Outline

- UMLS overview
- UMLS license agreement
- Accessing the Metathesaurus
- SNOMED CT® in the Metathesaurus
- Mappings
- RxNorm
What is the UMLS?

- Unified Medical Language System
- Objective: Help interpret and understand medical meaning across systems
- Overcome:
  - disparities in language
  - disparities in granularity and perspective
  - problems in mapping and aggregating information across systems
The UMLS consists of …

- **Metathesaurus**: Biomedical Concepts
- **Semantic Network**: Broad Categories + Relationships
- **SPECIALIST Lexicon & Lexical Tools**: Lexical information, language processing programs

… 3 Knowledge Sources used together or separately
Knowledge Sources are …

- Machine-readable data files (relational tables)
  - Records (rows)
  - Fields (columns)

- Applications
  - MetamorphoSys (installation and customization)
  - RRF Browser (view)
  - lvg (lexical programs)

... sets of data files and programs

the UMLS is not an end-user application
UML Uses

- Information retrieval
- Thesaurus construction
- Natural language processing
- Automated indexing
- Electronic health records (EHR)

- Distribution mechanism for U.S. vocabulary standards
License Agreement
First step  license agreement

Online Web-based license:

- 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
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License Restriction Levels 0-4

Level 0  (28.2%)  
- unrestricted

Level 1  (1.6%)  
- negotiate to translate

Level 2  (0.4%)  
- negotiate to use in health data creation

Level 3  (30.6%)  
- negotiate to use in production
- Explicitly prohibited to provide Internet access

Level 4  (39.2%)  
- unrestricted for U.S. use and distribution

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

Remote access
- UMLS Knowledge Source Server (UMLSKS)
- download files
- search and browse

Local access
- MetamorphoSys: install files locally, create customized Metathesaurus subsets
- RRF Browser: search, browse, view customized subsets
Remote Access UMLSKS

- UMLS Knowledge Source Server (UMLSKS):
- Web search interface

Returning users log in
New users create account with license #
UMLSKS Home Page

Tabs across top access basic searching of 3 Knowledge Sources

Advanced searching options on right-hand side
UMLSKS Basic Concept Report

Concept Name/CUI: Addison's disease
CUI: C0001403

Semantic Type(s): Disease or Syndrome

Definition:
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. (MeSH)

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. (CRISP Thesaurus)

Synonyms:
- Addison’s disease
- Addison’s disease (disorder)
- ADRENA L INSUFFICIENCY (ADDISON’S DISEASE)
- ADRENO CORTICAL INSUFFICIENCY, PRIMARY FAILURE
- Asthenia pigmentosa
- Bronzed disease
**MetamorphoSys**

- Multi-platform Java software with each UMLS release
- Installs Knowledge Sources to local storage
- Customizes a local Metathesaurus

Customization is critical and requires:
- Understanding of source vocabularies
- License arrangements
- Functional requirements, purpose and perspective
- Technical expertise
RRF Browser tree browser
UMLS 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, and the relationships among them.
Metathesaurus Sources

- Varying purposes, structures, properties
- “Sets of valid values” for data elements:
  - Thesauri, e.g., MeSH
  - Statistical Classifications, e.g., ICD
  - Billing Codes, e.g., CPT
  - Clinical coding systems, e.g., SNOMED
  - Lists of controlled terms
- Full list in Appendix to License Agreement

The UMLS serves as a vehicle for regulatory standards (HIPAA, CHI, PHIN)
Metathesaurus highlights

- Represents the meaning in each source
- Tags source information
- Adds context-free unique identifiers
  - “the name that never changes”
- Adheres to principle of “source transparency”
- Includes normalized word and string indexes produced using UMLS lexical tools
Metathesaurus organization

- **Concepts** (MRCONSO)
  - Synonymous terms are clustered into a concept
  - Additional information
    - From source (definitions, attributes)
    - Supplied by NLM (CUI, Semantic Type)

- **Relationships** (MRREL)
  - Concepts are related to other concepts
  - Relationship attributes (type, source of relationship)

- **Attributes** (MRSAT)
Metathesaurus

- Concept (1.2 M) CUI
  - Set of synonymous concept names

- Term (> 4.1 M) LUI
  - Set of normalized names

- String (> 4.7 M) SUI
  - Distinct concept name

- Atom (> 5.4 M) AUI
  - Concept name in a given source

<table>
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<tr>
<th>A1412439 headaches</th>
<th>(BI)</th>
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<tr>
<td>S1459113 headaches</td>
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<tr>
<td>A2882187 Headache (SNOMED)</td>
<td></td>
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<tr>
<td>A0066000 Headache (MeSH)</td>
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<td>S0046854 Headache</td>
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<tr>
<td>L0018681 headache</td>
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<tr>
<td>A1641293 Cranial Pain (MeSH)</td>
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<tr>
<td>S1680378 Cranial Pain</td>
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<tr>
<td>S0375902 HEAD PAIN CEPHALGIA</td>
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<td>L0290366 cephalgia head pain</td>
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<tr>
<td>C0018681 Headache</td>
<td></td>
</tr>
</tbody>
</table>
Relationships types

- Hierarchical
  - Parent / Child
  - Broader / Narrower than

- Associative: other

- Source asserted or editor-assigned
Relationships Examples: Hierarchical

**SNOMED Clinical Terms**

- Clinical finding
- Clinical history and observation findings
  - General finding of observation of patient
  - Symptom
    - Pain
      - Pain finding at anatomical site
        - Pain of head and neck region
          - Headache
Relationships Examples: Children

- Ocular headache
- Aural headache
- Nasal headache
- Posttraumatic headache
- Analgesic overuse headache
- Benign exertional headache
- Cough headache syndrome
- Generalized headache
- Headache due to cold exposure
Relationships: Associative

Other source relationships to Headache:

Facial Pain (SNOMED Clinical Terms)

Seizures (Clinical Problem Statements)
Sinusitis (Clinical Problem Statements)

Sinusitis (MedlinePlus)
Semantic Network categorizes

135 high-level categories (semantic types)

- **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 Network relations

- Relations define useful relations between types
- Types + Relations = Network

Biomedical Occupation or Discipline

- ASSOCIATED_WITH Social Behavior
- ISSUE_IN Biomedical Occupation or Discipline

Manufactured Object

- CAUSES Injury or Poisoning

Diagnostic Procedure

- USES Manufactured Object

Health Care Activity

- ISA Occupational Activity
- AFFECTS Disease or Syndrome
Core content U.S. English, Spanish
- Concepts, Descriptions, Relationships tables
- Generic drugs
- History table
- ICD-9-CM mapping

Not included:
- US, UK SNOMED branded drug extensions
- non-English, non-Spanish content
SNOMED CT in the Metathesaurus 2

- Different senses of concepts, synonyms, hierarchies

- UMLS concepts vs. SNOMED CT concepts
  - Useful level of distinction in biomedical practice

- UMLS hierarchies vs. SNOMED CT hierarchies
  - Single vs multiple hierarchies

- Occasional disagreement in synonymy and concepts
Different Concept Views 1

- Jan. 2005 data
- SNOMED CT differs from UMLS view (14%)
  - Most differences are intentional
  - Some undiscovered synonymy fixed via collegial working relationship
- SNOMED CT 2 concepts / UMLS 1 concept
- For example: substance / product
  - Antacid (substance)
  - Antacid (product)
"This patient is on acetaminophen."

"Tylenol contains acetaminophen."

Acetaminophen (product)

Acetaminophen 500 mg tablet (product)

Acetaminophen 80 mg suppository (product)

Acetaminophen (substance)

Is-a

Has-active ingredient
Representing and Resolving Differences

- Merging
  - Two or more SNOMED CT concepts merged into one UMLS concept
  - 38,485 SNOMED CT concepts involved (12.9%)

- Splitting
  - One or more SNOMED CT atoms split into different UMLS concepts
  - 3037 SNOMED CT synonyms in 1,970 concepts (0.7%)
SNOMED CT in the Metathesaurus

SNOMED CT January 2004 (English)
- Concepts: 298,090
- Descriptions: 736,946
- Relationships: 1,010,873

Represented in 2004AA UMLS Release as:
- Concepts: 37,089
- Descriptions: 350,464 (mostly fully-specified names e.g. Pneumonia (disorder))
### SNOMED CT Concepts Table:

<table>
<thead>
<tr>
<th>ConceptId</th>
<th>ConceptStatus</th>
<th>FullySpecifiedName</th>
<th>CTV3ID</th>
<th>SNOMEDID</th>
<th>IsPrimitive</th>
</tr>
</thead>
<tbody>
<tr>
<td>271737000</td>
<td>0</td>
<td>Anemia (disorder)</td>
<td>XM05A</td>
<td>DC-10009</td>
<td>1</td>
</tr>
</tbody>
</table>

### MRCONSO.RRF:

- CUI | AUI | SCUI | SAB | TTY | STR |
- C0002871 | A3597593 | 271737000 | SNOMEDCT | FN | Anemia (disorder) |
- C0002871 | A2878480 | 271737000 | SNOMEDCT | PT | Anemia |
- C0002871 | A2952250 | 271737000 | SNOMEDCT | SY | Absolute anemia |
- C0002871 | A3095181 | 271737000 | SNOMEDCT | PTGB | Anaemia |
- C0002871 | A3089808 | 271737000 | SNOMEDCT | SYGB | Absolute anaemia |
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
NLM and Health Data Standards

- Health Data Standards
  - Administrative (HIPAA)
  - Clinical (CHI)
  - Public Health (PHIN)

- NLM’s current contributions
  - Maintains UMLS as distribution mechanism for standards
  - Negotiated and maintains US license for SNOMED CT

- NLM’s evolving role
  - Reduce overlap
  - Establish and support mappings between standard clinical vocabularies (SNOMED, LOINC, RxNorm), and between other administrative and clinical standards
Mappings
Mappings

- Link concepts between two or more specific vocabularies

- Inter-terminology mappings facilitate reuse of information

- Source-asserted
  - SNOMED CT-NIC/NOC/NANDA (coming)
  - ICD-10-I CPC
Code to Code Mappings

- NLM mapping projects underway:
  - SNOMED CT to CPT/HCPCS
  - SNOMED CT to ICD-9-CM (specifically to support reimbursement)
  - LOINC to CPT

- Mapping projects being discussed:
  - SNOMED CT to MedDRA
  - SNOMED CT to MeSH
Mappings examples

- **LOINC - CPT**
  - LOINC
    - 1795-4 AMYLASE:CCNC:PT:FLU:QN
    - 1798-8 AMYLASE:CCNC:PT:SER:QN
    - 1799-6 AMYLASE:CCNC:PT:UR:QN
  - All map to CPT
    - 82150 Amylase (blood or urine)

- **SNOMED CT - CPT**
  - SNOMED CT
    - 313500004 Urine protein/creatinine ratio measurement
  - Maps to two CPT codes
    - 82570 Creatinine; other source
    - 84155 Protein, total, except by refractometry; serum
Key NLM Assumptions about Mapping

- Participants must include *producers* on both ends, and *users* of output
- Mappings must keep current at both ends
- Mappings will be distributed in the UMLS
- Mapping is an R & D problem; iteration is required to build highly functional maps
RxNorm
NLM’s RxNorm Project

- Addresses problems:
  - **missed synonymy** in UMLS clinical drugs
  - **patient safety**

- CHI recommended standard
  Designated standard for use in U.S. Federal Government systems for the electronic exchange of clinical health information

- Supports e-prescribing
RxNorm Source and Subset

**RxNorm Source**
- Normalized forms of clinical drugs
- Created by NLM
- Forms only from existing commercial and government clinical drug vocabularies

**RxNorm Subset**
- RxNorm forms from NLM
- Clinical drug content including SNOMED CT, First DataBank NDDF, FDA NDC

*Users must respect license restrictions*
RxNorm Normalized Forms

- Active standardized (generic) ingredients
- Strengths
- Units of measure
- Dosage form
- As administered to a patient
Creating RxNorm Concepts

- PALIFERMIN 6.25 MG INTRAVENOUS INJECTION, POWDER, LYOPHILIZED, FOR SOLUTION [KEPIVANCE]

- Standardize measurement and strength
  - Convert 6.25 MG to mg/ml
  - 5 mg/ml solution (per prescribing information)

- Identify brand name, base ingredient, dose form
  - Brand name (BN) = Kepivance
  - Base Ingredient (IN) = Palifermin
  - Dose Form = Injectable Solution

- SBD = Palifermin 5 mg/ml Injectable Solution [Kepivance]
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 concepts
RxNorm Nav

RxNorm Subset

- More than 8 full and partial sources
- Full sources
  - FDA, Medispan, Micromedex, NDDF, MDDB
  - More to come
- Partial sources
  - SNOMED CT, MeSH
- NOT in RxNorm
  - USP Medicare Model Guidelines

<table>
<thead>
<tr>
<th>RxNorm (NLM)</th>
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</thead>
<tbody>
<tr>
<td>SNOMED CT (CAP)</td>
</tr>
<tr>
<td>MTHFDA (FDA NDC)</td>
</tr>
<tr>
<td>VANDF (VA)</td>
</tr>
<tr>
<td>NDDF (FirstDataBank)</td>
</tr>
<tr>
<td>MDDB (Medispan)</td>
</tr>
<tr>
<td>MMX (Micromedex)</td>
</tr>
<tr>
<td>Multum</td>
</tr>
</tbody>
</table>
RxNorm Subset  2 ways

- Download RxNorm Source zip files
  - More frequent updates than UMLS
  - Monthly schedule to begin soon
  - Regular updates of individual sources coming

- Extract RxNorm Subset from UMLS
  - 3-4 releases per year
  - MetamorphoSys extracts preconfigured subset
  - Default setting includes Level 0 sources and SNOMED CT
  - Option to select specific sources
What’s Not in RxNorm

- Inactive ingredients (e.g., lactose, preservatives, alcohol)
- Pack size
- Flavors
RxNorm Development

Clinical Drugs from sources are released but RxNorm forms are not created:

- Radiopharmaceuticals
- Multi-vitamins
- More than 3 ingredients
- Drug Delivery Devices
Conclusion
Current UMLS Release

- UMLS 2005AB Release (June)
  - Metathesaurus
    - 133 sources including SNOMED CT, RxNorm
    - 1,196,265 concepts; 4,752,383 names
  - Semantic Network
  - SPECIALIST Lexicon
- Next release: 2005AC (November)
  - Updated sources include July 2005 SNOMED CT, MeSH 2006, RxNorm, ICD-9-CM 2006
  - New sources include HL7, PNDS
  - MetamorphoSys, Browser enhancements
Current RxNorm Release

August 2005 (7/29/05)
- Consistent with 2005AB UMLS Metathesaurus
- Updates: SNOMED CT (Jan. 2005), NDDF, VA NDF, NDF-RT

September 2005 (on or about 9/12)
- FDA NDC
Documentation and Support

- **UMLS homepage:**
  - links to all other UMLS information

- **UMLSKS homepage:**
  - links to the User’s and Developer’s guides

- **UMLSUSERS-L**
  - subscribe to discussion list

- **NLM Customer Service:**
  - custserv@nlm.nih.gov
Additional training

- UMLS Basics
- NLM
- American Medical Informatics Association (AMIA) tutorials
- Medical Library Association (MLA) CE class
- American Health Information Management Association (AHIMA)
- More SNOMED CT content coming
Thank you!