

AMIA 2006 Tutorial T12 Unified Medical Language System[®] UMLS OVERVIEW

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National Institutes of Health U.S. Dept. of Health & Human Services





Schedule

- 1:00 1:30 UMLS Overview
 1:30 2:00 Metathesaurus
 2:00 2:15 Electronic Health Data Standards
 2:15 2:30 UMLS Knowledge Source Server
 2:30 3:00 MetamorphoSys: Customizing the Metathesaurus
- 3:00 3:30 break -
- 3:30 4:00 SPECIALIST Lexicon and Lexical Tools 4:00 – 4:30 MetaMap Technology Transfer (MMTx)



Unified Medical Language System

9/30/06: Draft LOINC to CPT Mappings now available for download from the UMLSKS. ••• New to the UMLS? Register now.

About the UMLS Resources

Metathesaurus; Semantic Network; SPECIALIST Lexicon and lexical programs; MetamorphoSys

Accessing UMLS Knowledge Sources

Metathesaurus license; Semantic Network; SPECIALIST Lexicon; DVD

Knowledge Source Server

Download files; searching; additional tools and resources

Documentation

<u>Help</u>

Training; contact us; FAQs; listserv

About

NLM's Unified Medical Language System (UMLS) project develops and distributes multipurpose, electronic "Knowledge Sources" and associated lexical programs for system developers. Researchers will find the UMLS products useful in investigating knowledge representation and retrieval questions.

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Metathesaurus Source Vocabularies

- SNOMED CT
- LOINC
- <u>RxNorm</u>
- <u>MeSH</u>
- List of Sources
- Source FAQs
- <u>Mappings</u>

More Resources

- Metathesaurus License
- <u>Tools</u>
- Learning Resources
- MetaMap Transfer (MMTx)
- <u>Archives</u>

The UMLS consists of



3 Knowledge Sources + associated tools

UMLS Objectives

Intellectual middleware

A set of multi-purpose tools for system developers

 Knowledge Sources used to overcome:
 disparities in language and 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 Ivg (lexical programs) Distributed on DVD Downloaded from UMLS Knowledge Server (UMLSKS) The UMLS is *not* an end-user

application

UMLS Access

Remote access

 UMLS Knowledge Server (UMLSKS) http://umlsks.nlm.nih.gov/
 Browsers, Navigators
 APIs
 Download files and programs
 Documentation
 Local access
 MetamorphoSys: install files locally, create

- customized Metathesaurus subsets
- Subset Browser: search, browse, view customized subsets

Metathesaurus License Agreement

Online Web-based license:

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

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



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

License Agreement Restrictions

- 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.
 See list of current sources in Appendix A.1.

Metathesaurus

The Metathesaurus is

very large
 multi-purpose
 multi-lingual

It contains information about biomedical and health related concepts their various names and associated codes the relationships among them

Metathesaurus: clusters terms by meaning

Concepts contain synonymous terms
 Preferred term is chosen (default can be changed)
 Unique identifier (CUI) is assigned

term	source	term type	source ID
Addison's disease	/letathesaurus	PN	
Addison's disease	SNOMED CT	PT	363732003
Addison's Disease	MedlinePlus	PT	T1233
Addison Disease	MeSH	PT	D000224
Bronzed disease	SNOMED Intl	SY	DB-70620
Primary Adrenal Insufficiend	cy MeSH	EN	D000224
Primary hypoadrenalism syndrome, Addison	MedDRA	LT	10036696



Organization of Concepts



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

54 Semantic Relationships

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

Semantic Network Relationships



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

SPECIALIST Lexicon

Syntactic English lexicon of common words, biomedical terms (250K+ words, 400K+ 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 Diseases of the adrenal

Adrenal gland diseases Diseases of the adrenal glands Adrenal disorder Disorder of adrenal gland 17 C0001621

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=in
entry=E0033870
 cat=prep
}

Lexical Tools

JAVA programs to manage lexical variation, indexing, normalization in biomedical text

Wordind

breaks strings into words

produces the Metathesaurus word indexes (MRXW)

Ivg

performs various lexical transformations

58 flow components and 38 options in 2006

NORM

- a selection of LVG transformations
- 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

2006AD UMLS

November 2006

Metathesaurus:

- 120 sources
 1,352,403 concepts
 17 languages
- Semantic Network:
 135 Semantic Types
 54 Semantic Relationships

SPECIALIST Lexicon:

Over 297K records (over 482K inflectional forms)

Metathesaurus

Metathesaurus Sources

- Wide range of general and specialized biomedical terminologies
- Used in variety of settings and purposes:
 - Clinical
 - Administrative
 - Public Health Reporting
 - Research

Metathesaurus Sources

120 vocabularies in 17 languages
 Sets of valid values

 Thesauri, e.g., MeSH, CRISP, NCI
 Statistical classifications, e.g., ICD-9-CM
 Billing codes, e.g., CPT
 Clinical coding systems, e.g., SNOMED CT

See License Appendix, documentation

Metathesaurus: not a single vocabulary

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. In: Broering NC, ed. High- Performance Medical Libraries: advanced information management for the virtual era. Westport (CT): Meckler; 1993, p. 33.

Metathesaurus highlights

Concept based

Represents the meaning in each source
Represents and delivers data in common for

- Represents and delivers data in common format
- Adheres to principle of "source transparency"
- Source information tagged
 Context-free unique identifiers added

Normalized word and string indexes included

Source Data to Metathesaurus Files

Names, Synonyms Terms, Codes	\rightarrow	MRCONSO
Relationships	\rightarrow	MRREL
Hierarchies	\rightarrow	MRHIER
Attributes	\rightarrow	MRSAT
Definitions	\rightarrow	MRDEF
Mappings	\rightarrow	MRMAP, MRSMAP, (MRREL, MRCONSO)

CUIs links concept data across files 28

MRCONSO (sample rows 1..5)

D000224

10052381

D000224

MH

3 MSH

4 MDR

5 MSHFRE

	1	2	3	4	5	6	7	8	9	10	11	
	CUI	LAT	TS	LUI	STT	SUI	ISPREF	AUI	SAUI	SCUI	SDU	1
1	C0001403	ENG	Ρ	L0001403	PF	S0354372	Υ	A4367951				
2	C0001403	ENG	Ρ	L0001403	PF	S0354372	Ν	A2922421	48 <mark>562401</mark> 4	4 <mark>363732003</mark>		
3	C0001403	ENG	Ρ	L0001403	VC	S0010794	Υ	A0019740		M0000346	D0002	24
4	C0001403	ENG	S	L0494851	PF	S2164152	Ν	A2018589				
5	C0001403	FRE	Ρ	L3246333	PF	S3773545	Y	A3996251			D0002	21
	12	13		14			1	5	1	5 17		
	SAB	TTY		CODE		STR 위 SUI					ESS	CVF
N	ITH	PN	N	OCODE	Addis	ddison's disease 0						
S	NOMEDCT	PT	36	63732003	Addis	son's disea	se		4	N		

Primary adrenal insufficiency

Addison's Disease

Addison, maladie

0

3

3

Ν

Ν

Ν

MRCONSO (sample rows 6..10)

D000224

	1	2	3	4	5	6	7	8	9		10	11	
	CUI	LAT	SL	LUI	STT	SUI	ISPREF	AUI	SAUI		SCUI	SDL	JI
6	C0001403	FRE	S	L1272481	PF	S1514427	Y	A1464383					
7	C0001403	GER	Ρ	L1229627	PF	S1471573	Y	A4030156				D0002	224
8	C0001403	GER	S	L1239271	PF	S1481217	Y	A4034094				D0002	224
9	C0001403	JPN	Ρ	L3437833	PF	S3965327	Y	A4264008			D000		224
10	C0001403	JPN	S	L3465347	PF S3992841		Y	A4291522				D0002	224
	12		13	14				15		16	17		18
	SAB		TTY	CODE			S	TR		SRL	SUPPRE	SS	CVF
6	WHOFRE	ľ	Г	0410	MAI	LADIE D'AI	DDI	SON		2	N		
7	MSHGER	N	1H	D000224	Addison-Krankheit					3	N		
8	MSHGER	S	Y	D000224	Bro	Bronzehautkrankheit					N		
9	MSHJPN	N	1H	D000224	Add	lison病				3	Ν		
10						⊽ ⊾⊥ == ,+,,,-				_			

Appendix - Metathesaurus relational files (RRF)

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MRREL (sample rows)

	1	2	3	4	5		6
	CUI1	AUI1	STYPE1	REL	CUI2	AUI2	STYPE2
1	C0001403		CUI	RB	C0001621		CUI
2	C0001403	A0019738	AUI	SY	C0001403	A0049628	AUI
3	C0001403	A2922421	SCUI	CHD	C0085859	A2977940	SCUI
4	C0001403	A6326321	SCUI	RO	C0688490	A6339383	SCUI
5	C0001403	A0019743	AUI	PAR	C0935495	A1988358	AUI

7	8	9	10	11	12	13	14	15
RELA	RUI	SRUI	SAB	SL	RG	DIR	SUPPRESS	CVF
	R02837989		MTH			Ν	Ν	
	R18849683		MSH	MSH			Ν	
isa	R19859511	1658795027	SNOMEDCT	SNOMEDCT	0	Y	Ν	
may_treat	R27600039		NDFRT	NDFRT			Ν	
has_member	R08110401		PSY	PSY			Ν	

MRHIER (sample rows)

	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_clus	ster		
4	C0001403	A2922421	1	A3307650	SNOMEDCT	isa			
5	C0001403	A2922421	2	A3307650	SNOMEDCT	isa			
			7			8	9		
			PTR			HCD	CVF		
1	A0434168.A236	7943.A2366890. <i>i</i>	40135391. <i>.</i>	A0054194.A0020)267.A0020270	C19.053.264.263			
2	A0434168.A236	7943.A2366890.	A0135391.	A0072566.A0028	3022	C20.111.163			
3	A0449751.A198	8279.A1988358							
Л	A3684559.A388	A3684559.A3886745.A2880798.A3398606.A3399335.A33 <u>98961.A2872359</u> .							
	A2872360.A330	7650							
5	A3684559.A388	6745.A2880798.	43398606.	A3399335.A3398	3961.A2872359.				
	A2933400.A298	9549.A3307650							

MRSAT (sample rows)

	1	2	3	4	5	
	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	Ν	
2	AT33411754		MESH_UI	NDFRT	D000224	Ν	
3	AT24166602		DESCRIPTIONS TATUS	SNOMEDCT	0	N	
4	AT27438950		REFINABILITY	SNOMEDCT	0	Ν	
5	AT02925340		ST	MTH	R	N	



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	

Metathesaurus Metadata Files

MRFILES:
MRCOLS:
MRDOC:
MRSAB:
MRRANK:

files authority data elements data element valid values source vocabularies source/term types ranks

Generate UMLS release documentation
 Full set of metadata files in subset

Metathesaurus Indexes

 Word: each word in each unique string in each language
 Heart disease yields ENG disease |C0024117|L0024117|S0058458| ENG heart |C0018787|L0018787|S0047194|

 Normalized word: each normalized word in each unique English string
 Disease, diseases, diseased yields ENG | disease |C0024117 |L0024117 |S0058458 |

Normalized string obstructive lung disease ENG | disease lung obstructive | C0024117 | L0024117 | ... S0058458 |
Different Formats for Different Purposes

- Original Release Format (ORF)
- Lexical View
- Natural language processing

- Rich Release Format (RRF)
- Atomic View
- Greater specificity
 - Facilitates maintenance
 - Enables other types of changes and applications
- RRF Browser in MetamorphoSys

How do I use the Metathesaurus?

 Identify useful sources
 Identify useful content from specific sources
 Create customized Metathesaurus using MetamorphoSys

Use UMLS Release Documentation to understand file content and structure

Metathesaurus Use Cases

- Information retrieval
- Thesaurus construction
- Natural language processing
- Automated indexing
- Electronic patient records

Distribution of health data standard vocabularies

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 heart attack myocardial infarction heart attack, nos myocardial infarction heart attacks myocardial infarction heart attacks myocardial infarction heart attacks Electronic Health Data Standards A U.S. Government Health Information Technology Web Site



An initiative of the U.S. Department of Health & Human Services

	··· ··· ···
Health IT Home	Health IT
General Information	Health IT initiatives harness current and emerging information technologies to improve patient safety and convenience while reducing the cost of providing care. Some of the numerous benefits of health IT
Federal Efforts	initiatives will include: a reduction in medical errors, avoidance of costly duplicate testing, and elimination of unnecessary hospitalizations.
Consumer Awareness	The benefits of health IT initiatives range from consumer convenience, as patients will not have to fill out renetitive paper work, to life-saving early detection of an infectious disease outbreak, as anonymous
Focus Areas	data from emergency rooms is sent to public health systems instantly. These initiatives aid in fulfilling the President's goal for most Americans to have electronic health records by the year 2014.

American Health Information Community (AHIC) Topics

The American Health Information Community (AHIC) is a federal advisory body, chartered to make recommendations to the Secretary of HHS on how to accelerate the development and adoption of Health IT. The Community will make recommendations to the Secretary of HHS to enable advancement in four areas of focus by the end of 2006.

- <u>Consumer Empowerment</u> Make available a consumer-directed and secure electronic record of health care registration information and a medication history for patients.
- <u>Chronic Care</u> Allow the widespread use of secure messaging, as appropriate, as a means of communication between doctors and patients about care delivery.
- <u>Biosurveillance</u> Enable the transfer of standardized and anonymized health data from the point of health care delivery to authorized public health agencies within 24 hours of its collection.
- <u>Electronic Health Records</u> Create an electronic health record that includes laboratory results and interpretations, that is standardized, widely available and secure.

Highlighted Federal Efforts

Health & Human Services

<u>Health & Human Services (HHS)</u> - HHS is facilitating the development of standards for Health IT systems that will improve patient care and increase efficiency across the health care system. HHS, through several of its agencies, also provides funding to organizations engaged in building and testing Health IT systems, standards and projects.

Department of Defense

Department of Defense (DoD) - Currently, thousands of military medical providers use the DoD's electronic health record system, AHLTA, and nearly 300,000 outpatient visits are captured digitally every week. DoD's vision is to provide each patient with a continuously updated digital medical record from the

http://www.hhs.gov/healthinformationtechnology/

<u>Veterans Health Administration (VHA)</u> - The VHA is a division of the U.S. Department of Veteran's Affairs, and provides care for over five million veterans of the United States Armed Services. The VHA's

Mission Statement

"We will make wider use of electronic records and other health information technology to help control costs and reduce dangerous medical errors."

-- President Bush, January 31, 2006

"..to link all health records through an interoperable system that protects privacy as it connects patients, providers and payers. Resulting in fewer medical mistakes, less hassle, lower costs and better health."

-- HHS Secretary Mike Leavitt

News

08/22/2006 - <u>President Directs Federal Agencies</u> to Provide Health Care Quality and Price Information for Consumers

05/17/2006 - <u>American Health Information</u> <u>Community Approves First Set of</u> Recommendations

01/17/2006 - HHS Announces Pilot Project Launched to Expand Electronic Prescribing

Speeches

10/07/2005 - <u>Remarks at the First Meeting of the</u> American Health Information Community.

- HHS Secretary Mike Leavitt

Testimony

09/28/2005 - Full Committe Hearing on Healthcare and the IT Revolution

-- Dr. David J. Brailer, HHS National Coordinator for Health IT

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Health Information Technology at NLM

B Printer-friendly Version

NLM is the central coordinating body for clinical terminology standards within the Department of Health and Human Services (HHS). NLM works closely with the Office of the National Coordinator for Health Information Technology (ONC) to ensure NLM's efforts are aligned with HHS Secretary Mike Leavitt and President Bush's goal for the nationwide implementation of an interoperable health information technology infrastructure to improve the quality and efficiency of health care.

Health Data Standards

NLM supports the development, enhancement, and distribution of clinically specific vocabularies to facilitate the exchange of clinical data and improve retrieval of health information.

Clinical Vocabularies supported, licensed, or developed by NLM:

SNOMED_CT | LOINC | RxNorm

• Uniform distribution mechanism for HIPAA and clinical vocabulary standards through the UMLS Metathesaurus

NLM coordinates efforts to develop mappings between HIPAA code sets and standard clinical vocabularies.

SNOMED CT to ICD-9-CM | SNOMED CT to CPT | LOINC to CPT ICD SNOMED CT to ICD-9-CM | SNOMED CT to CPT | LOINC to CPT

NLM promotes harmonization between standards.

 <u>Contract with HL7</u> to align HL7 message standard with CHI standard vocabularies and create implementation guides for exchange of entire EHRs.

System Development Tools

- UMLS Resources
- Language and Knowledge Processing
- Image Processing

Grants and Funding

Education and Training

- <u>Funded Support for Academic Training in Biomedical Informatics & Bioinformatics</u> Formal programs and individual fellowships are offered to assist medical informaticians in pursuit of a degree.
- <u>Medical Informatics Training Program</u> Support for visiting scientists and students to participate in research at the Lister Hill National Center for Biomedical Communications. <u>Medical Informatics Course at Woods Hole</u> sponsored by NLM and the Woods Hole Marine Biology Laboratory.

Key Reports Supported or Produced by NLM

- Ending the Document Game (2005)
- Networking Health: Prescriptions for the Internet (2000)
- Evaluating the Coverage of Controlled Health Data Terminologies: Report on the Results of the NLM/AHCPR Large Scale Vocabulary Test (1997)
- For the Record: Protecting Electronic Health Information (1997)
- Making a Powerful Connection (1995)

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Related Sites

<u>HealthIT</u> (U.S. Health Information Technology Web Site)

Go

Office of the National Coordinator for Health Information Technology (ONC)

NIH Initiatives

<u>NIH Re-engineering</u> <u>the Clinical Research</u> <u>Enterprise</u>

<u>CaBIG (Cancer</u> <u>Biomedical</u> <u>Informatics Grid)</u>

NLM Participation in Standards Development

Federal Health Architecture/Consolidated Health Informatics (EHA/CHI) (committee member)

National Committee on Vital and Health Statistics (NCVHS) (staff to Standards and Security subcommittee)

<u>Healthcare</u> <u>Information</u> <u>Technology</u> <u>Standards Panel</u> (<u>HITSP</u>) (board member)

Public Health Data Standards Consortium (PHDSC) (founding member)

Electronic Health Data Standards

- Include standard vocabularies
- Key element of health information technology infrastructure for:
 - Effective decision support
 - Safe, evidence-based, and coordinated health care
 - Cost-effective care
 - Increased/informed choice
 - More efficient clinical, public health, and health services research
 - Timely public health and bioterrorism surveillance

NLM-led Support for Development and Maintenance

- 1999 LOINC (lab tests/instrument observations) contract support
- 2002 RxNorm (clinical drugs) direct development
- **2003** SNOMED CT contract & license for U.S-wide use (as distributed by NLM in UMLS)
- 2004 NLM designated central coordinating body for clinical terminology standards within U.S. Dept. of Health and Human Services (HHS)
- 2006 Draft LOINC to CPT mapping distributed

SNOMED CT

CHI recommended standard
 Comprehensive clinical vocabulary
 Substantially increased Metathesaurus content

 Concepts: ~300K (+37K to Metathesaurus)
 Descriptions: ~737K (+350K to Metathesaurus)
 Relationships: 1.3M
 Generic drugs, History table

Updates

■ January SNOMED CT update → Spring UMLS
■ July SNOMED CT update → Fall UMLS

RxNorm

- CHI recommended standard
- Developed to address missed synonymy in UMLS clinical drugs
- Provides standardized (normalized) forms of U.S. clinical drugs
- Supports
 - effective sharing of drug data across systems
 - electronic health record (EHR)
 - computerized physician order entry (CPOE)



Link specific terms or codes between two sources

NLM given the responsibility for funding, coordinating, and/or performing official mappings between standard clinical terminologies and HIPAA code sets

Represented in Metathesaurus files MRMAP, MRSMAP

Mapping projects planned/underway

Clinical standards → HIPAA code sets:

- LOINC → CPT (Draft now available for testing!)
- **SNOMED CT** \rightarrow **ICD-9-CM**, **ICD-10-CM**
- **SNOMED** $CT \rightarrow CPT$

SNOMED CT → "other" vocabularies:

- International Classification of Functioning, Disability and Health (ICF)
- International Classification of Primary Care (ICPC)
- Medcin
- Medical Dictionary for Regulatory Affairs (MedDRA)
- Medical Subject Headings (MeSH®)
- Nursing Vocabularies (NIC, NOC, NANDA)

Key NLM Assumptions about Mapping

- Mapping is an R & D problem
- Iteration required to build highly functional maps
- Testing, validation, and use in real world settings
- Participants include producers of vocabularies on both ends, and prospective users, recipients of maps
- Mapping will prompt changes/corrections to vocabulary content and update schedules
- Mappings must be updated when either end is updated
- Mappings will be distributed in the UMLS (not exclusively)

Draft LNC215 to CPT2005 Mappings

Use case assumes

LOINC codes used to order lab tests, observations
 CPT codes used for billing (one-way)

2000+ most common mappings contributed

Created by Intermountain Healthcare
 Reviewed by Regenstrief Institute and AMA

Future developments to include:
 Radiology tests
 Document names (consults, progress notes)
 More Laboratory LOINC mappings
 Clinical findings (vital signs, height, weight)

Mapping examples – LOINC → CPT

LOINC Code

- 1795-4 AMYLASE:CCNC:PT:FLU:QN \rightarrow
- 1798-8 AMYLASE:CCNC:PT:SER:QN →
- 1799-6 AMYLASE:CCNC:PT:UR:QN →

CPT Code

82150 Amylase (blood or urine)

C1704201|LNC|ATX104051419| |1799-6|CODE|RN||<mark>82150</mark>|CODE||

C1704201|LNC|ATX104051418| |1798-8|CODE|RN||82150|CODE||

C1704201|LNC|ATX104051417| |1795-4|CODE|RN||82150|CODE||

LNC215_TO_CPT2005 MRSMAP

LNC215_TO_CPT2005 MRCONSO

C1704201 | ENG | P | L6107611 | PF | S7015223 | Y |A10924448 | | | | LNC | XM | NOCODE | LNC21 5 to CPT2005 Mappings |0|N | |

LNC215_TO_CPT2005 MRSMAP

C1704201|L6107611|S7015223|A10924448|AUI|NOCODE|...

ATX104051002||MAPSETNAME|LNC|LNC215 to CPT2005 Mappings|N|| ATX104051003||MAPSETTYPE|LNC|LOINC has associated CPT code|N|| ATX104051007||FROMVSAB|LNC|LNC215|N|| ATX104051008||TOVSAB|LNC|CPT2005|N||

• • •

ATX104051018||MAPSETVERSION|LNC|Sept 2006|N||



UMLS Knowledge Source Server (UMLSKS)

UMLS Knowledge Source Server

Liscensed users access online: <u>http://umlsks.nlm.nih.gov</u>

Web search interface for the three Knowledge Sources

Application Programming Interface (API)

Using the UMLSKS

- Create login ID and password using the form on the right
- Each UMLS license can have multiple login IDs
- NLM does not maintain a copy of the password or Login ID

Passwords may be reset, not login IDs

	wledge Source Server	(UMLSKS)
	Later Hill Natio	II.I. National Library of Medicine nal Center for Biomedical Communications (LHNCBC)
About the UNLSAS Log in Overview Frequently Asked Questions UMLS Metathesanrus® Semantic Network SPECIALIST Lexicon Deconnectation User's Guide Developer's Guide UMLS Documentation Set	Login ID: Login Password Login Password Login Porget your password? What's New The primary database server for the UMLSKS has been restored and users should now have access to all available UMLS releases. We're sorry for any inconvenience this may have cause. (September 22, 2006 10:00 ET) 2006AC UMLS release is available for searching and for download within the UMLSKS. For more information, click here. (July 31, 2006 13:45PM ET) On June 14, 2006 heensees who have out athorited their angel report (as	New UMLS Lisence #

UMLSKS Basic Concept Report

	UMLS Knowledge Source Server (UMLSKS) Metathesaurus Semantic Network SPECIALIST Lexicon
	Home Advanced Search Logout
Metathesaurus Search for: (Oryetolagus cuniculus in UbdLS Release 2006AC
Display Display All	
	Concept: Oryctolagus cuniculus
Concept	CUI: C0034493
 Definition 	Samuelia Tanan Menand
🗹 Synanyme	semantic type: Charlies
🗹 Other Languages	
🗹 Suppressible Synonyme	Definition:
M Sources	The species Oryctolagus cumculus, in the family Lepondae, order LAGOMORPHA. Rabbits are born in burrows, turiess, and with
Context	eyes and ears closed. In contrast with HARLS, rabbits have 22 chromosome pairs (<u>MeSH</u>)
M Ancestors	the of the model marine of the same Tanas among the common Freezone marine (Tanas amindus), and the common Amorica
Parents	Any or the similar species or the genus Lepos, especially the common European species (Lepos conclusis), and the common Americal subject (Lepos conclusion) for any other and the common Lepose and the common Americal
M Siblings	subject: systematic sector research manal. (Four December 1) [1992] The subject of the subject o
M Children	9
Relations Vorcent	Symonymes:
E Broader	Comparison and the
D Sailes	Diropean rabon
R Other	Amount Dabler
Related and search to	Relation Companying
manna an pornay	Sufficient Accounting

Includes:

Definitions Synonyms Other Languages Sources Context Information Relations Co-occurring Concepts Can be used to find: Source IDs Other Language Synonyms Term Type Information Term Variants

Metathesaurus Advanced Search Options

Focused Search

Search by release, source vocabulary or language Partial string matching with right or left truncation
XML Query Send Standard API commands or specialized requests Run against the UMLSKS, results returned in tab delimited text

Request relational records

View records by CUI for the files MRCONSO, MRDEF, MRSTY, MRCXT, MRREL, MRSAT, & MRCOC as tab delimited text



Semantic Network search

 Enter search term or select from drop down box
 View Type, TUI, and Definition
 Select checkboxes to see Meta Concepts, Relations And Raw Records

	Metathesaurus	Semantic Network	SPECIALIST Lexicon
ick here to view the Java SWING b	ased version of the Semantic N	letwoek beowsee.	
his is best viewed with Netsnape 6.3	canil above or IE 5 x and above	e	
	Semantic Net	work.	
Semantic Types: <u>Entity</u> , <u>Event</u>		Semant	ic Relations: isa, associated with
ease Enter a Semantic Type of	r Relation in the box below	or select from the list below and	click on the Find button:
0]	
Semantic Types:			
C Alga	-		
Semantic Relations:			
C adjacent_to 💌			
Find			
Fetter		Meta Concepts	ERAL Relations 🗆 Raw Records
Physical Object			
Physical Object Organism		Semantic Type: .	Jga
Physical Object Organism Plant		Semantic Type: J TUI: T003	Alga
Physical Object Organism Plant Alga		Semantic Type: J TUI: T003 Definition: A chia	Mga fly aquatic plant that contains
Physical Object Organism Plant Aliga Fungus		Semantic Type: A TUI: T003 Definition: A chin chlorophyll, but do	Mga fly aquatic plant that contains es not form embryos during
Physical Object Organism Plant Alga Fungus Varus		Semantic Type: / TUII: T003 Definition: A chir chlorophyti, but do development and I	Mga fly aquatic plant that contains es not form embryos during acks vascular tissue.

SPECIALIST Lexicon Search

Search by entering the complete term View record in relational or unit record format

UMLS Knowledge Source Server (UMLSKS)

MALE STATES STATES AND A STOLED AND A STOLED AND AND A STOLED AND AND A STOLED AND

Logout

Relational Records for "heart attack"

LRWD

heart|E0431577| attack|E0431577| attacks|E0431577|

UMLS Knowledge Source Server (UMLSKS)

S II I THESE TRANSFERRED DID DEDARE DIDLAC DODARE DIDLAR DODARE DIDLAR DIDLAR DIDHAR DIDHARE DIDLAR DIDLARE
Metathesaurus Semantic Network

SPECIALIST Lexicon

E0431577|heart attack|noun|count(thr_sing)|heart attack|heart atta E0431577|heart attacks|noun|count(thr_plur)|heart attack|heart att

LRTYP

LRAGR

E0431577|heart attack|noun|reg|

Specialist Lexical Record

{base=heart attack entry=E0431577 cat=noun variants=ceg

View the unit record for "heart attack"

View heart attack in relational format

Application Programming Interfaces

- Remote server at NLM
- Local application connected through

Java RMI

- Java-based applications
- Developer's Guide: Chapter 3
- Set of Java classes (part of the UMLSKS API download)
- Detailed Javadoc documentation online and with API download

TCP/IP socket

- XML-based queries
- Developer's Guide: Chapter 5
- XML schema
- Socket server
 - Host: umlsks.nlm.nih.gov
 - Port: 8042

You MUST register your IP Address! 65

New UMLSKS expected Spring 2007

Web services based
Create your own tabs
Forms auto-complete, learning as you go
Looking for beta testers

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
1 GB RAM recommended (512 MB min.)
6x (or better) DVD drive
13 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



How do 1?

Specify sources for a customized subset?



Sources to	Exclude				
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		
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		
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		
DSM-III-R, 1987	DSM3R_1987	DSM3R	ENG	3	
DSM-IV, 1994	DSM4_1994	DSM4	ENG	3	
DXplain, 1994	DXP94	DXP	ENG	0	
Gene Ontology, 2004_12_20	GO2004_12_20	GO	ENG	0	
HCPCS Version of Current Dental Terminology 2005 (CDT-5), 5	HCDT5	CDT	ENG	3	

Highlighted rows are excluded from the subset.

Extract an RxNorm subset?

 On the File menu select "Enable/Disable Filter"
 Selecting any filter opens a new tab for that filter.
 Make your selections on the tab for that filter



Input Optio	ns Output Opti	ons Source List	Precedence	Suppressibility	R×Norm Filter		
Restricts t	he subset to cor	ncepts that meet or	ne of the follow	ving criteria:			
c) Semant	a) KxNorm atoms, e.g., SCD c) Semantic Type = Drug Delivery Device.						
The RxNor	The RxNorm default subset includes only RxNorm concepts from sources at restriction level 0. Use the Source List tab to include or exclude						
RxNorm c	RxNorm concepts from other sources.						
	R×Norm Filter						
	ly RyNorm Filter						

Other filters allow users to remove terms: Attribute Type List Relationship Type List Semantic Types List Source Term Types

Create a custom database load script?

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

Input Options Output Options Source List Precedence Suppressibility

Select data output options to customize the subset and create additional files if desired. 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"

	₽ A	
	Output Format Options	
Select Output Format		
Rich Release Format		Browse
	Write Database Load Scripts	_
Write Oracle load script.	Write MySQL load script.	

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

Input Options | Output Options | Source Let 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.

18/hen you have made your selections on this tab, you may proceed to the other Ontions tabs in any order. If you are finished customizing

Precedence					
Full Source Name		Source Abbreviation		Term Type	
UMLS Metathesaurus		МТН	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		

Remove specific term types from subset?

Input Options | Output Options | Source List | Precedence Suppressibility

This screen contains the Suppressibility Filter, which spectres source/term two 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 Supp	ressible Term Types		
Source	Source Abbreviation	Term Type	
Al/RHEUM, 1993	AIR93	DI	
Al/RHEUM, 1993	AIR93	FI	
Al/RHEUM, 1993	AIR93	HT	
Al/RHEUM, 1993	AIR93	SY	

Select Suppressibility tab

Highlight rows to exclude

Select Options \rightarrow Advanced Suppressibility Options Select the term types to remove



Reset default MMSYS Options?

UMLS Metathesaurus Configuration 2004AB						
File	Edit	Options	Reset	Done		
		Res	et Input Options			
Input Options 0 Customize the inj When you have i		Reset Output Options		recedence		
		Res	et Source List			
		Reset Precedence		more morm		
		Res	et Suppressibility	b, you may pi		

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

👙 Rich Release Format Browser 2006AD C0004057		
File Edit Options		Help
Monospaced ▼ ▼	C:\Documents and Settings\kleinsr\Desktop\test\2006AD\META	•
CUI Search Tree Browser Word Search Enter search terms: (English) aspirin Search Select a concept. (1 result) C0004057 Aspirin	Raw View Report View • Concept: [CUI: C0004057] Aspirin • Semantic Types Organic Chemical Pharmacologic Substance • Definitions CSP/PT prototypical analgesic used in the treatment of mild to moderate pain; has antiinflammatory and antipyretic properties and acts as an inhibitor of cyclooxygenase which results in the inhibition of the biosynthesis of prostaglandins; aspirin also	

More information and help

MetamorphoSys Documentation at: 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

SPECIALIST Lexicon and Lexical Tools



MetaMap and MetaMap Transfer (MMTx)



Purpose of MetaMap/MMTx
The MetaMap/MMTx Algorithm
Availability
Demo

Purpose of MetaMap/MMTx

To map biomedical text to concepts in the UMLS Metathesaurus
 Or, equivalently, to find Metathesaurus concepts in text

MMTx was created to provide a distributable version of MetaMap

MetaMap/MMTx Example

Text: <u>Termination</u> of <u>clinical trials</u>: the <u>beta-blocker</u> <u>heart attack</u> <u>trial</u>...

Concepts

Termination *

Clinical Trials

Adrenergic beta-Antagonists

Myocardial Infarction

Heart attack (Myocardial Infarction)

Clinical Trials Trial (Clinical Trials)

Beta-blocker (Adrenergic beta-Antagonists)

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

Technical Details and Availability

<u>http://skr.nlm.nih.gov/</u>

Click 'Research Information' for technical details

<u>http://mmtx.nlm.nih.gov/</u>

Click 'Documentation' and 'Prerequisites'

Use restrictions

- Sign UMLS license agreement; then access MetaMap and download MMTx using UMLS ID/password
- Respect UMLS constituent vocabulary copyrights



http://skr.nlm.nih.gov

Example: normal processing

Phrase: "lung cancer."

Meta Candidates (8):
1000 Lung Cancer (Malignant neoplasm of lung) [Neoplastic Process]
1000 Lung Cancer (Carcinoma of lung) [Neoplastic Process]
861 Cancer (Malignant Neoplasms) [Neoplastic Process]
861 Lung [Body Part, Organ, or Organ Component]
861 Cancer (Cancer Genus) [Invertebrate]
861 Lung (Entire lung) [Body Part, Organ, or Organ Component]
861 Cancer (Specialty Type - cancer) [Biomedical Occupation or Discipline]
768 Pneumonia [Disease or Syndrome]

Meta Mapping (1000):

1000 Lung Cancer (Carcinoma of lung) [Neoplastic Process] Meta Mapping (1000):

1000 Lung Cancer (Malignant neoplasm of lung) [Neoplastic Process]

Example: Variants (-v)

Phrase: "lung cancer."

. . .

lung cancer [noun] variants (n=1): lung cancer{[noun], 0=[]}

lung [noun] variants (n=9): lung{[noun], 0=[]} lungs{[noun], 1="i"} pneumonia{[noun], 5="ds"} pneumoniae{[noun], 5="ds"} pneumonias{[noun], 5="ds"} pneumonic{[adj], 2="s"} pulmonal{[adj], 4="ss"} pulmonary{[adj], 2="s"} pulmonic{[adj], 2="s"}

cancer [noun] variants (n=4): cancer{[noun], 0=[]} cancerous{[adj], 3="d"} cancers{[noun], 1="i"} carcinomatous{[adj], 2="s"}

Example: Compound mappings

Phrase: "obstructive sleep apnea." Meta Candidates (8):

without --best_mappings_only

Meta Mapping (1000):

1000 Obstructive sleep apnoea (Sleep Apnea, Obstructive) [Disease or Syndrome]

Meta Mapping (901):

827 Obstructive (Obstructed) [Functional Concept]

901 Apnea, Sleep (Sleep Apnea Syndromes) [Disease or Syndrome] Meta Mapping (851):

827 Obstructive (Obstructed) [Functional Concept]

827 Sleep [Organism Function]

827 APNOEA (Apnea) [Pathologic Function]

•••

. . .

Example: show sources (-G)

Phrase: "scorpion sting."

Meta Candidates (4):
1000 Scorpion sting {MDR,DXP} [Injury or Poisoning]
861 Sting (Sting Injury {MTH,MSH,MDR,RCD,SNM,SNOMEDCT,SNMI,WHO}) [Injury or Poisoning]
694 Scorpion (Scorpions {LCH,MSH,MTH,SNM,SNOMEDCT,SNMI,CSP,RCD,NCBI}) [Invertebrate]
694 SCORPION (Scorpion antigen {MTH,LNC}) [Immunologic Factor]

Meta Mapping (1000): 1000 Scorpion sting {MDR,DXP} [Injury or Poisoning]

Example: restrict to sources (-GR LCH)

Phrase: "scorpion sting."

Meta Candidates (1): 694 Scorpion (Scorpions {LCH}) [Invertebrate]

Meta Mapping (694): 694 Scorpion (Scorpions {LCH}) [Invertebrate]

Example: restrict to STs (-J neop)

Phrase: "lung cancer."

Meta Candidates (3):

1000 Lung Cancer (Malignant neoplasm of lung) [Neoplastic Process]1000 Lung Cancer (Carcinoma of lung) [Neoplastic Process]861 Cancer (Malignant Neoplasms) [Neoplastic Process]

Meta Mapping (1000):

1000 Lung Cancer (Carcinoma of lung) [Neoplastic Process] Meta Mapping (1000):

1000 Lung Cancer (Malignant neoplasm of lung) [Neoplastic Process]

Questions?

UMLS Documentation and Support

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



Register: sign the license agreement
Create UMLSKS account
Explore Knowledge Sources
Download files or request DVD
Create subsets using MetamorphoSys

Center Drive

Library of Medicine

