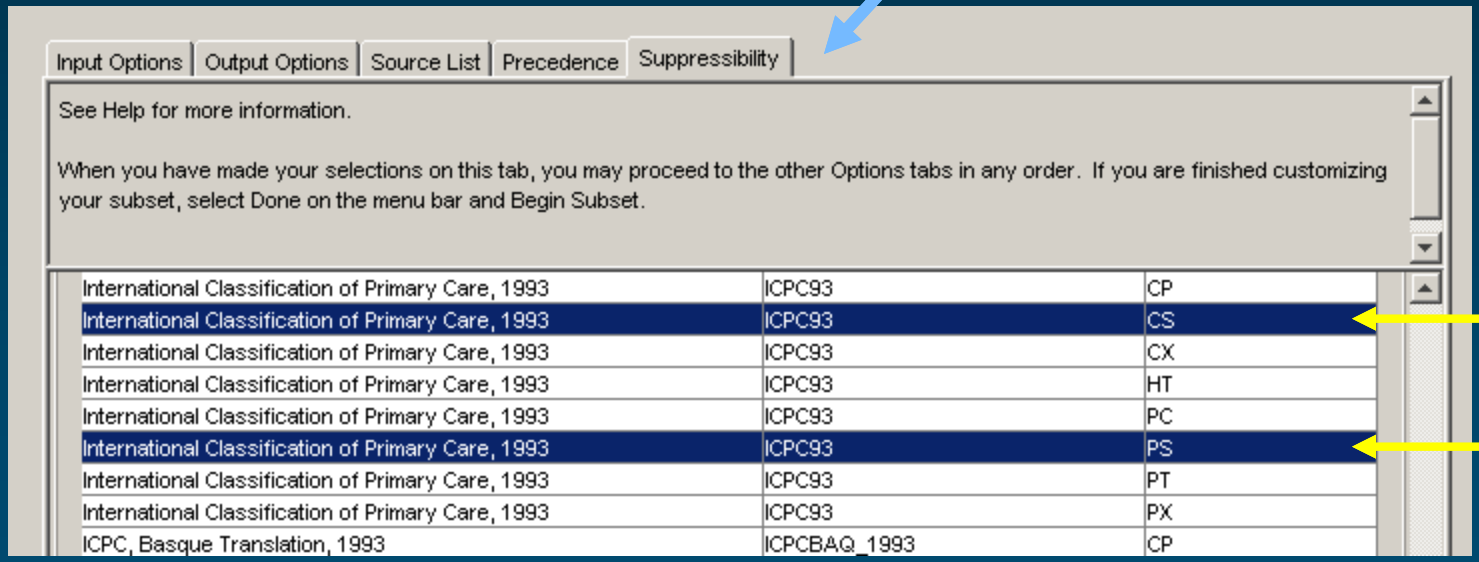
When you have made your selections on this tab, you may proceed to the other Options tabs in any order. If you are finished customizing

Precedence

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

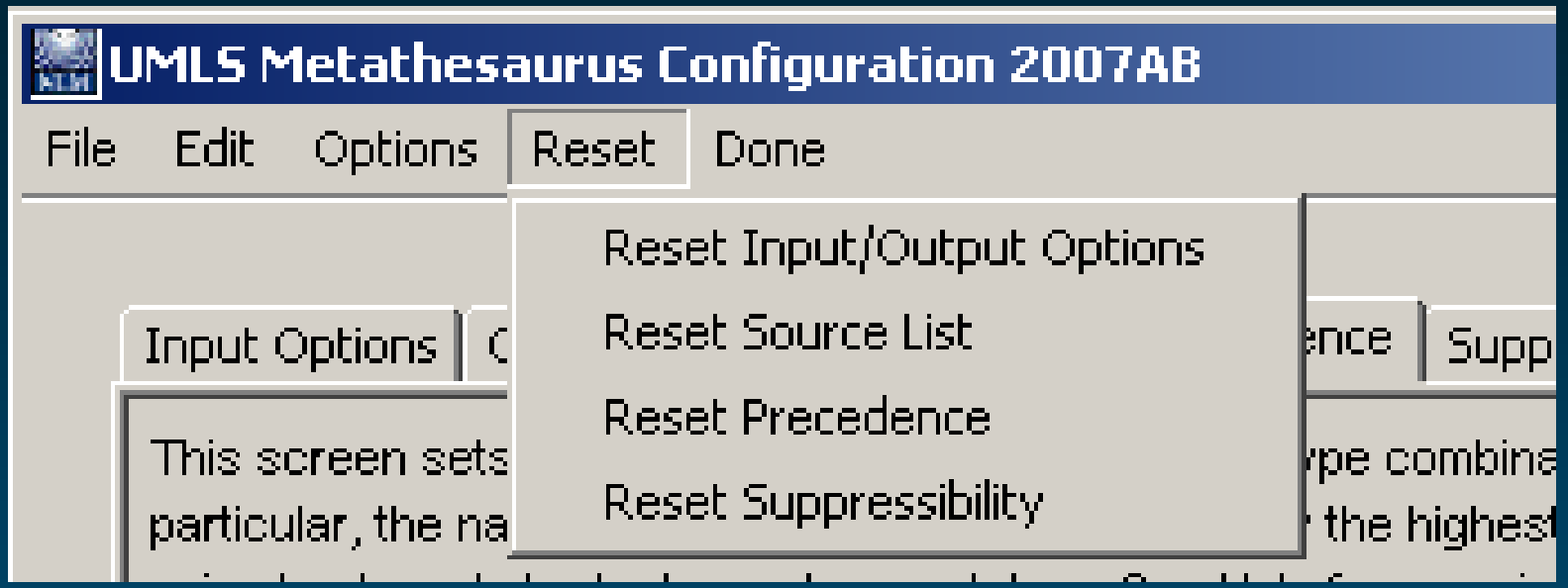
Cut and paste rows to alter the preferred name

Suppressibility Tab



Highlighted source term types will be marked suppressible

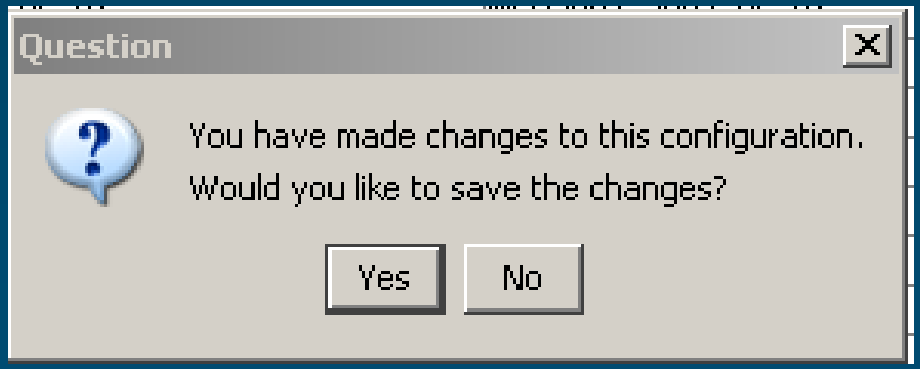
Reset menu



- 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

- Complete configuration options
- Done menu
- Begin Subset



MetamorphoSys log

mmsys.log - Notepad

File Edit Format View Help

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

MetamorphoSys log

mmsys.log - Notepad

File Edit Format View Help

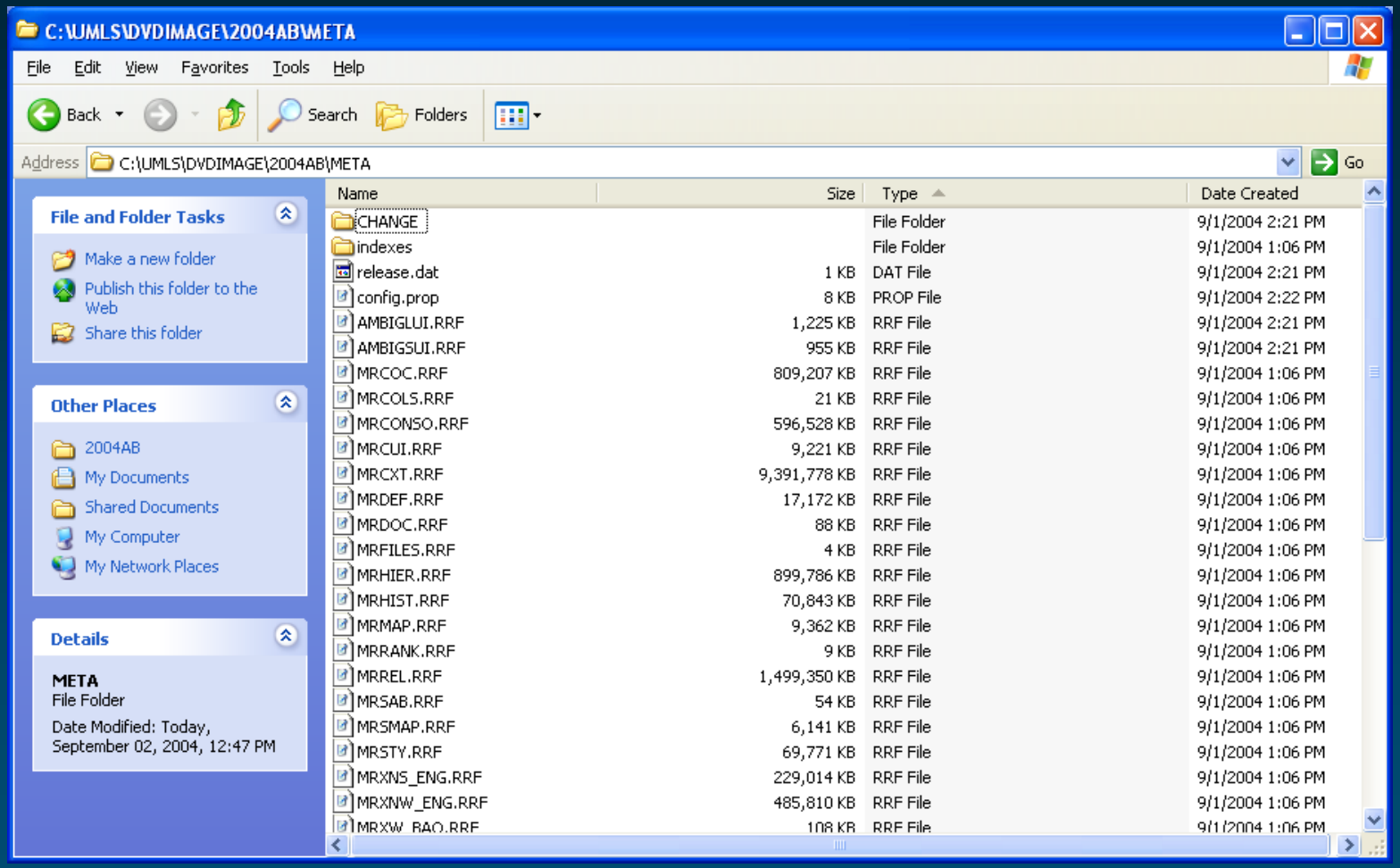
```
--------------------------------
Metathesaurus Output: Rich Release Format
Long fields were not truncated.
Source Abbreviations were written out with a versionless (root) representation.
Fields containing UTF-8 characters were not removed.


Excluded Sources
  <none>

Kept Sources
  AI/RHEUM, 1993                                                  AIR93
  Alternative Billing Concepts                                    ALT2003
  Alcohol and Other Drug Thesaurus, 2000                          AOD2000
  Beth Israel Vocabulary, 1.0                                     BI98
  Canonical Clinical Problem Statement System, 1999               CCPSS99
  Clinical Classifications Software, 2003                         CCS2003
  Current Dental Terminology (CDT), 4                             CDT4
  COSTAR, 1989-1995                                               COSTAR_89-95
  Medical Entities Dictionary, 2003                               CPM2003
  Physicians' Current Procedural Terminology, Spanish Translation,... CPT01SP
  Physicians' Current Procedural Terminology, 2004                CPT2004
  CRISP Thesaurus, 2004                                           CSP2004
  COSTART, 1995                                                   CST95
  Diseases Database, 2000                                         DDB00
  German translation of ICD10, 1995                               DMDICD10_1995
  German translation of UMDNS, 1996                               DMDUMD_1996
  DSM-III-R, 1987                                                 DSM3R_1987
  DSM-IV, 1994                                                    DSM4_1994
  DXplain, 1994                                                   DXP94
  Gene Ontology, 2004_03_02                                       GO2004_03_02
  HCPCS Version of Current Dental Terminology (CDT), 4            HCDT4
  HCFA Common Procedure Coding System, 2004                       HCPCS04
  HCPCS Version of Current Procedural Terminology (CPT), 2004     HCPT04
  Health Devices Alerts, 1999                                     HDA99
  Home Health Care Classification, 2003                           HHC2003
  Health Level Seven Vocabulary, 1998-2002                        HL7_1998-2002
  ICPC2E-ICD10 relationships from Dr. Henk Lamberts, 1998         HLREL_1998
  Health Product Comparison System, 1999                          HPC99
  ICD10, American English Equivalents, 1998                       ICD10AE_1998
  International Statistical Classification of Diseases and Related... ICD10AMAE_2000
```

Kept Sources

Output directory contents

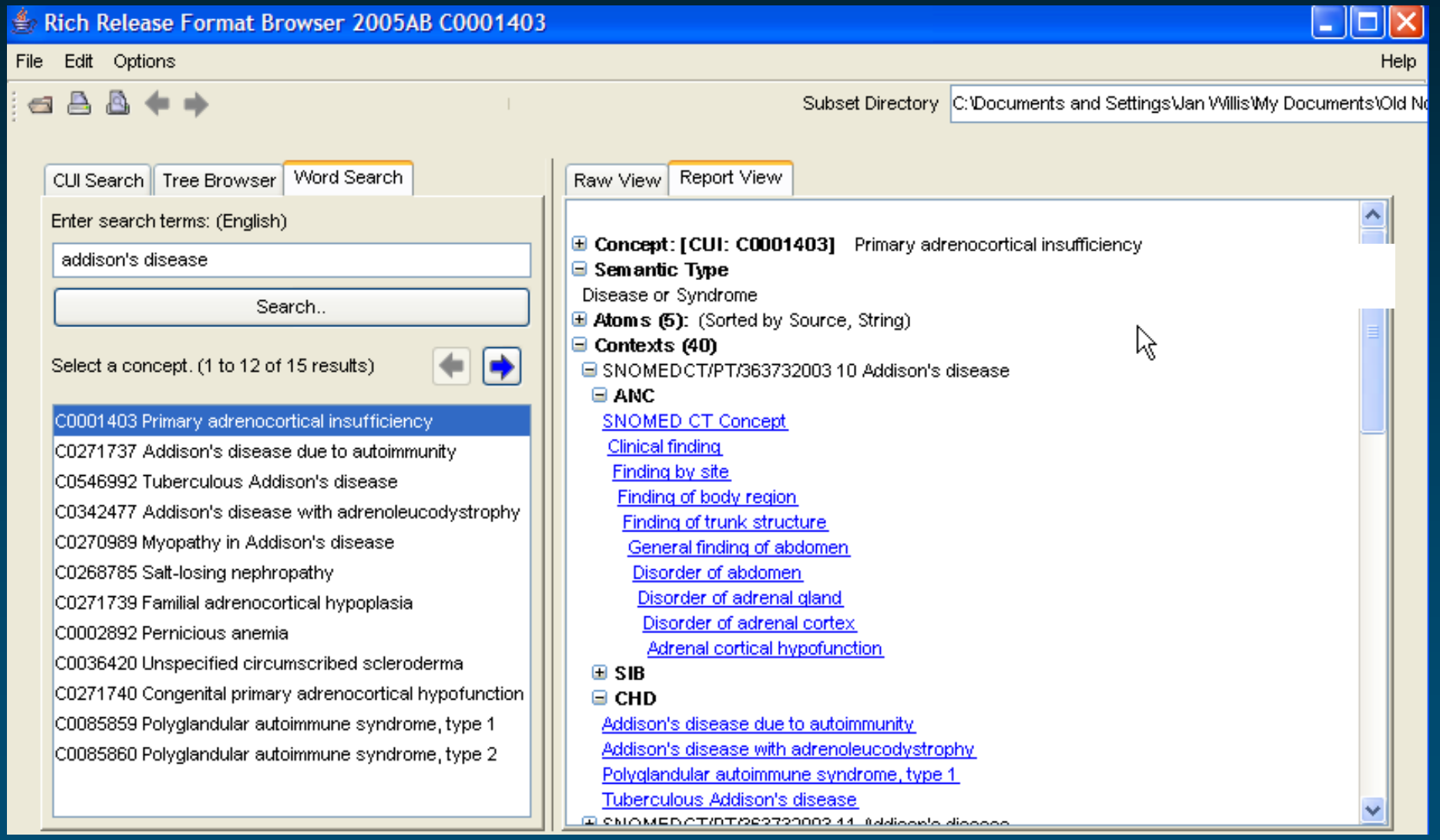


RRF Subset Browser

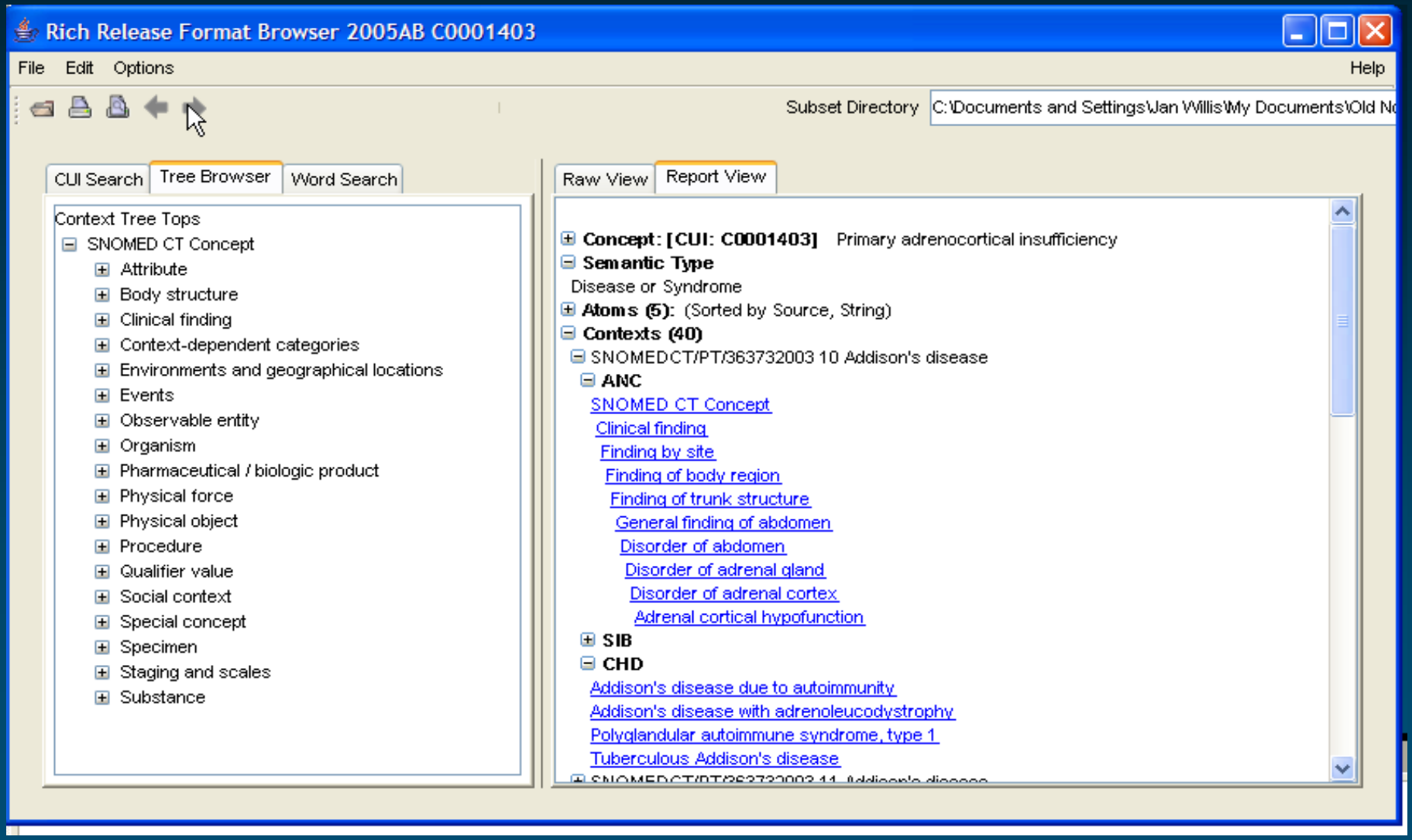
- Must have an RRF Subset
- View customized subsets
  - Tree, report, or raw data for each term
  - Expandable Concept Report View
  - Search for CUI or more information
- Quickly find terms within a subset
- Hyperlinked concepts

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

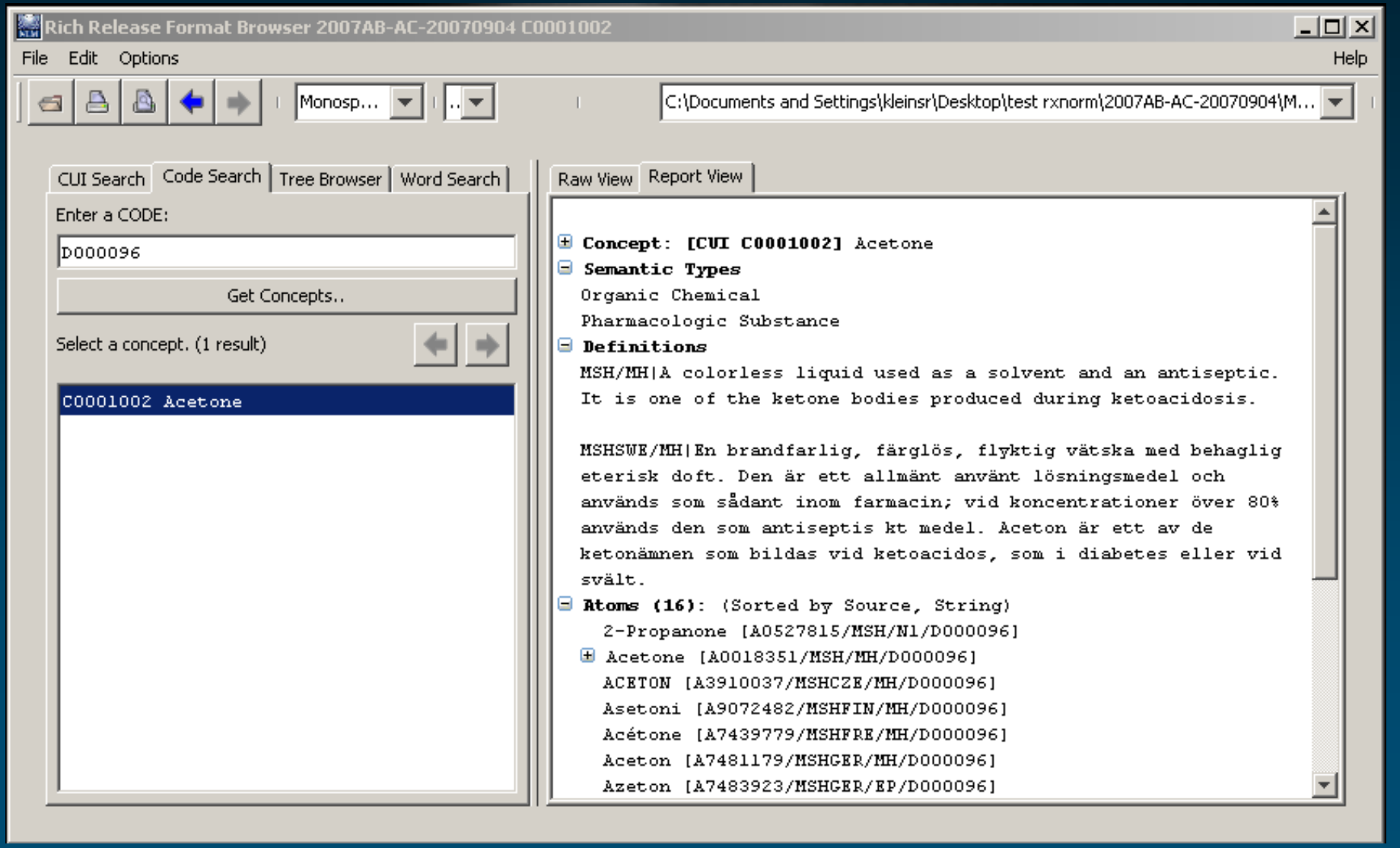
RRF Browser word search & report view



RRF Browser tree browser & report view

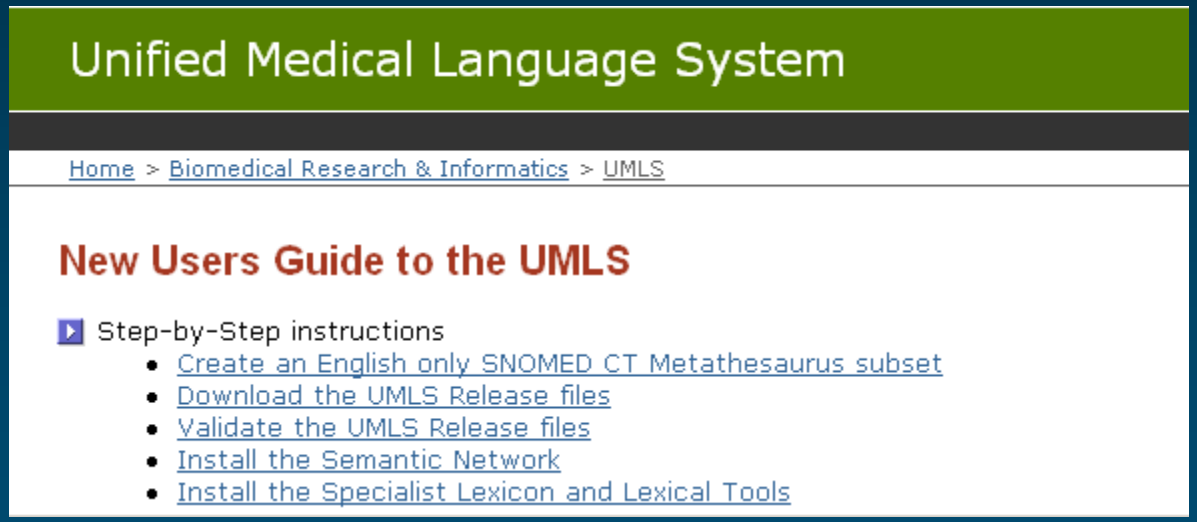


RRF Browser -- search by code



MetamorphoSys Step-by-Step help

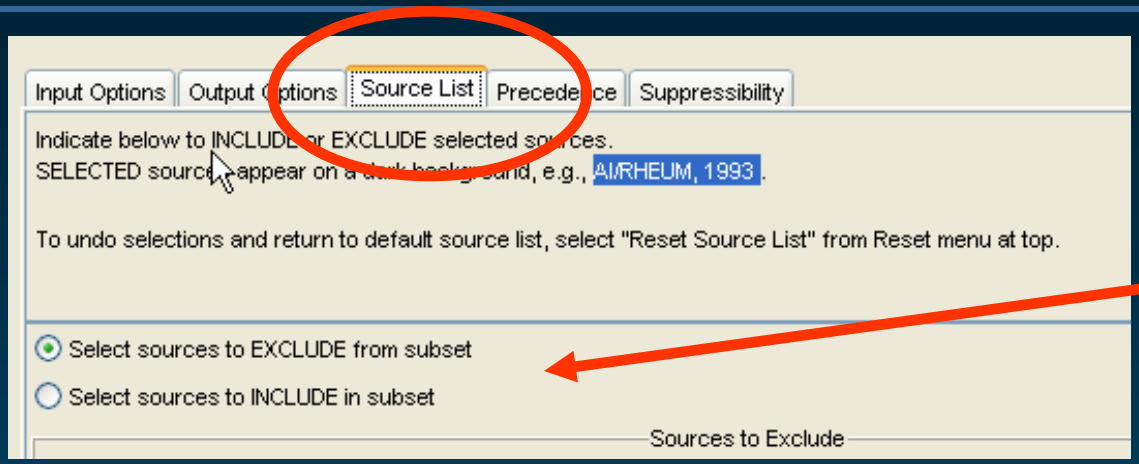
- MS Word files
  - Screen grabs with explaination
  - Cover common MMSYS tasks such as installing the UMLS Knowledge Sources and creating a custom subset
  - Suggestions for new Step-by-Steps requested!



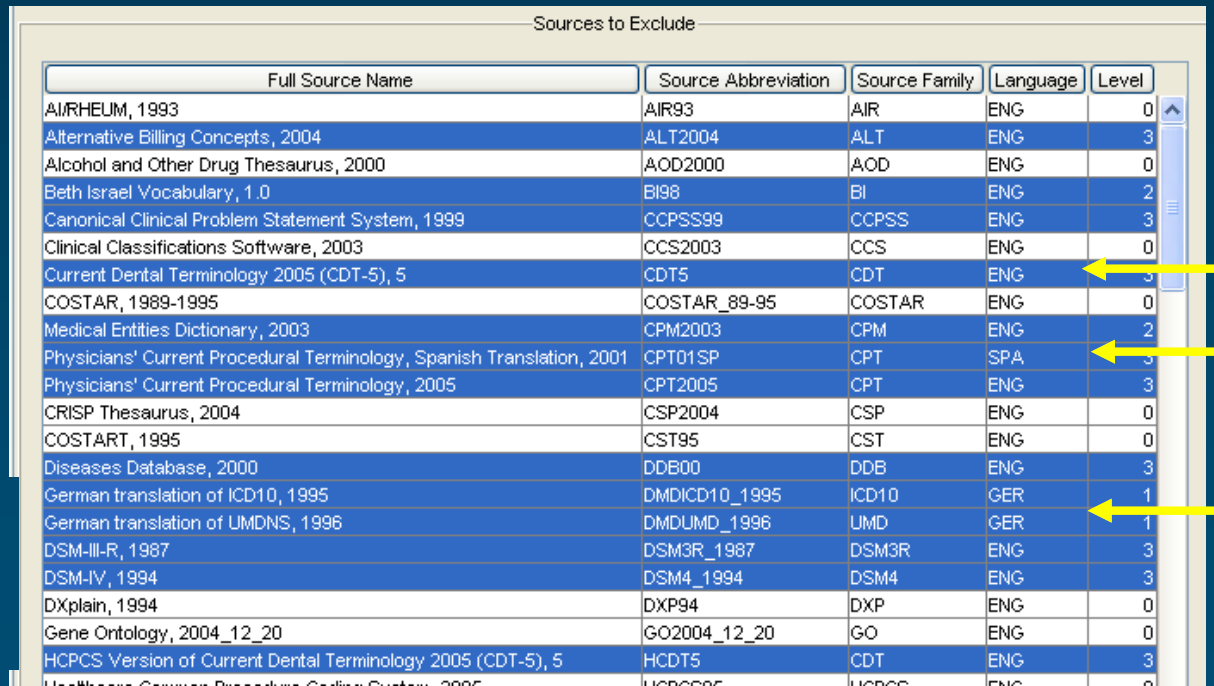
⑩http://www.nlm.nih.gov/research/umls/new_users.html

How do I?

Specify sources for a customized subset?



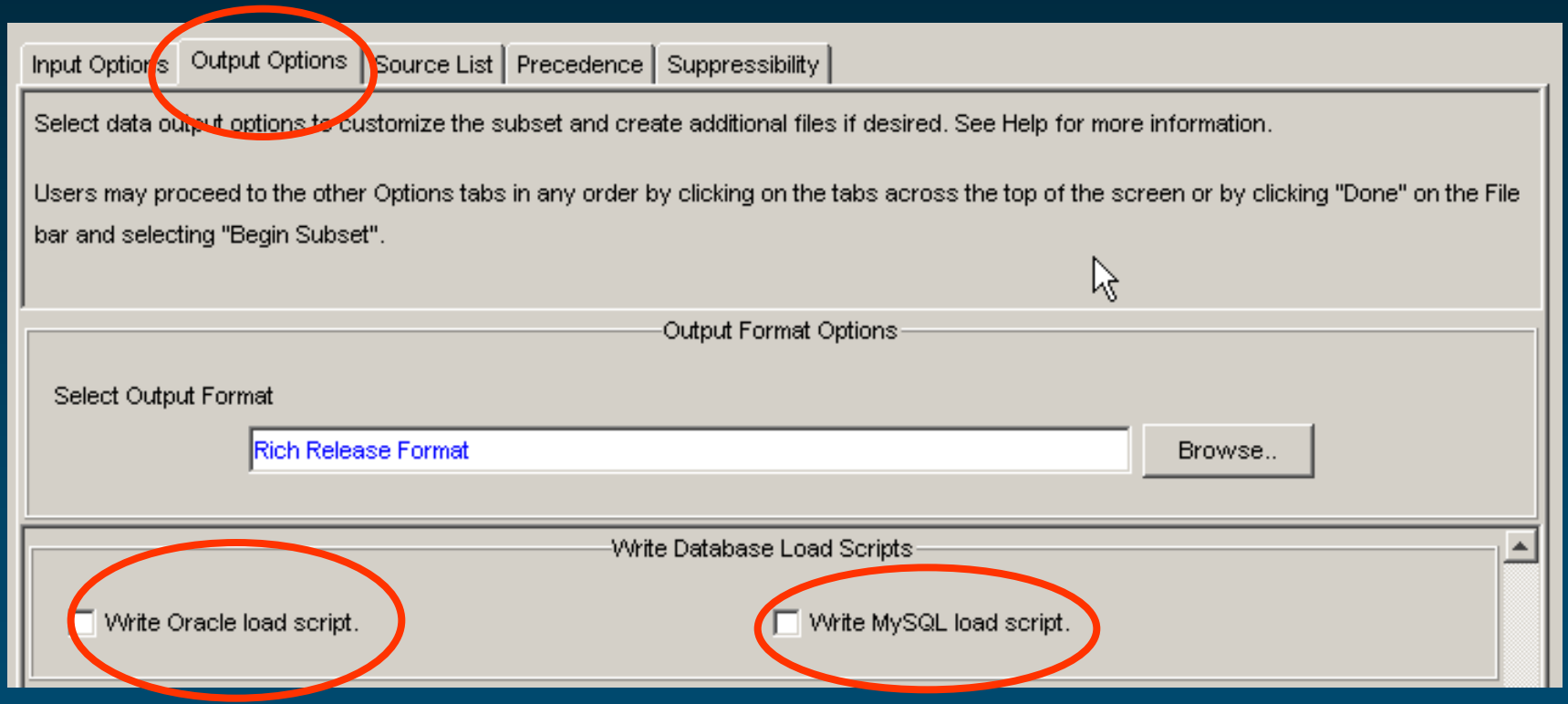
⑩ Exclude or Include



⑩ Highlighted rows are excluded from the subset.

Create a custom database load script?

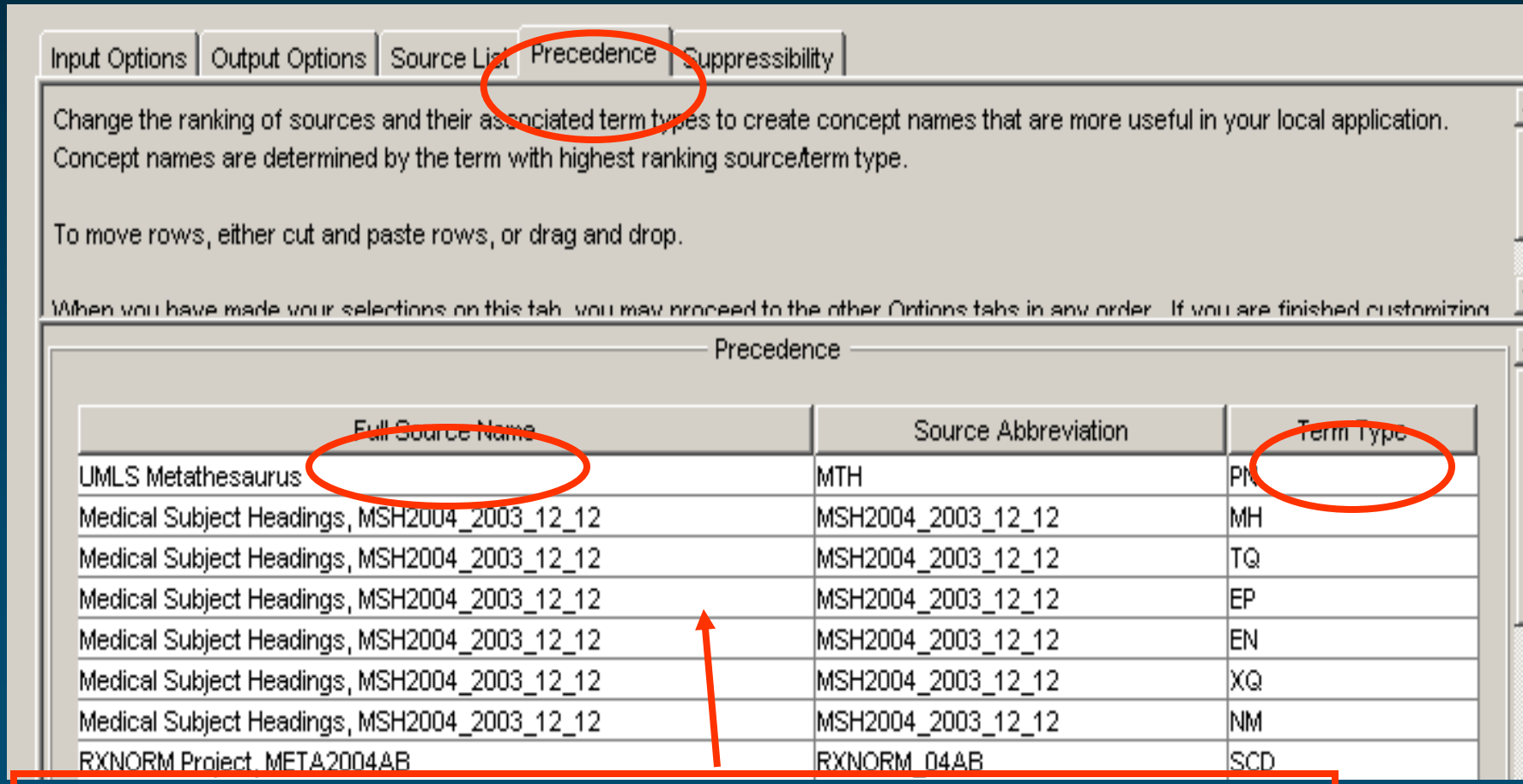
- Select the Output Options tab
- Check the box next to the type of load script you require



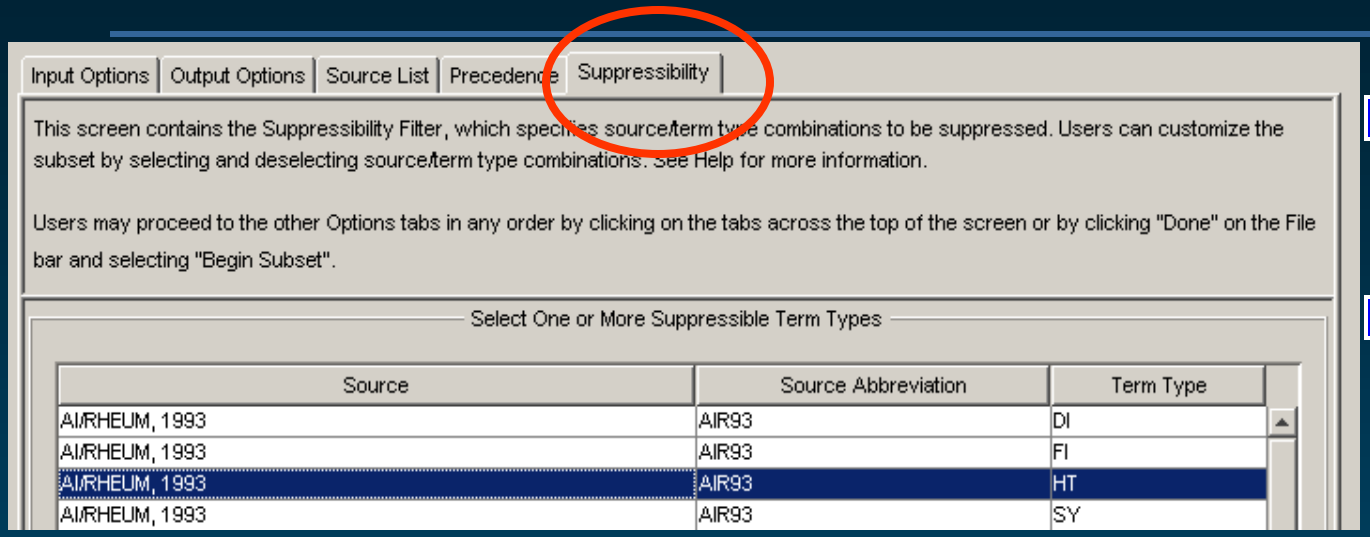
- Other Options on this tab

Change how preferred term is set?

- Select the Precedence tab
- Cut and paste or drag and drop source and term types to reflect your preferred ranking order

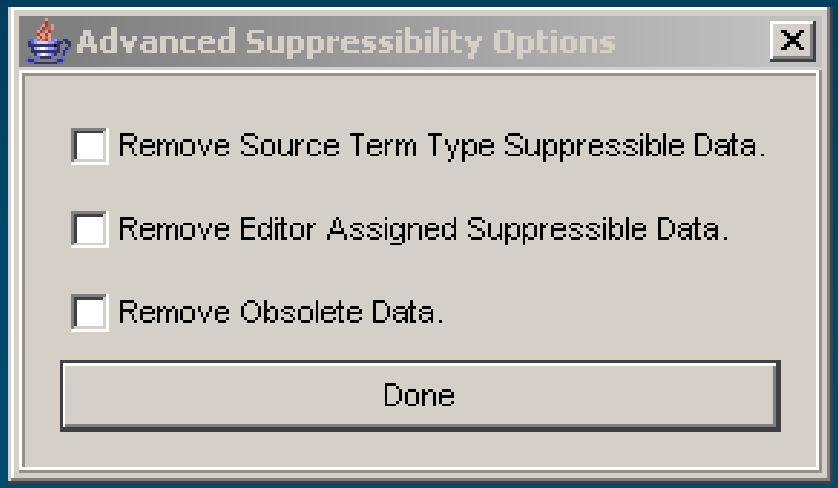


Remove specific term types from subset?

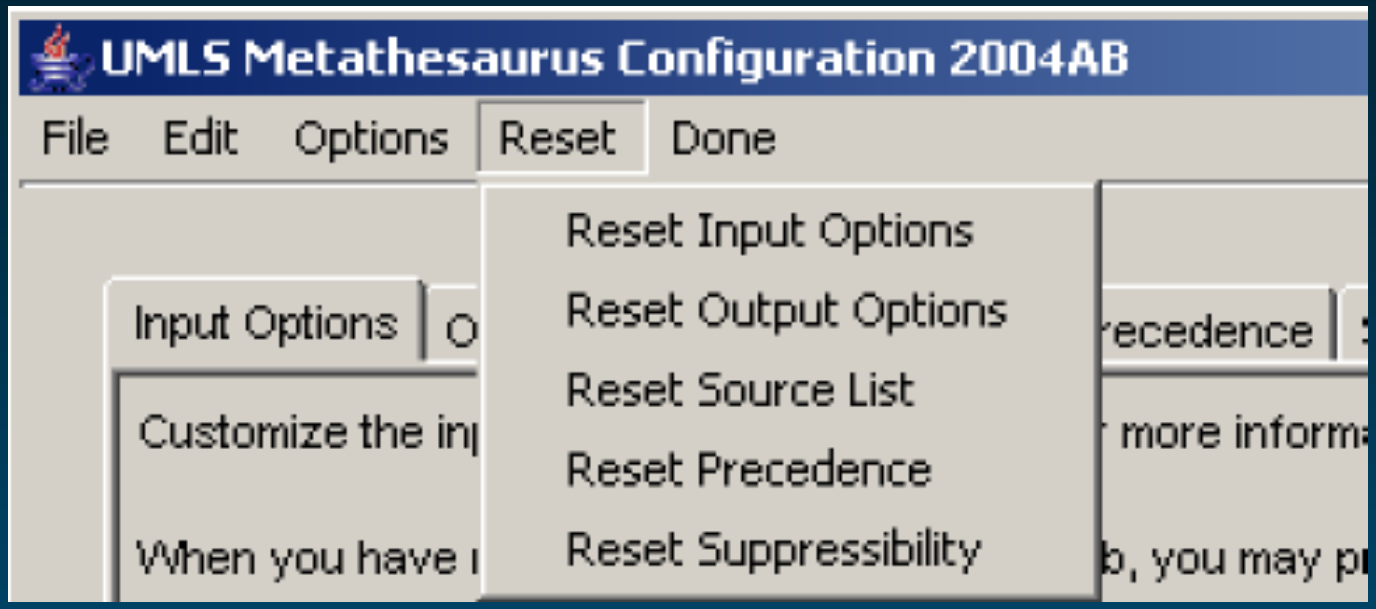


- Select Suppressibility tab
- Highlight rows to exclude

- Select Options → Advanced Suppressibility Options
- Select the term types to remove



Reset default MMSYS Options?



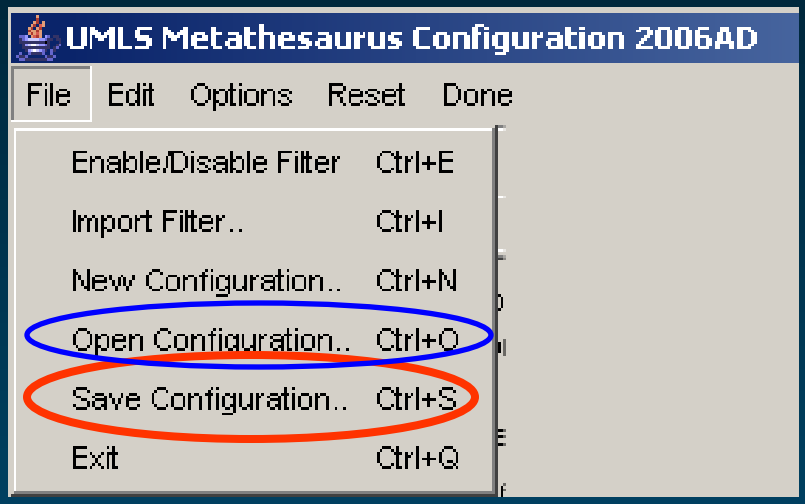
- Returns all filters to default selections
- Default selections in “mmsys.prop.default file” in config folder
- mmsys.prop.default contains properties in last run

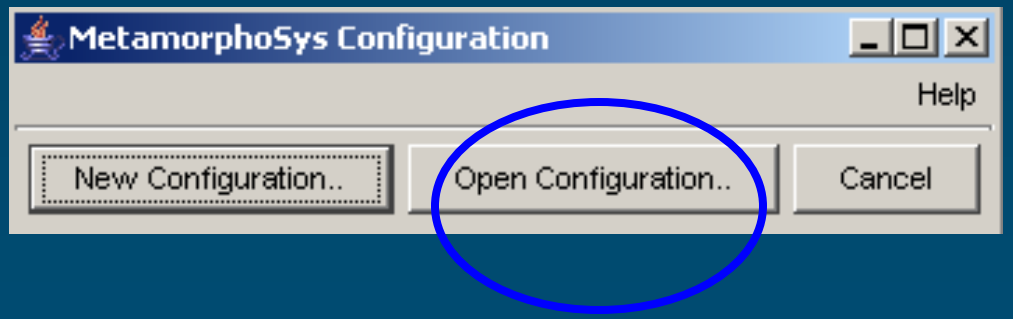
Ensure all team members have the same subset?

- On File menu "Save Configuration"
- Share configuration file with team members
- Have team members select "Open Configuration" from File Menu

/or/

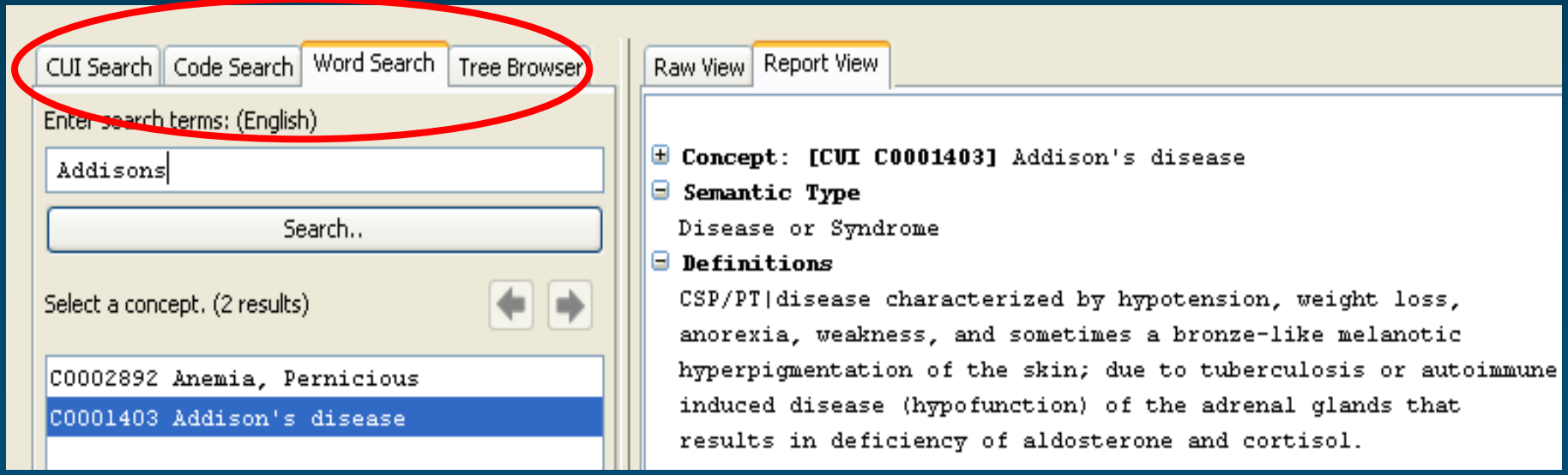
Select "Open Configuration" from Configuration Screen





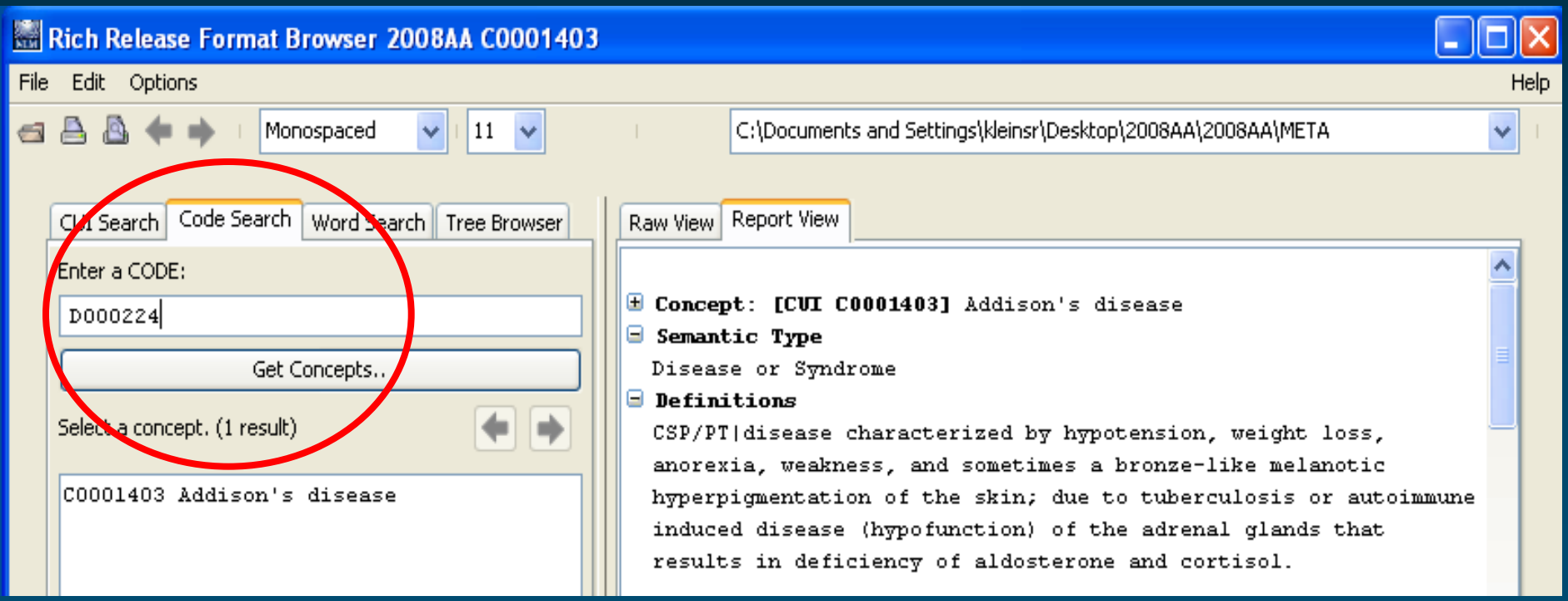
Search for a term in my RRF subset?

- Select "Browse my Subset" from welcome screen
- Browse to your subset location
- Search by term, string or CUI
- Reports include:
  - Hyperlinked concepts
  - Raw data view
  - Attributes and Relations



Search for a code in my RRF subset?

- Select "Browse my Subset" from welcome screen
- Browse to your subset location
- Select "Code search"



More information and help

- MetamorphoSys Documentation

http://www.nlm.nih.gov/research/umls/meta6.html

- Readme file on the DVD or downloaded from the UMLSKS

- Help Menu from any page in MetamorphoSys

UMLS Tools

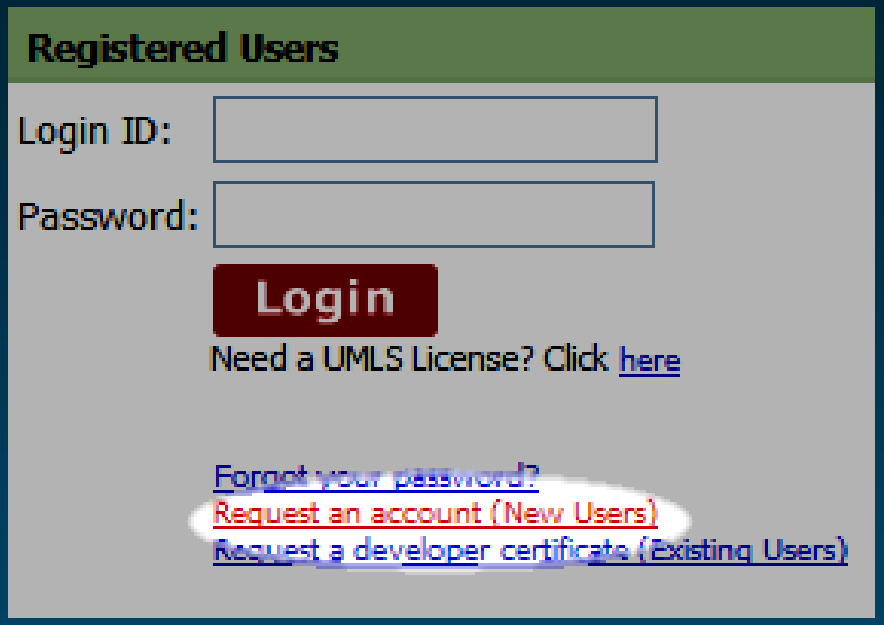
UMLS Knowledge Source Server

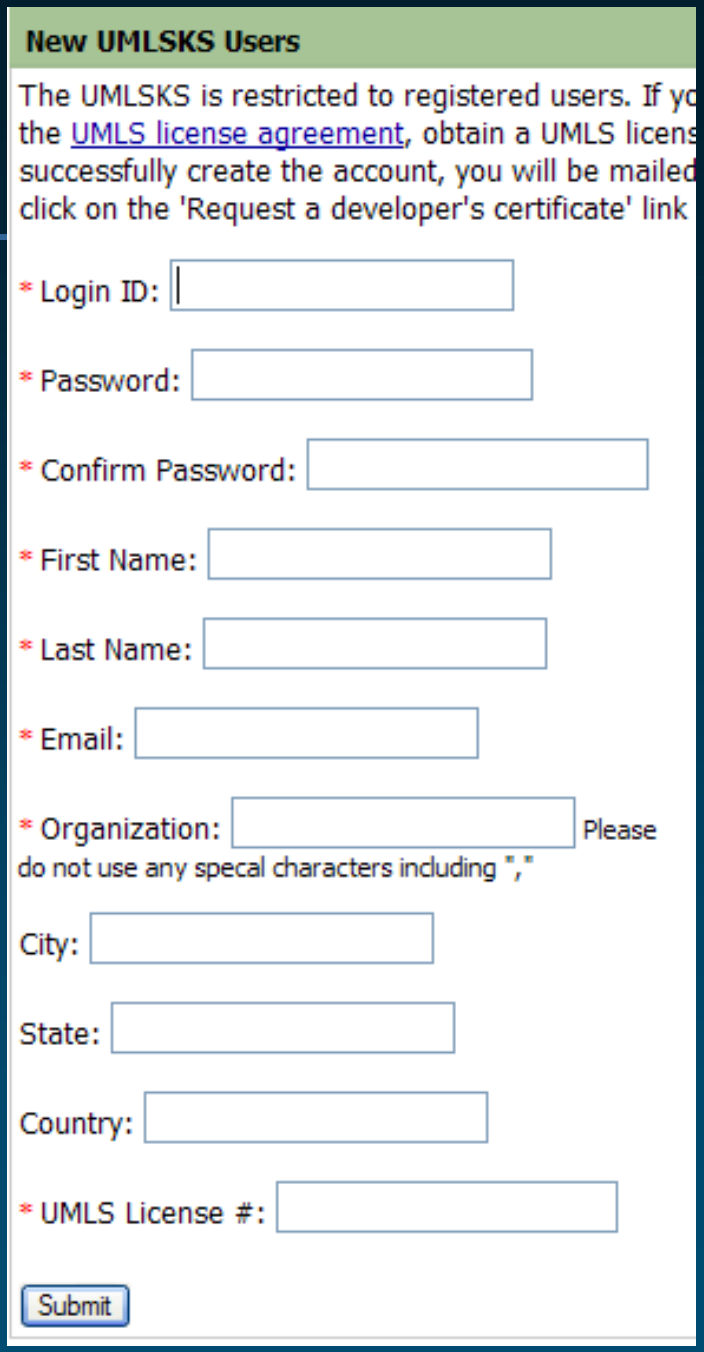
- Licensed users access online:

  http://umlsks.nlm.nih.gov

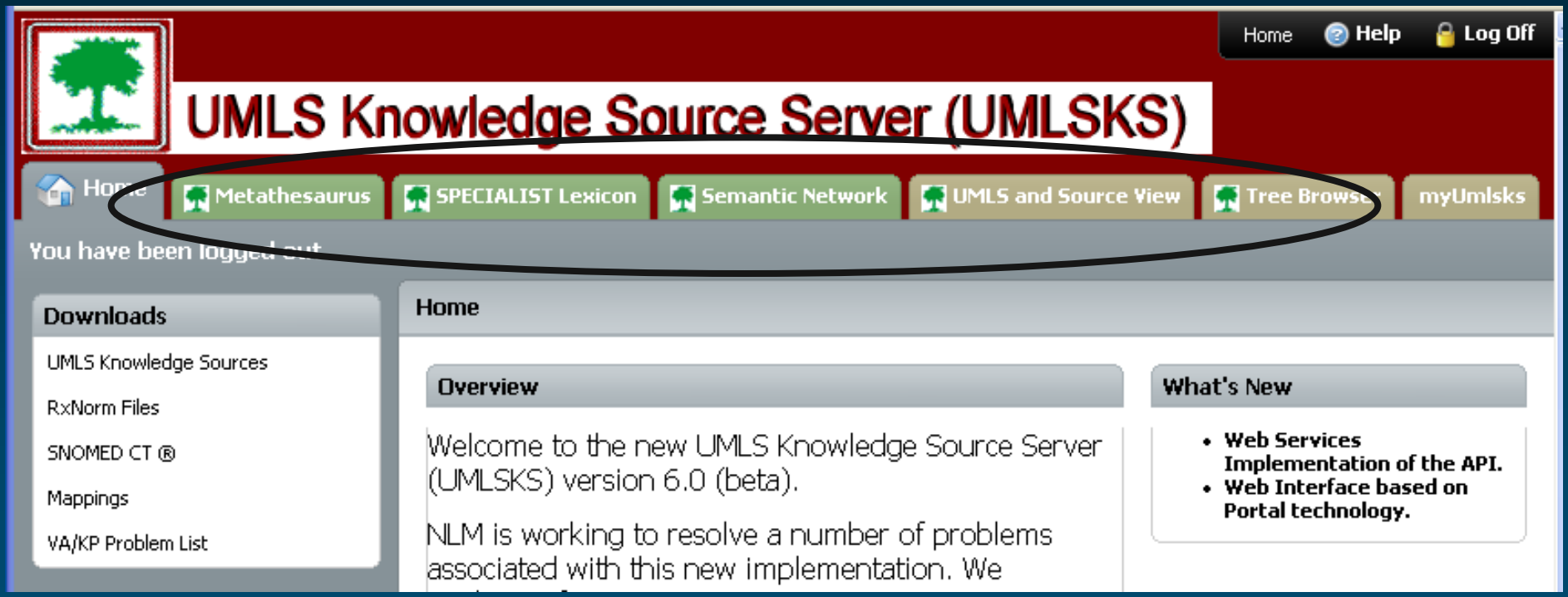
  Web search interface for the three Knowledge Sources (Metathesaurus, Semantic Network, Specialist Lexicon)
- Application Programming Interface (API)
- File downloads
- Documentation

Create a UMLSKS Account

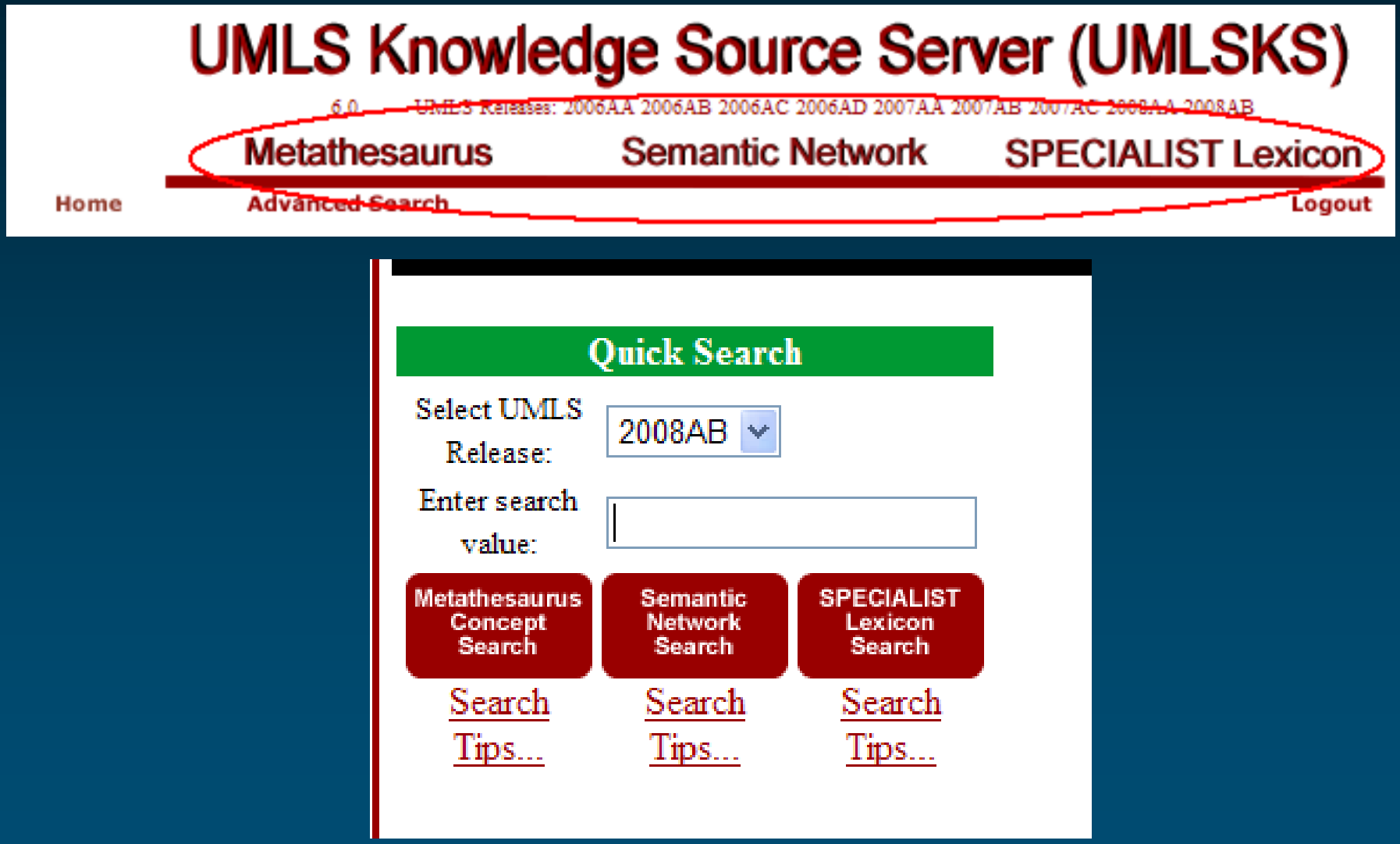




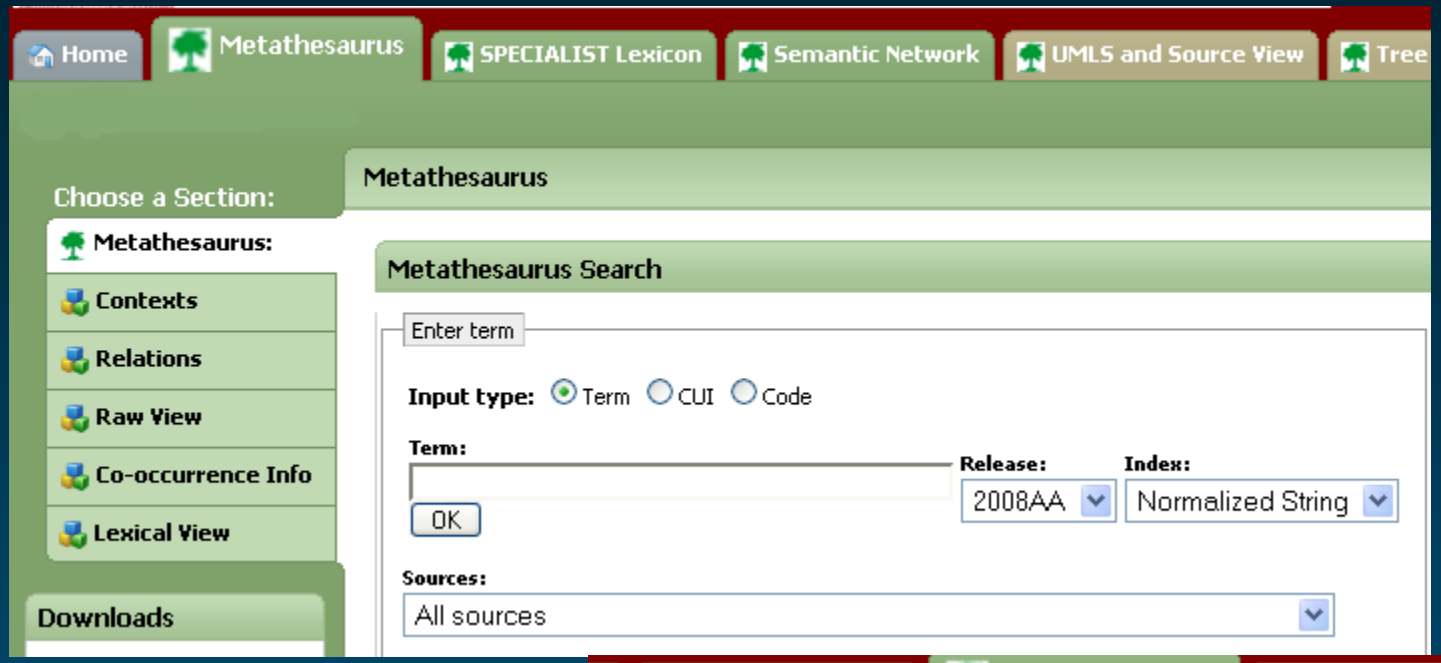
UMLSKS Search Options

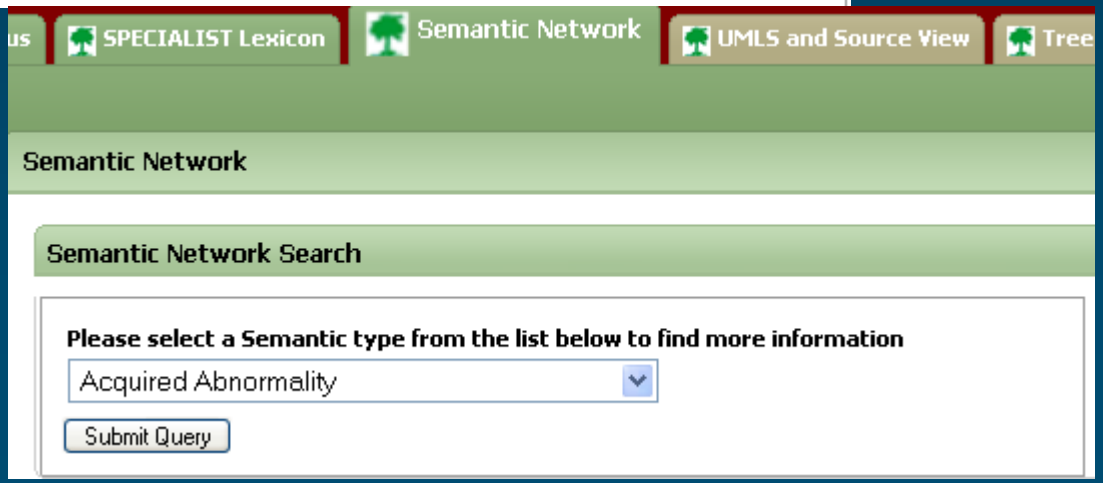


UMLSKS v5 Search Options

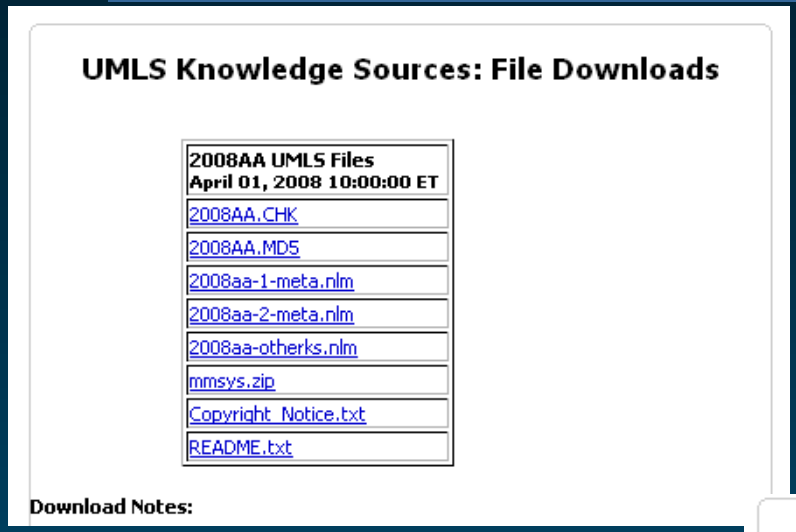


Search Options





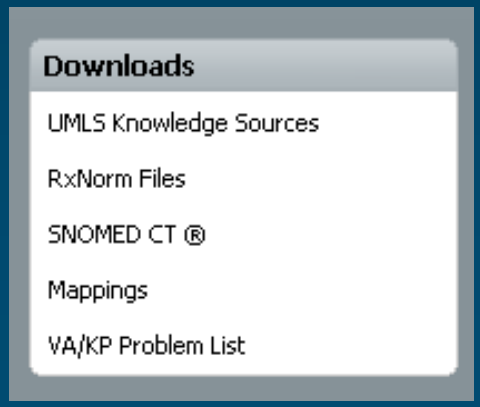
UMLSKS: Downloading files



UMLS Knowledge Sources: File Downloads

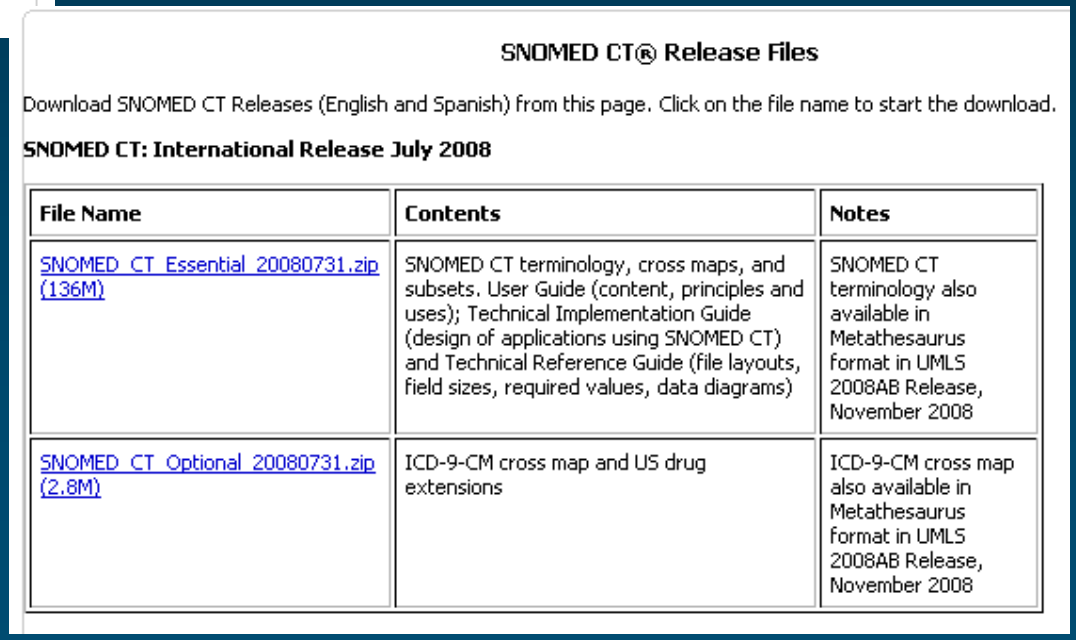
2008AA UMLS Files April 01, 2008 10:00:00 ET
2008AA.CHK
2008AA.MD5
2008aa-1-meta.nlm
2008aa-2-meta.nlm
2008aa-otherks.nlm
mmsys.zip
Copyright_Notice.txt
README.txt

Download Notes:



Downloads

- UMLS Knowledge Sources
- RxNorm Files
- SNOMED CT ®
- Mappings
- VA/KP Problem List



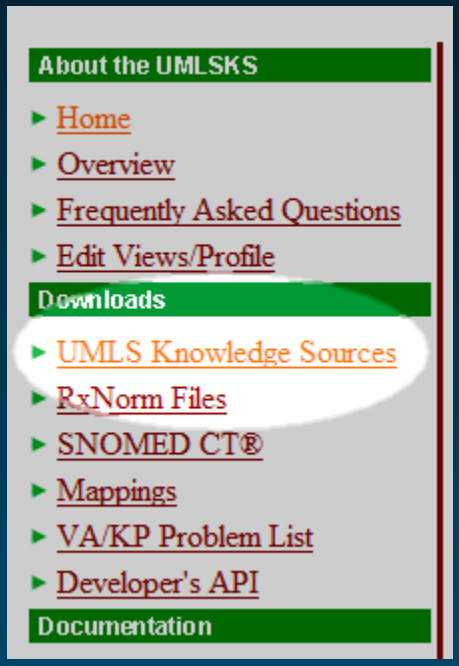
SNOMED CT® Release Files

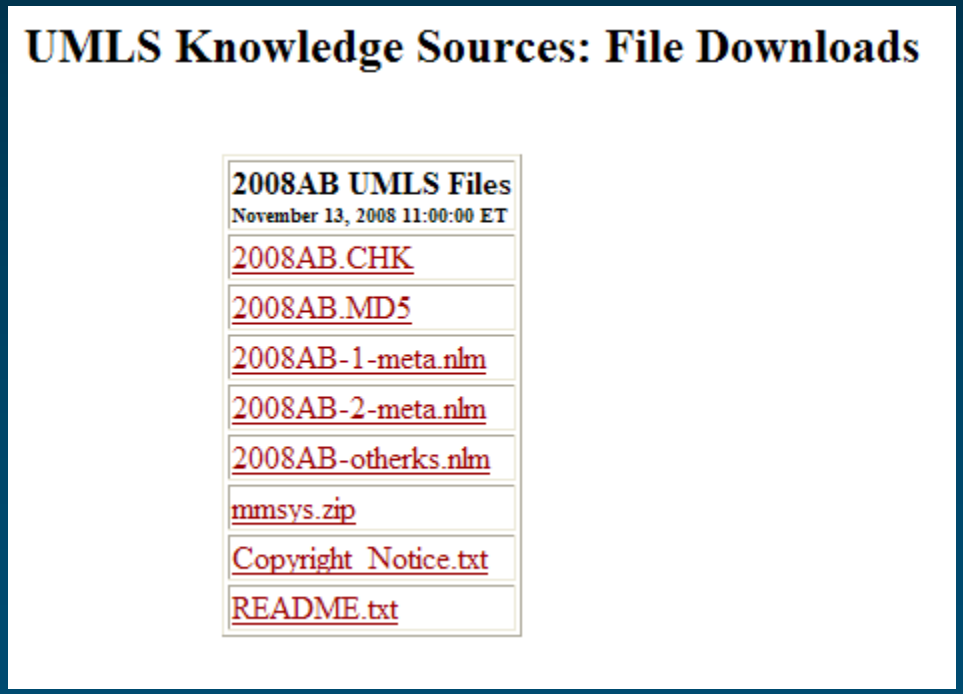
Download SNOMED CT Releases (English and Spanish) from this page. Click on the file name to start the download.

SNOMED CT: International Release July 2008

File Name	Contents	Notes
SNOMED_CT_Essential_20080731.zip (136M)	SNOMED CT terminology, cross maps, and subsets. User Guide (content, principles and uses); Technical Implementation Guide (design of applications using SNOMED CT) and Technical Reference Guide (file layouts, field sizes, required values, data diagrams)	SNOMED CT terminology also available in Metathesaurus format in UMLS 2008AB Release, November 2008
SNOMED_CT_Optional_20080731.zip (2.8M)	ICD-9-CM cross map and US drug extensions	ICD-9-CM cross map also available in Metathesaurus format in UMLS 2008AB Release, November 2008

UMLSKS v5: Downloading files





The SPECIALIST Lexicon and Lexical Tools

SPECIALIST Lexicon

- Syntactic English lexicon of common words, biomedical terms (330K+ words, 550K+ variants)
- Word properties
  - Syntax (how words are put together)
  - Morphology (inflection, derivation, and compounding)
  - Orthography (spelling)
- Used by SPECIALIST Natural Language Processing System to process text and terms
  - Customizable
  - Used to maintain Metathesaurus, indexes

⑩ Adrenal gland diseases

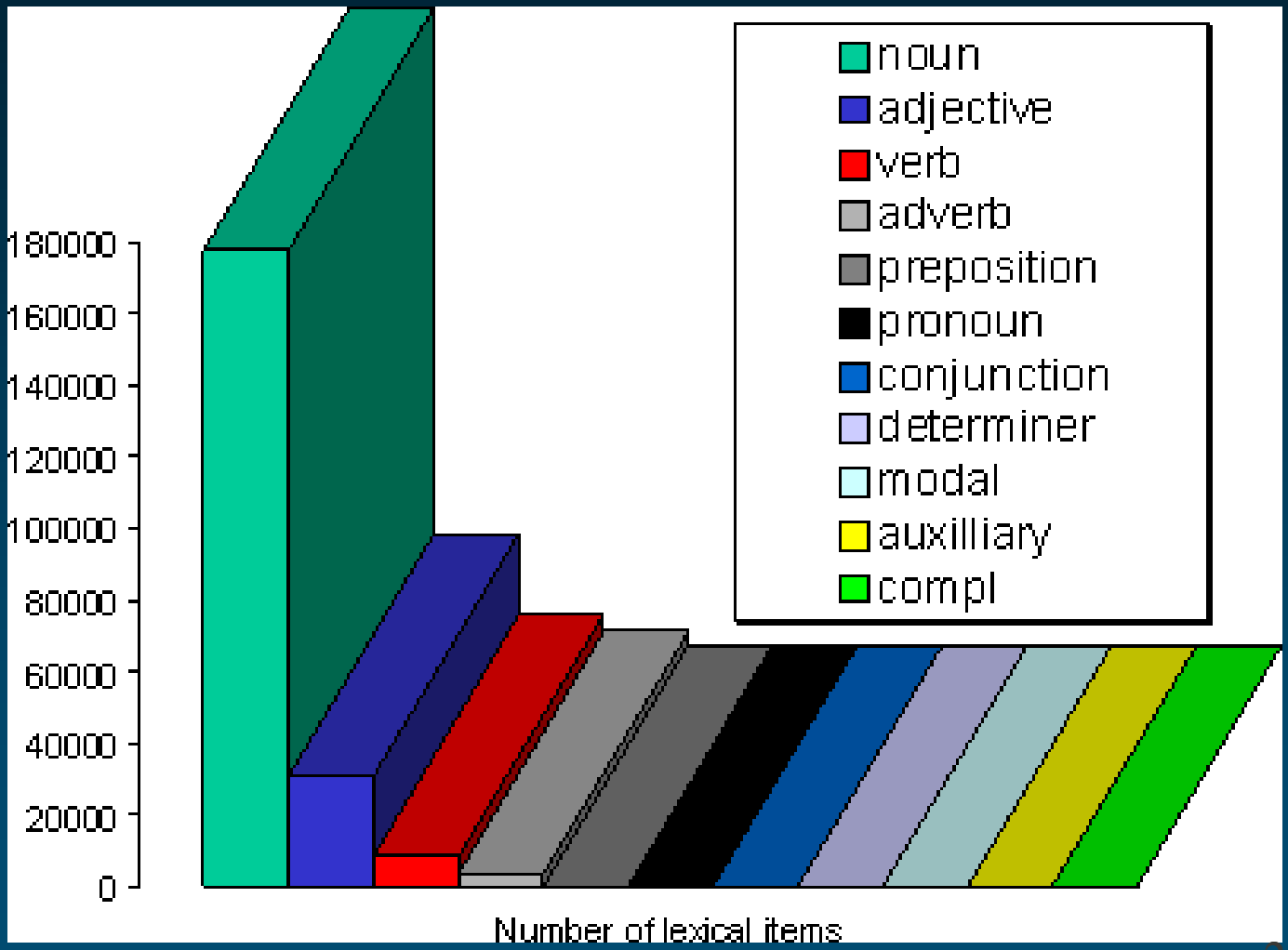
⑩ Diseases of the adrenal glands

⑩ Adrenal disorder

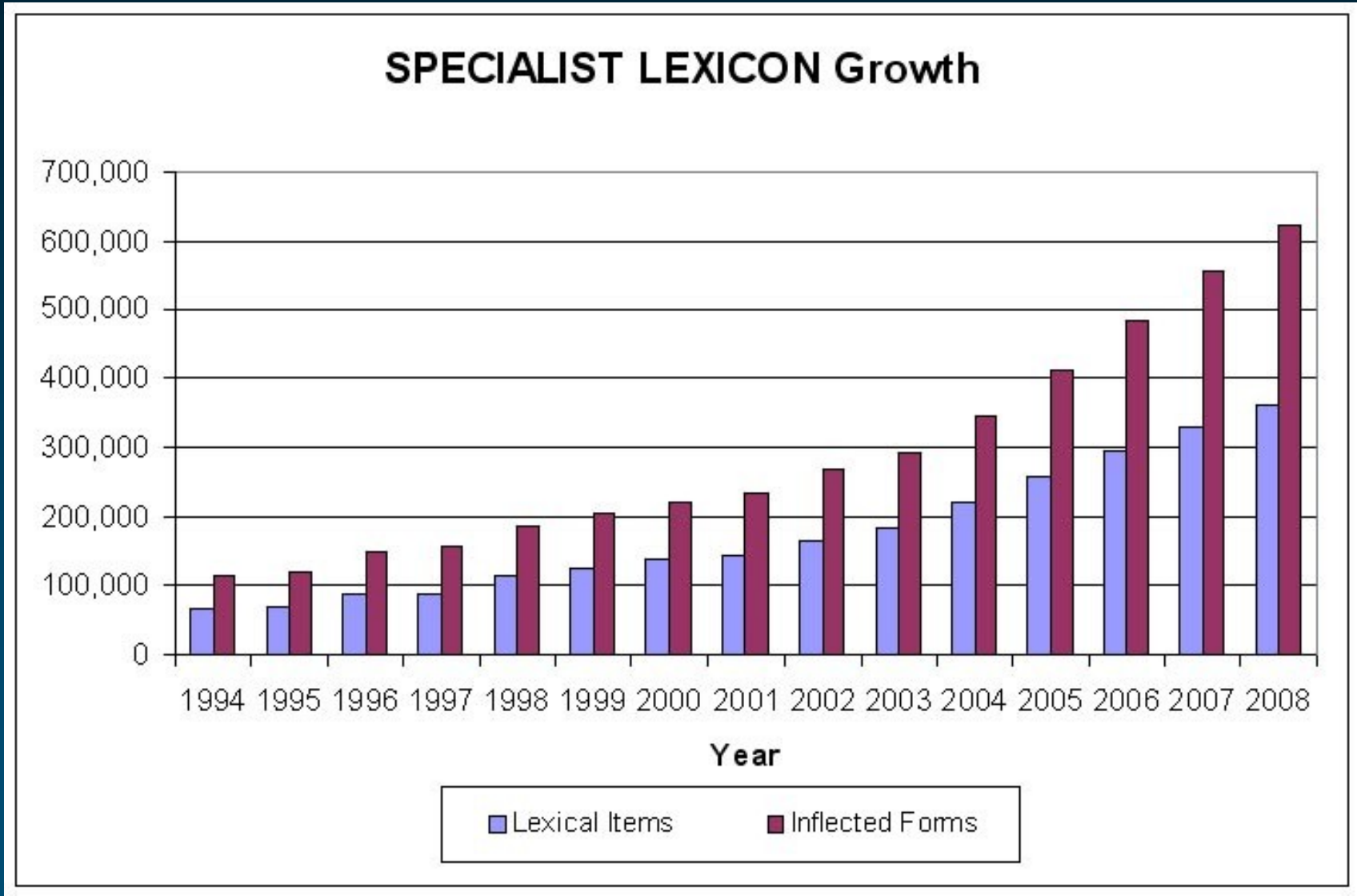
⑩ Disorder of adrenal gland

⑩ C0001621

SPECIALIST Lexicon by part of speech



Specialist Lexicon Growth



Lexicon Growth by Year

Release	Items	Forms	New Items	New Forms
2008	360,688	625,041	30,233	67,644
2007	330,455	557,397	33,141	75,169
2006	297,314	482,268	40,515	69,755
2005	256,799	412,473	35,916	68,490
2004	220,883	343,983	37,582	51,004
2003	183,301	292,979	19,413	24,187

SPECIALIST Lexicon Lexical Entry

```
{base=disease
    entry=E0023270
        cat=noun
        variants=reg
        variants=uncount
        compl=pphr(of,np|bone|)
        compl=pphr(of,np|breast|)
        compl=pphr(of,np|liver|)
        compl=pphr(of,np|ovary|)}
```

- Base form
- Unique identifier
- Part of speech
- Lexical variants
- Prepositional phrase complements

Orthography

- Spelling variants
  - oe/e
  - ae/e
  - ise/ize
  - genitive mark

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

- British-American variants
  - ➉ criticise -- criticize
  - ➉ centre -- center
  - ➉ foetus -- fetus

SPECIALIST Lexicon lexical records

```
{base=Kaposi's sarcoma
spelling_variant=Kaposi sarcoma
entry=E0003576
        cat=noun
        variants=uncount
        variants=reg
        variants=glreg
}
{base=aspirate
entry=E0010803
    cat=verb
    variants=reg
    tran=np

nominalization=aspiration|noun|E0010804
}
```

```
{base=chronic
entry=E0016869
        cat=adj
        variants=inv
        position=attrib(1)
        position=pred
        stative
}

{base=deja vu
spelling_variant=deja-vu
spelling_variant=déjà vu
entry=E0021340
        cat=noun
        variants=uncount
}
```

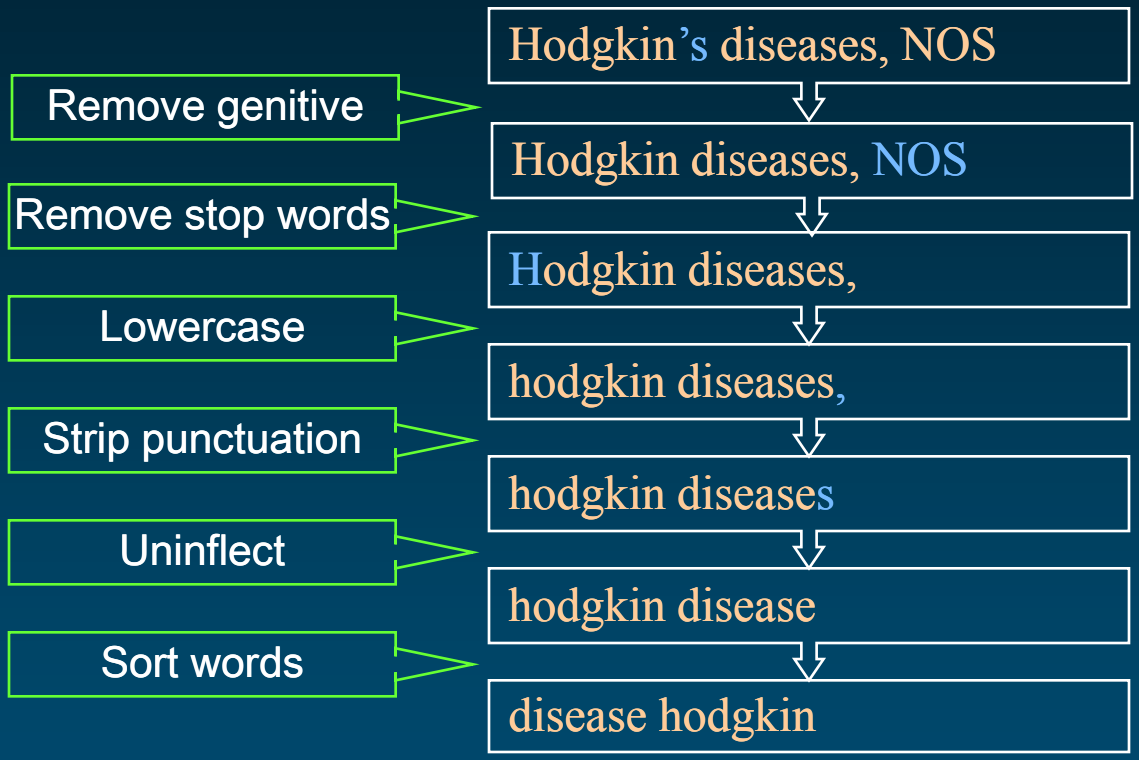
Lexical Tools

- Manage lexical variation in biomedical terminologies and text
- Used separately or with SPECIALIST Lexicon
- Perform transformations selected and ordered by users
- 3 primary programs: normalizer, word index generator, lexical variant generator

Lexical Tools

- Wordind
  - breaks strings into words
  - produces the Metathesaurus word indexes (MRXW)
- lvg
  - performs various lexical transformations
  - 58 flow components and 32 options in 2005
- NORM
  - a selection of LVG transformations
  - used for Metathesaurus indexing
  - produces Metathesaurus normalized word and string indexes (MRXNW & MRXNS)
  - used to access those indexes

Normalization



Normalization: Example

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

Normalized term is not necessarily readable

Lexical Systems Group

Official Websites	http://SPECIALIST.nlm.nih.gov http://mmtx.nlm.nih.gov
Mail Addresses	Allen Browne - browne@nlm.nih.gov Guy Divita - divita@nlm.nih.gov Chris Lu - lu@nlm.nih.gov Lexical Systems group - umlslex@nlm.nih.gov

MetaMap/MMTx

- To map biomedical text to concepts in the UMLS Metathesaurus
- To find Metathesaurus concepts in text

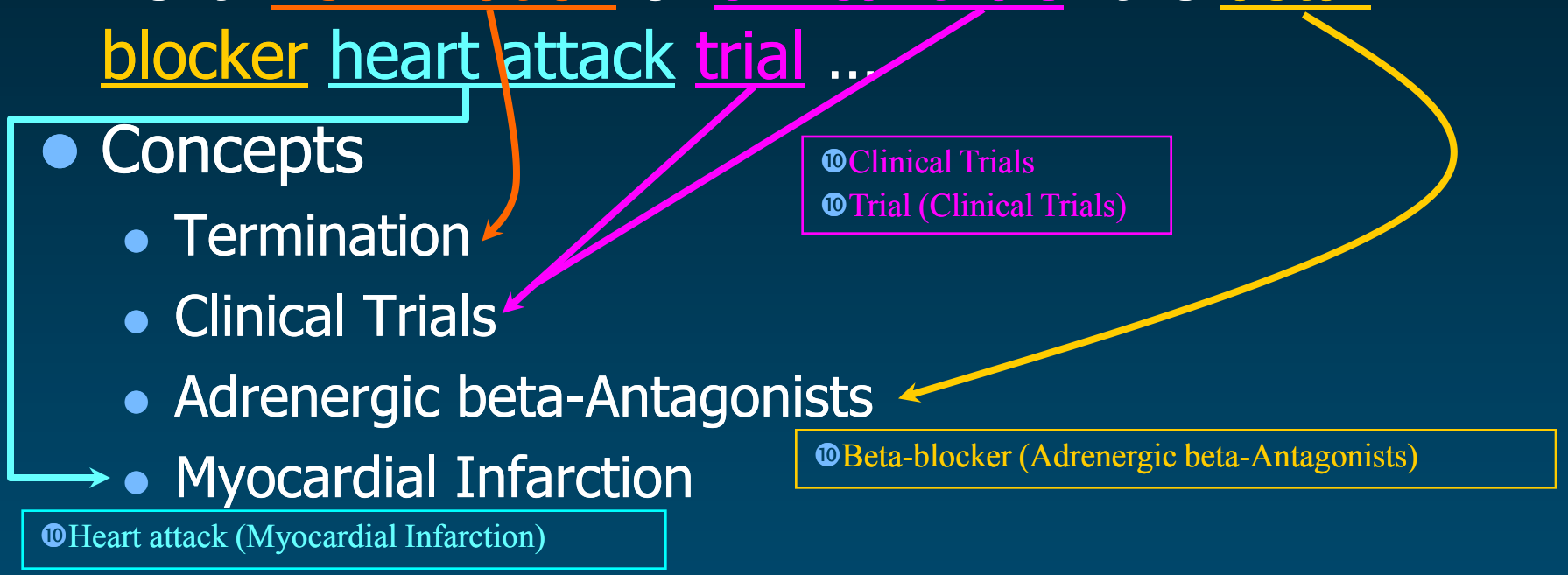
- MMTx was created to provide a distributable version of MetaMap

The MetaMap/MMTx Algorithm

- Parsing
  - Using SPECIALIST minimal commitment parser, SPECIALIST lexicon, a part of speech tagger
- Variant generation
  - Using SPECIALIST lexicon, Lexical Variant Generation (LVG)
- Candidate retrieval
  - From the Metathesaurus
- Candidate evaluation
- Mapping construction

MetaMap/MMTx Example

- Text: Termination of clinical trials: the beta-blocker heart attack trial ...
- Concepts
  - Termination
  - Clinical Trials
  - Adrenergic beta-Antagonists
  - Myocardial Infarction



Semantic Network

Semantic Network

- Semantic Types
  - 135 broad subject categories (Clinical Drug, Virus)
  - Organized into 2 hierarchies: entity, event

    Addison's Disease (concept)

    Semantic Type: Disease or Syndrome
- Semantic Relationships
  - 54 possible, useful links between categories
  - Hierarchical: isa
  - Associative, e.g. causes

    Virus **causes** Disease or Syndrome
- Types + Relationships = Semantic Network
- http://www.nlm.nih.gov/research/umls/meta3.html

Why have a Semantic Network?

- Semantic Types
  - High level categories assigned to Metathesaurus concepts
  - Independent of position in source hierarchies
- Semantic Relations
  - Useful links between Semantic Types
  - Relationships may hold at the concept level
  - Other relationships may apply at the concept level

Semantic Types categorize

- Concept: Discipline of Nursing
- Semantic Type: Biomedical Occupation or Discipline

- Concept: Nursing Homes
- Semantic Type: Health Care Related Organization
  Manufactured Object

- Concept: Home Nursing
- Semantic Type: Health Care Activity

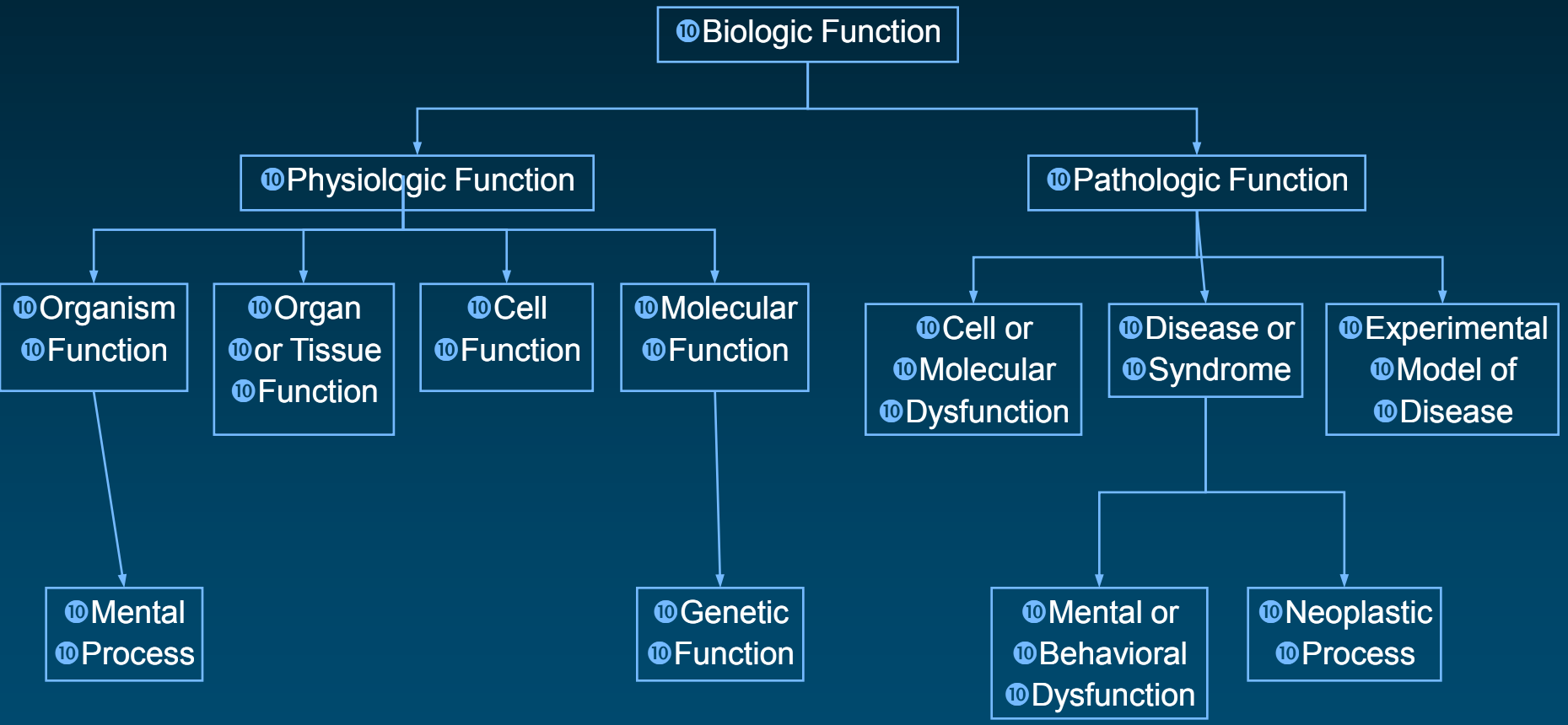
Semantic Type: Disease or Syndrome

- Natural Phenomenon or Process
  - Biologic Function
    - Physiologic Function
      - Organism Function
        - » Mental Process
      - Organ or Tissue Function
      - Cell Function
      - Molecular Function
        - » Genetic Function
  - Pathologic Function
    - ***Disease or Syndrome*** 
    - Mental or Behavioral Dysfunction
      - Neoplastic Process

54 Semantic Relationships

- Hierarchical (isa = is a kind of)
  - among types
    - Animal *isa* Organism
    - Enzyme *isa* Biologically Active Substance
  - among relationships
    - prevents *isa* affects
- Non-hierarchical
  - Sign or Symptom *diagnoses* Pathologic Function
  - Pharmacologic Substance *treats* Pathologic Function

“Biologic Function” hierarchy (isa)



Semantic Relationships

Define useful relations between types

Biomedical Occupation or Discipline	ASSOCIATED_WITH	Social Behavior
Language	ISSUE_IN	Biomedical Occupation or Discipline
Manufactured Object	CAUSES	Injury or Poisoning
Diagnostic Procedure	USES	Manufactured Object
Health Care Activity	ISA	Occupational Activity
Health Care Activity	AFFECTS	Disease or Syndrome

Semantic Relationships

Disease or Syndrome associated_with Finding

Disease or Syndrome result_of Pathologic Function

Body Part, Organ, or Organ Component location_of Disease or Syndrome

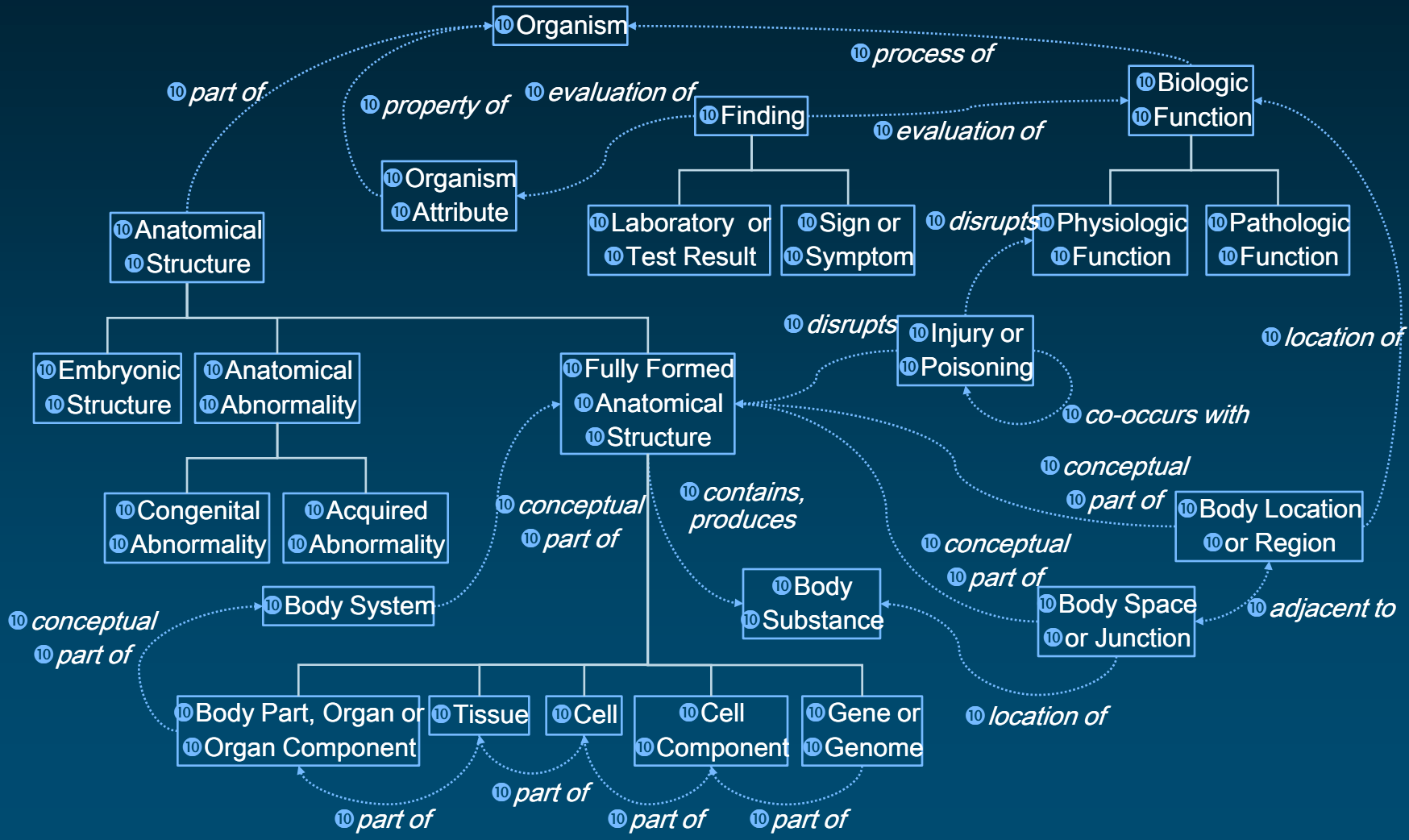
Hormone affects Disease or Syndrome

Hormone complicates Disease or Syndrome

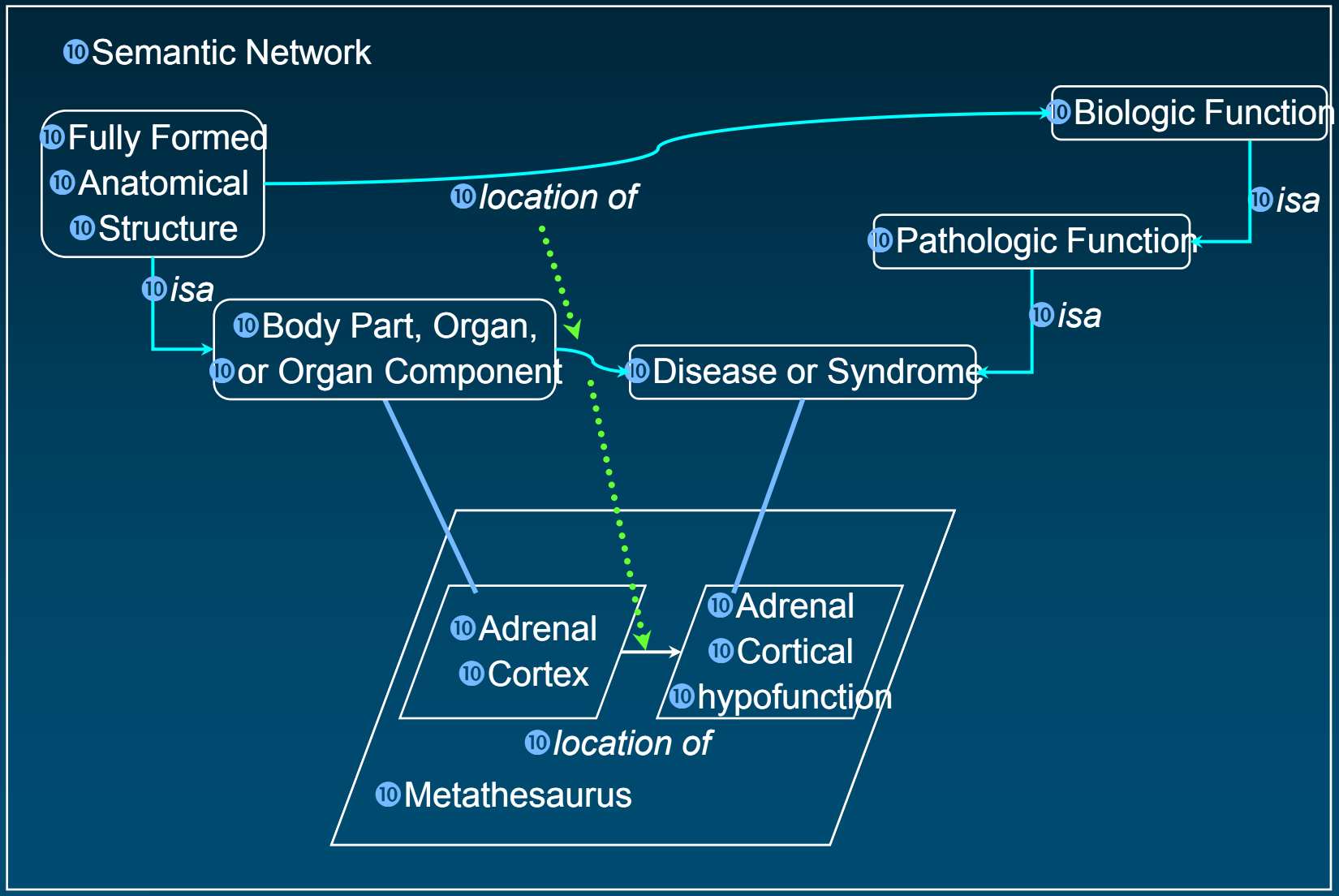
Laboratory Procedure diagnoses Disease or Syndrome

Pharmacologic Substance treats Disease or Syndrome

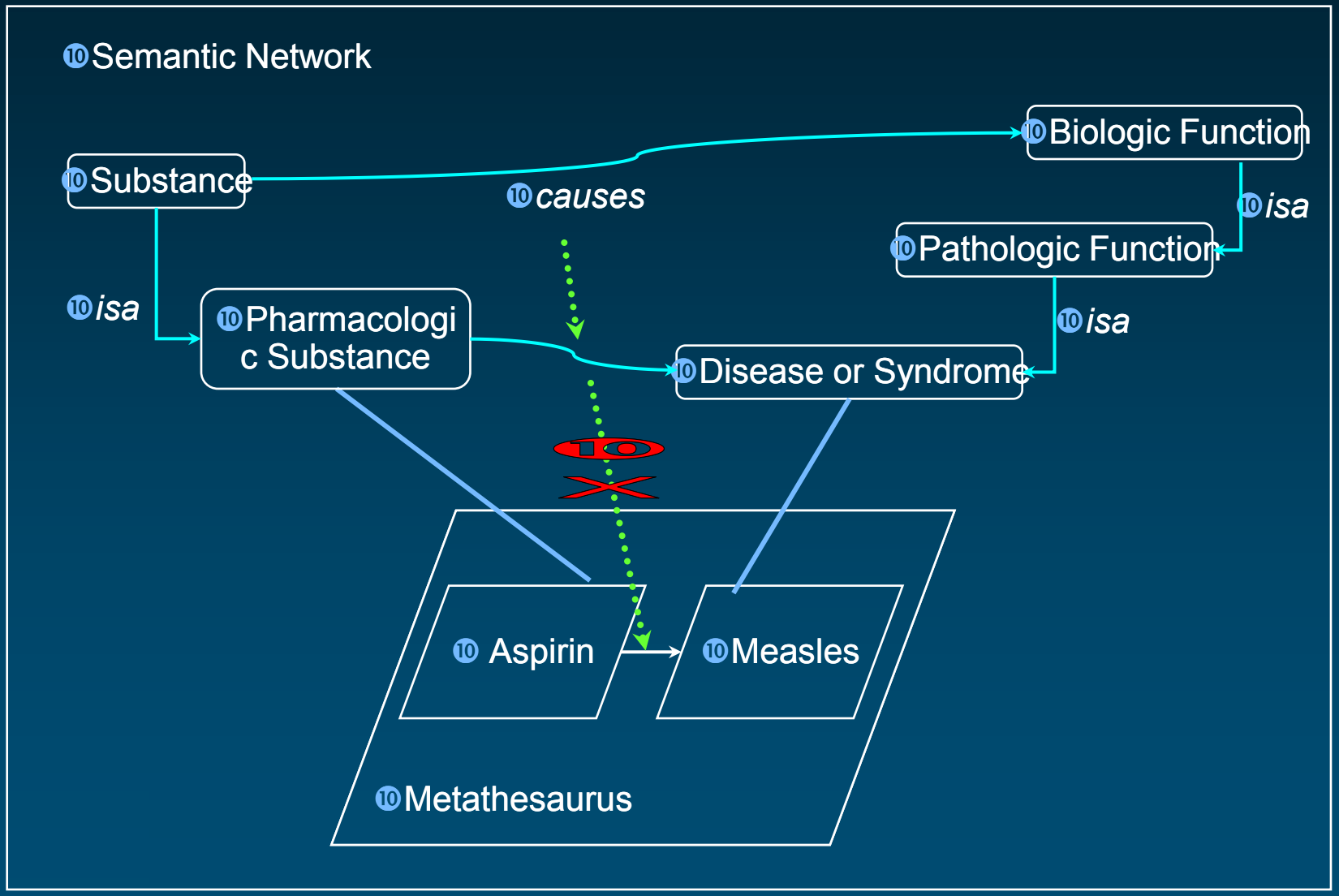
Associative (non-isa) relationships



Relationships can inherit semantics



Relationships don't always inherit semantics



Why have a Semantic Network?

- Semantic Type serves as a high level category assigned to each Metathesaurus concept, *independent of its position in its original source hierarchy*

- A Semantic Relation is a possible link between 2 concepts assigned those Semantic Types
  - The relationship may hold at the concept level
  - Other relationships may apply at the concept level

UMLS and Health Data Standards

Toward a nationwide system of health information

- Adoption
  - Incentives, Regulatory Reform, Workforce Needs and Impacts, Public Awareness
- Interoperability
  - Product Certification
  - Data Standards
  - Standard Product Identifiers and Vocabulary
  - Drug Records
- Connectivity
  - Privacy Standard and Criminal Sanctions, Nationwide Health Information Network (NHIN), Consumer Protections

Electronic Health Records (EHR)

- National collaboration toward secure, Internet-accessible EHR in 10 years
- Federal and private-sector to develop standards with agreement from all stakeholders
- Electronic health records and other information technology to transform health care system
  - reduce medical errors, improve quality of care
  - minimize paperwork hassle, lower costs

Secretary Leavitt Takes News Steps to Advance Health IT, June 15, 2005 (www.hhs.gov/healthit)

The call for standards

- 1991 – IOM: *The Computer-Based Patient Record*
- 1995 – PHS: *Making a Powerful Connection*
- 1996 – Congress: *HIPAA*
- 2000 – IOM: *To Err is Human*
- 2000 – NCVHS: *Report on Standards for PMRI*
- 2001 – IOM: *Crossing the Quality Chasm*
- 2002 – NCVHS: *PMRI recommendations (1st Set)*
- 2002 – AMIA Congress: *Drug Nomenclature Information*
- 2002 – AAMC conference: *IT Enabling Clinical Research*
- 2003 – CHI eGov: *U.S. gov't target standards (1st set)*
- 2003 – Connecting for Health: *Standards Wk.Gr.Report*

Strategy for U.S. health data standards

- Establish mechanisms for designating U.S. standards
  - ✔ Pick best available as starting point
  - ✔ Support development, maintenance, distribution
- Coordinate development of selected standards to achieve non-overlapping, interlocking set
- Broaden participation in standards development
- Promote use and improvement

Federal standards selection mechanisms

- HIPAA (1996) - Health Insurance Portability and Accountability Act of 1996 requires administrative standards
- NCVHS - National Committee on Vital and Health Statistics, a long-standing (50+ years) advisory committee to HHS expanded by HIPAA, recommends standards
- CHI (2001) Consolidated Health Informatics project, a cross-agency eGov initiative designates U.S. gov't-wide clinical data standards
- Medicare Modernization Act (2003) - requires e-Prescribing standards; establishes Commission on Systemic Interoperability

Standards implementation

- CHI (clinical) *LOINC*
  - E.g., lab test results, problems, diagnoses, history, physical
  - Electronic exchange of clinical health information in U.S. Government systems
- HIPAA (administrative) *CPT*
  - e.g., health insurance claims, billing, ordering
  - HIPAA Administrative Simplification provisions
  - Designated DHHS national standards for electronic healthcare transactions
- PHIN (public health) *ICD-9-CM*
  - e.g., disease surveillance, immunization rates, environmental monitoring
  - CDC designated standards for public health reporting

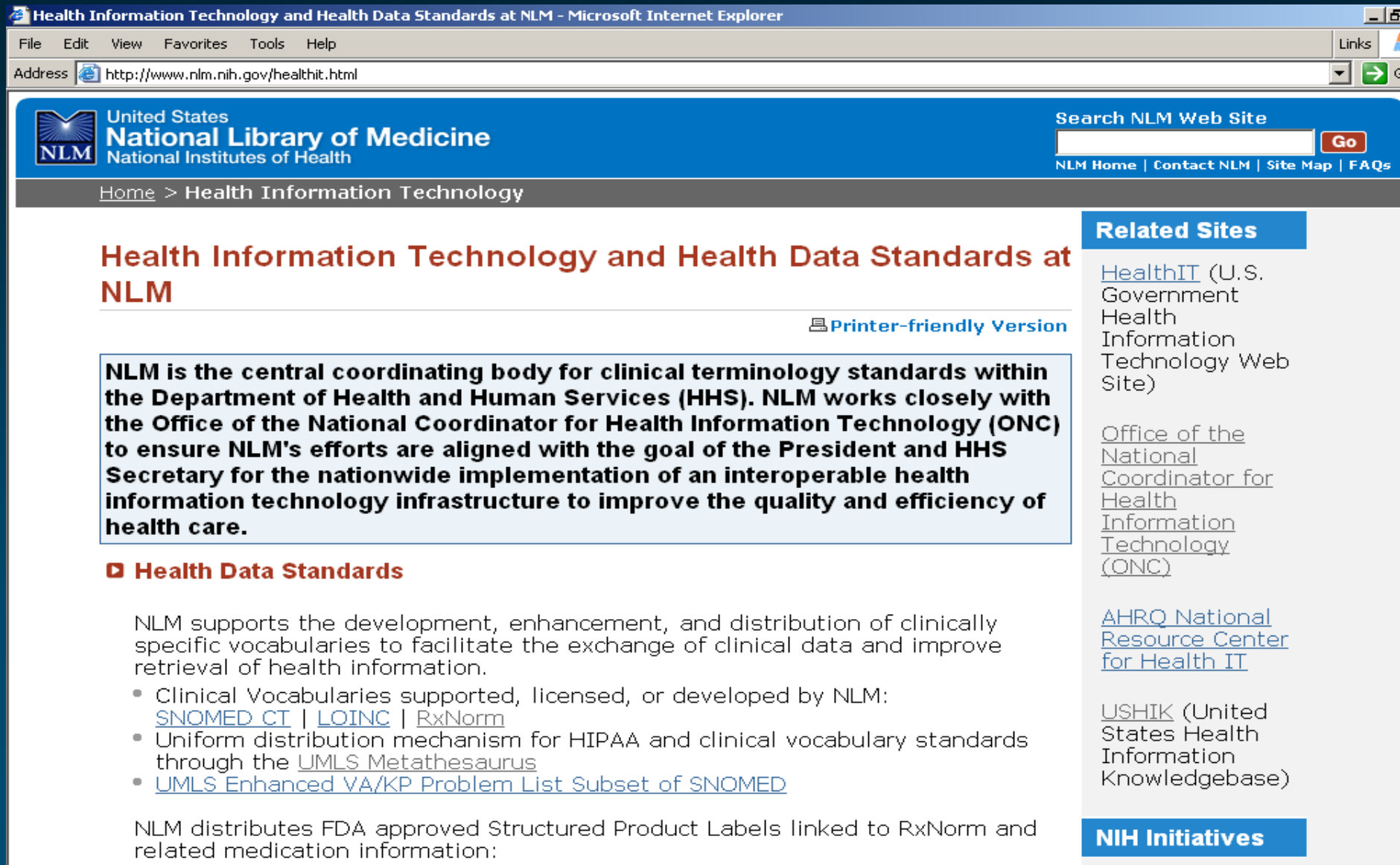
NLM role

- HHS central coordinating body for Patient Medical Record Information (PMRI) terminologies
- Work with agencies, organizations to support standards
- Support mappings between standard clinical vocabularies (SNOMED, LOINC, RxNorm) and others
- Reduce overlap between standards vocabularies

NLM role 2

- Maintain UMLS as uniform distribution mechanism for
  - HIPAA code sets, recommended PMRI terminologies
  - mappings between standard codes and PMRI terminologies
  - information on valid vocabularies for transaction standards

Health Information Technology Homepage



International Health Terminology Standards Development Organisation (IHTSDO)

- National Library of Medicine (NLM) is U.S. member

- Promotes SNOMED CT as one of key US standards
- Distributes SNOMED CT

SNOMED CT in the UMLS

- English descriptions and relationships (2004)
  - Concepts: 298,090
  - Descriptions: 736,946 (synonyms)
  - Relationships: 1,315,910
- Added to Metathesaurus:
  - Concepts: 37,089
  - Descriptions: 350,464
- Spanish terms added in 2004AB

SNOMED CT in the Metathesaurus

- Core content U.S. English, Spanish
  - Concepts, Descriptions, Relationships tables
  - Generic drugs
  - History table
  - ICD-9-CM mapping

- SNOMED updates in UMLS
  - January SNOMED CT update → Spring UMLS
  - July SNOMED CT update → Fall UMLS

Additional information on NLM Web site

- SNOMED CT® in the UMLS® Metathesaurus®: Inversion Source Transparency Achieved Jan. 2005
- SNOMED CT in the 2005AB Release
- SNOMED CT® in the UMLS® Metathesaurus®:Release Source Transparency Achieved Jan. 2005
- SNOMED CT in the 2005AB Release
- SNOMED CT Information from the College of American Pathologists

RxNorm

RxNorm Project

- Developed by NLM to address
  - missed synonymy in UMLS clinical drugs
  - medication errors
- CHI recommended standard
- Intended to support
  - effective sharing of drug data across systems
  - electronic health record (EHR)
  - computerized physician order entry (CPOE)

RxNorm Contents

- Full sources
  - FDA, NDDF, Medispan, Micromedex, Multum
  - More to come
- Partial sources
  - SNOMED CT, MeSH
- NOT in RxNorm
  - USP Medicare Model Guidelines
  - MedDRA (adverse reactions)

RxNorm (NLM)
SNOMED CT (CAP)
MTHFDA (FDA NDC)
VANDF (VA)
Gold Standard
NDDF (FirstDataBank)
MDDB (Medispan)
MMX (Micromedex)
Multum

RxNorm Release Files

- RxNorm (standalone vocabulary)
  - Normalized forms of clinical drugs
  - Created by NLM
  - From commercial and government clinical drug vocabularies
  - Full monthly, and weeky additional updates
- RxNorm subset (extracted from UMLS)
  - RxNorm forms (NLM)
  - Clinical drug content including SNOMED CT, VANDF, FDA NDC

Respect license restrictions

RxNorm Normalized Forms

- Semantic Normal Form (SNF)
  - Active standardized (generic) ingredients
  - Strengths
  - Units of measurement
  - Dosage form
- As administered to a patient
  - Acetaminophen 500 MG Oral Tablet
  - Acetaminophen 500 MG Oral Tablet [Tylenol]

RxNorm Term Types

- SBD = Palifermin 5 mg/ml Injectable Solution [Kepivance] ➔ 7 additional concepts

- IN= Palifermin
- BN= Kepivance
- SBDC= Palifermin 5 mg/ml [Kepivance]
- SBDF = Palifermin Injectable Solution [Kepivance]
- SCD= Palifermin 5 mg/ml Injectable Solution
- SCDC= Palifermin 5 mg/ml
- SCDF= Palifermin Injectable Solution

all are separate RxNorm concepts

Mappings

Mappings

- Synonymy within concept (implied)
- Explicit links between concepts, terms, codes or other content in specified vocabularies
- Source-asserted
  - SNOMED CT→ ICD-9-CM
- Created/supported by NLM
- Represented in
  - MRREL (simple only)
  - MRMAP, MRSMAP (simple and complex)

UMLS – MeSH mapping file

Used in MEDLINE/PubMed searching
Based on synonymy

...

myocardial infarction|attack coronary
myocardial infarction|attack heart (nos)
myocardial infarction|cardiac infarction
myocardial infarction|cardiac infarction, nos
myocardial infarction|cardiac; infarction
myocardial infarction|heart attack
myocardial infarction|heart attack, nos
myocardial infarction|heart attacks
myocardial infarction|heart infarction

Other Types of Mappings

LOINC - CPT

- 1795-4 AMYLASE:CCNC:PT:FLU:QN →
- 1798-8 AMYLASE:CCNC:PT:SER:QN →
- 1799-6 AMYLASE:CCNC:PT:UR:QN →
- 82150 Amylase (blood or urine)

SNOMED CT – CPT

- 313500004 Urine protein/creatinine ratio measurement →
- 82570 Creatinine; other source
- 84155 Protein, total, except by refractometry; serum

Mapping projects planned/underway

- CHI standards → HIPAA code sets
  - SNOMED CT → ICD-9-CM (support reimbursement)
  - SNOMED CT→ ICD-10-CM
  - SNOMED CT → CPT/HCPCS
  - LOINC → CPT
- SNOMED CT →
  - MedDRA
  - MeSH
  - Nursing (NIC, NOC, NANDA)

Key NLM Assumptions about Mappings

- Participants must include producers on both ends and users
- Mappings may prompt changes to content and adjustment to update schedules
- Mappings must keep current at both ends
- Mappings will be distributed in the UMLS with license terms applicable to both ends (MRMAP, MRSMAP)
- Mapping is an R & D problem; iteration is required to build highly functional maps
- Webcast: http://www.nlm.nih.gov/research/umls/umlswebcasts/index.html

Questions?

Summary

- UMLS = 3 knowledge sources
  - Metathesaurus
  - Semantic Network
  - SPECIALIST Lexicon and Lexical Tools
- UMLSKS
  - Remote access and browser
  - Download files
  - Documentation
- MetamorphoSys
  - Installs UMLS, customizes Metathesaurus
  - RRF Browser

Bibliography of UMLS Articles

- Search MEDLINE®/PubMed® for recent research journal articles on the UMLS (1997 to present)

- Comprehensive Bibliography 1986-1996
  - Selden, Catherine
    Unified Medical Language System (UMLS): January 1986 through December 1996 : 280 citations / prepared by Catherine R. Selden, Betsy L. Humphreys. -- Bethesda, Md. (8600 Rockville Pike, Bethesda 20894) : U.S. Dept. of Health and Human Services, Public Health Service, National Institutes of Health, National Library of Medicine, Reference Section ; Pittsburgh, PA : Sold by the Supt. of Docs., U.S. G.P.O., 1997.
    -- (Current bibliographies in medicine ; 96-8)

References

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

UMLS Documentation and Support

- UMLS homepage
  - http://umlsinfo.nlm.nih.gov/
- UMLSKS homepage
  - http://umlsks.nlm.nih.gov
- UMLSUSERS-L
  - subscribe to discussion list
- NLM Customer Service email:
  - custserv@nlm.nih.gov

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- Create UMLSKS account
- Explore Knowledge Sources
- Download files or request DVD
- Create subsets using MetamorphoSys


Center
Drive
Library
of
Medicine
Thank you