Medical Data, Standard Vocabularies, Communication Standards

6.872/HST950

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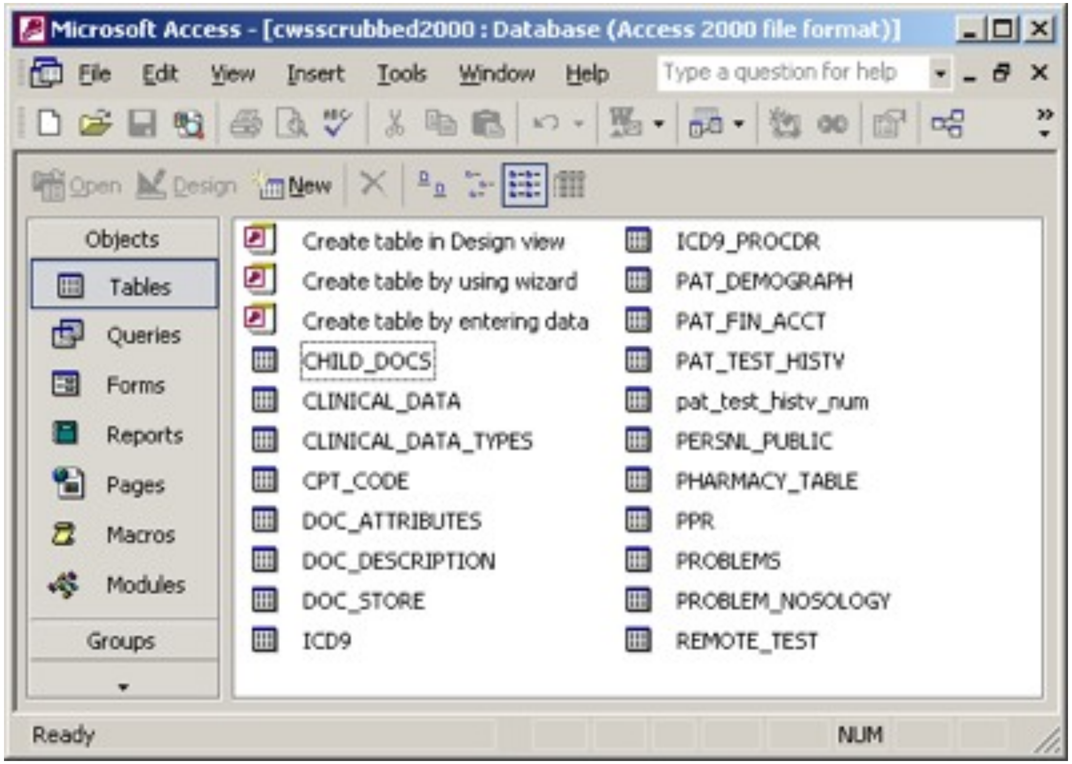
(with some material from Chris Cimino)

Recall Children’s Clinicians’ Workstation Database

- Demographics
- Problems
- Allergies
- Medications
  - Immunizations
- Lab Data
- Clinical Measurements
  - Growth Charts
- Visit History
- Reports and Letters

The Database



Create table in Design view
Create table by using wizard
Create table by entering data
CHILD_DOCS
CLINICAL_DATA
CLINICAL_DATA_TYPES
CPT_CODE
DOC_ATTRIBUTES
DOC_DESCRIPTION
DOC_STORE
ICD9
ICD9_PROCDR
PAT_DEMOGRAPH
PAT_FIN_ACCT
PAT_TEST_HISTV
pat_test_histv_num
PERSNL_PUBLIC
PHARMACY_TABLE
PPR
PROBLEMS
PROBLEM_NOSOLOGY
REMOTE_TEST

- Documents
    - DOC_STORE
    - DOC_ATTRIBUTES
    - DOC_DESCRIPTION
    - CHILD_DOCS
- Doctors
    - PERSNL_PUBLIC
    - PPR
- Patients
    - PAT_DEMOGRAPH
    - PAT_FIN_ACCT
    - PAT_TEST_HISTV
    - REMOTE_TEST
    - PHARMACY_TABLE
    - PROBLEMS

- General Information
    - CPT_CODE
    - ICD9
    - ICD9_PROCDR
    - PROBLEM_NOSOLOGY

Vocabularies and Terminology

- Why?
  - Surrogate for “messy reality”
  - Uses
- How?
  - Flat list
  - Taxonomy (Hierarchy, Nosology, …)
  - Heterarchy
  - Combinatorial Language
    - Derivation rules
    - Inference
    - … knowledge representation

“Ontology” for Computer Folks

- An organization of concepts (hierarchy or heterarchy)
- (Some) concepts are defined in terms of others
  - A *triangle* is a *polygon* with exactly 3 *sides*
  - A *dachshund* is a *dog* (with ???)
- Automatic classification
  - If P is a 3-sided polygon with …, it is recognized automatically as a triangle

OWL: “Semantic Web”

- *Description Logic*
  - Concepts and Instances
  - Is-a in virtue of
    - Primitive assertion: “A dog is a mammal.”
    - Definition: “A triangle is a three-sided polygon.”
  - Limited logical power of definition language assures tractable inference
    - Slot restriction
    - Number restriction
    - But, no negation or disjunction
  - Subsumption inferences are central
  - Other logical assertions may be made, but are typically not enforced or utilized in DL

Definitions

- Word – a set of characters including punctuation delimited by white space.
- Term – one or more words used as a unit.
- Concept – an idea, action, or thing.
- Synonym – two terms for the same concept.

Vocabulary Uses

- Indexing – Finding what you want
- Cataloging – Putting away what you have
  - E.g., WHO, DRGs
- Knowledge Representation
  - Representing the facts
  - Blurring the facts
  - Creating new shades of meaning

Describe a term for a Laboratory Test

- Where was it done?
- How was it done?
- Under what conditions was it done?
- How many minutes after eating carbohydrate was it measured?

Describe a Vocabulary for a Gene

- Whose gene?
- Gene fragment?
- Open Reading Frame?
- Promoter + all exons and introns
- Promoter + all exons + all introns + other binding sites affecting function?
- Final/draft/species/SNP/Alternative splicing?

Knowledge vs. Language

- Get two or more people to enumerate terms to describe the same set.
  - Do any terms match exactly?
  - Do terms differ by word order?
  - Do terms differ by word suffix or prefix?
  - Are there terms that some people think are synonyms that other people think are not?

History of 3 Vocabularies

- MeSH — Index
- ICD — Precoodinated
- SNOMED — Post-coordinated

History

- The modern history of medical controlled vocabularies begins with the U.S. Army General Surgeon who petitioned Congress to fund a medical library. (~Civil War)
- The position eventually became “The US Surgeon General” and the library the National Library of Medicine
  - http://www.nlm.nih.gov/

History

- Library collection was indexed with Index Medicus (created by NLM) which is published in book form.
- Index Medicus was extended to index medical literature articles.
- Index Medicus was extended further to provide on-line indexing (1960). This became the Medical Subject Headings (MeSH).

MeSH

- Purpose is to index the medical literature.
- Content of MeSH is driven by publications.
- Who “owns” MeSH?
- What impact do vocabulary changes have?

MeSH – Structure

http://www.nlm.nih.gov/mesh/

- MeSH is organized into a series of “trees”. (e.g. physical findings, diseases, chemicals)
- A MeSH main heading is a “concept”. (e.g. “Neurologic Disease”, “Epilepsy”)
- Main Heading (MH) is often called a term. (Try to avoid doing this.)

MeSH – Structure

- Each MH has a unique identifier.
- Each MH may have multiple synonyms.
- Each MH may have multiple locations in multiple trees. Each of these “contexts” has a unique tree address. The concept of “context” is synonymous with “multiple inheritance”.

MeSH – Structure

- There is a small set of subheadings (50) that “modify” MH based on tree address. (e.g. “diagnosis” applies to MH in the “Disease” tree but not to the “Chemical” tree).
- There is a small set of tag terms (15) which exist unrelated to the rest of MeSH. (e.g., “Review Article”, “Human”, “Animal”)

MeSH – Structure

- Every article is indexed with tag terms.
- Every article is indexed with MH terms for focus (main index term) and mention (minor index term).
- Every index term is checked for subheadings.
- This is all done by trained reviewers.
- The MeSH Vocabulary is revised annually.

http://www.nlm.nih.gov/mesh/

MESH Redux—
The Genome “Ontology”

biological process

molecular function

cell components

Image showing genome ontology removed due to copyright restrictions.

International Classification of Disease (ICD)

- Any agency that dispenses funds for health care needs a way to assess needs and effectiveness.
- The United Nations World Health Organization (WHO) funds health care prevention projects world wide and gathers statistics for member nations.
- Who “owns” ICD?
- What impact will changes have?

ICD – Structure

- ICD is divided into categories based on a 5-digit numeric code. (e.g., “133.21”)
- Usually round numbers are more general concepts (e.g., “100” subsumes “130” which subsumes “133”)
- The fourth and fifth digit is called a modifier but it isn’t really.

ICD – Structure

- The code is both the concept and the unique identifier. Multiple terms are linked to the same code.
- Every patient is coded with as many terms as possible.
- Terms should be the most specific one to describe a particular problem.

ICD – Structure

- Coding scheme limits the size of the vocabulary.
- Obsolete codes must be reused.
- Base ten results in limited flexibility and the need for “other”, “NOS”, and “NOC” terms.

ICD – Structure

- Lack of multiple contexts or multiple inheritance results in duplicate terms.
- Lack of overall organization results in ambiguous terms.

ICD – Structure

- ICD has been adopted by most insurance companies as a method for controlling billing and payment.
- Economic forces drive how the vocabulary is used which drives how ICD is modified which drives changes in reimbursement which drives how the vocabulary is used…
- Who “owns” ICD?
- The Vocabulary is revised sporadically.

SNOMED – Structure

- Developed by the American College of Pathologists to overcome the faults of ICD.
- Really describes 6 [now 12] different vocabularies, one for each “axis” of a concept (e.g., anatomy, environment, history).
- Every concept is built up from a term from each “axis” (e.g., “surgery of” “blue” “nevus” “of left” “forearm”).

SNOMED – Structure

- There is some overlap of the axes so it is possible to form two different versions for the same concept (e.g., “blue nevus” “nevus colored blue”).
- There are few rules for how to combine axes terms so it is possible to form valid nonsense terms (e.g., “nevus” “of left” “esophagus”).
- Who “owns” SNOMED?

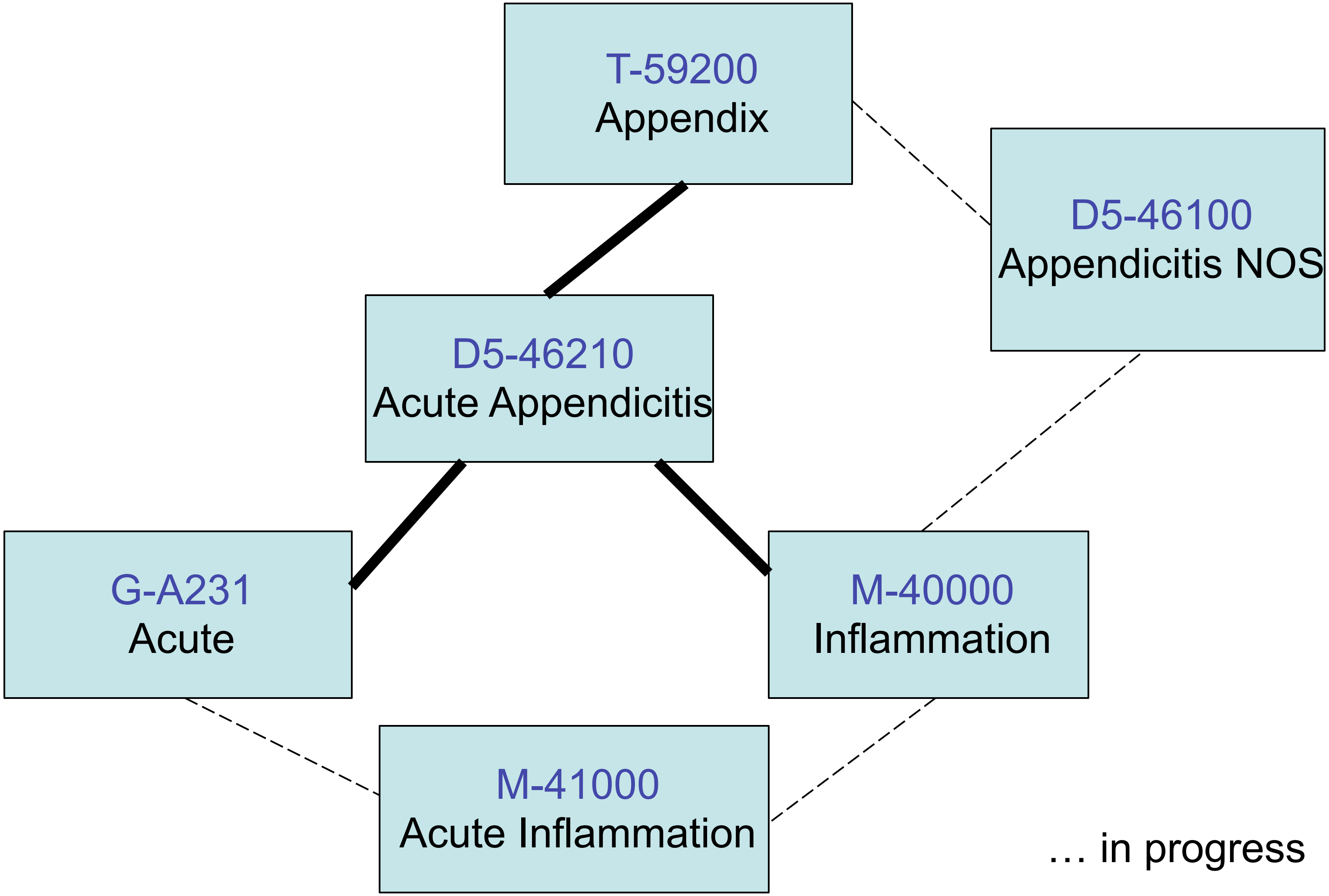
SNOMED Axes

- D – Diseases
- C – Drugs
- F – Function
- L – Living Organisms
- X – Manufacturers
- G – Modifiers
- M – Morphology
- J – Occupations
- A – Physical Agents
- P – Procedures
- S – Social Context
- T -- Topography

“Postcoordination”

- D5-46210 Acute Appendicitis
- G-A231 Acute, D5-46100 Appendicitis NOS
- G-A231 Acute, M-40000 Inflammation, T-59200 Appendix
- M-41000 Acute Inflammation, T-59200 Appendix
- T-59200 Appendix, M-41000 Acute Inflammation

Semantics of Postcoordination



… in progress

SNOMED-CT (Clinical Terminology)

- Combined SNOMED + Reed Codes
  - – SNOMED for diseases
  - – Reed for symptoms
- Licensed by NLM for anyone in US to use royalty-free
  - – Attempt to encourage standardization
  - – New international organization to maintain it

History

- For everyone who wants to “own” a medical vocabulary, there is a set of terms which are likely to overlap but be inconsistent with every other vocabulary.
- Read, CPT, COSTART, ChemAbstracts, …
- In theory they are all describing agreed upon concepts. A single standard vocabulary would improve the automated flow of medical information.

Ideal Vocabulary

?

Ideal Vocabulary

- Boundary
- Organization
- Completeness
- Absence of ambiguity
- Growth
- Aging

Ideal Vocabulary – Definitions

- String – a unique sequence of characters. The same set of characters may represent different concepts.
- Lexical variants – synonyms with “minor” differences. Word order, capitalization, and punctuation are usually included. Suffixes (plural) and prefixes may be included.
- One man’s lexical variant is another’s synonym.

Ideal Vocabulary – Definitions

- Related terms – distinct terms whose concepts overlap in some way. The most used relations are “broader” and “narrower” (e.g., “Neurologic Disease” includes but is broader than “Epilepsy”.)
- One man’s related term is another’s synonym.

Ideal Vocabulary – Definitions

- Controlled versus “free” text
  - Freedom of expression
  - Automatic indexing accuracy
- Atomic versus enumerated (Pre vs Post)
  - Handle the unexpected
  - Predict what to expect
- Definitions
  - “Free” text versus semantic

Unified Medical Language

- The Unified Medical Language System (UMLS) started as an NLM collaborative program with 7 centers around the country.
- Early years:
  - Explore ideas (1986)
  - One “winner” selected and developed (1988)
  - Usage Testing (1991)

UMLS – Structure

- Three components
  - Metathesaurus (META)
  - Semantic Network
  - Information Sources Map (ISM)
    - dropped
  - Specialist Lexicon

META – Structure

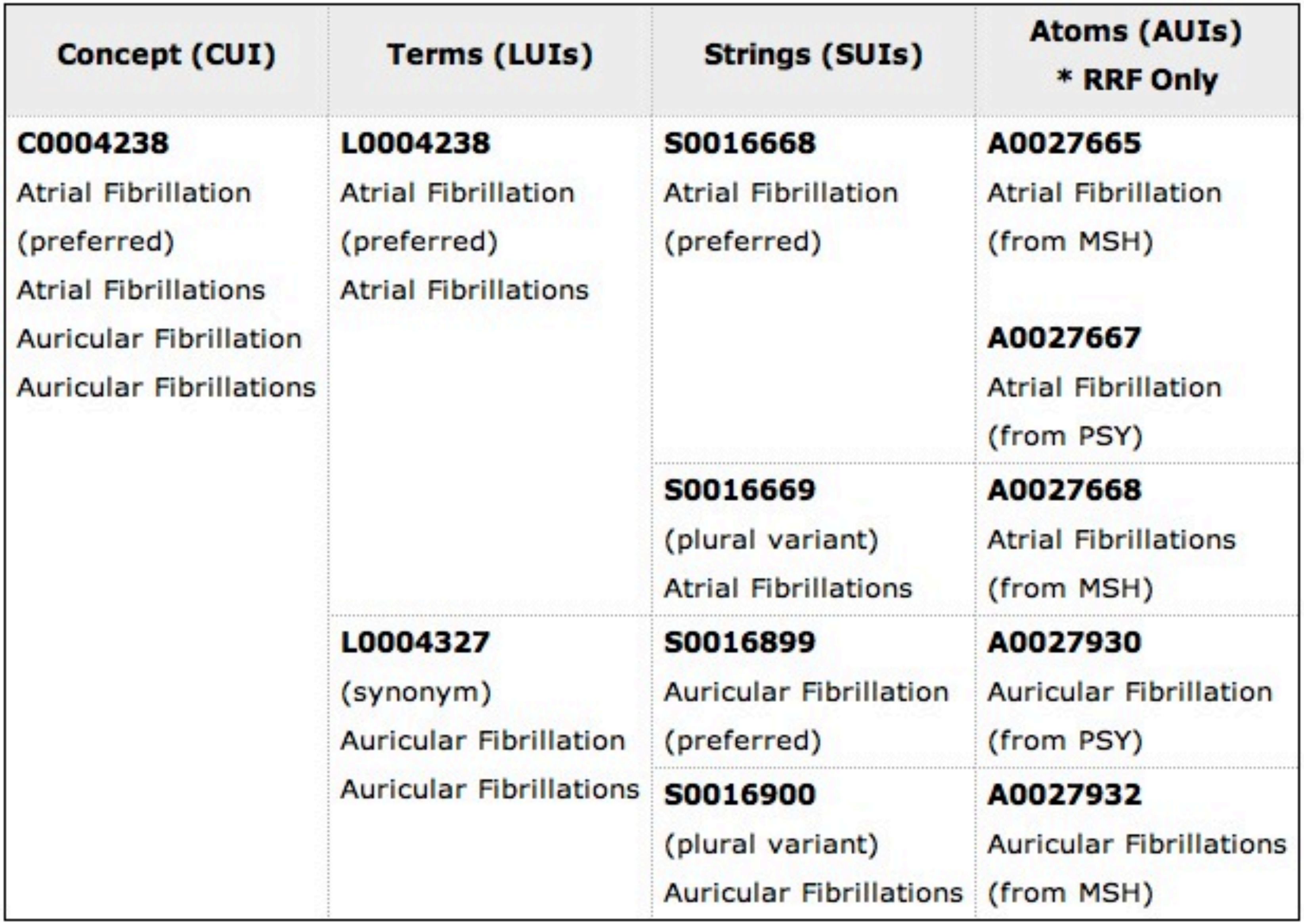
- NOT a controlled vocabulary.
- Database of information about other controlled vocabularies.
- Contains sufficient info to recreate most of the component vocabularies.
- Basic unit is the concept. A concept is linked to multiple strings from multiple vocabularies.

META - Structure

- Each concept-string pair is either a preferred term, synonym, or lexical variant.
- The same string may be linked to multiple concepts but a term, synonym, or lexical variant will only link to one concept each.
- Other links exist based solely on the existence of those links in a source vocabulary.

META – Structure

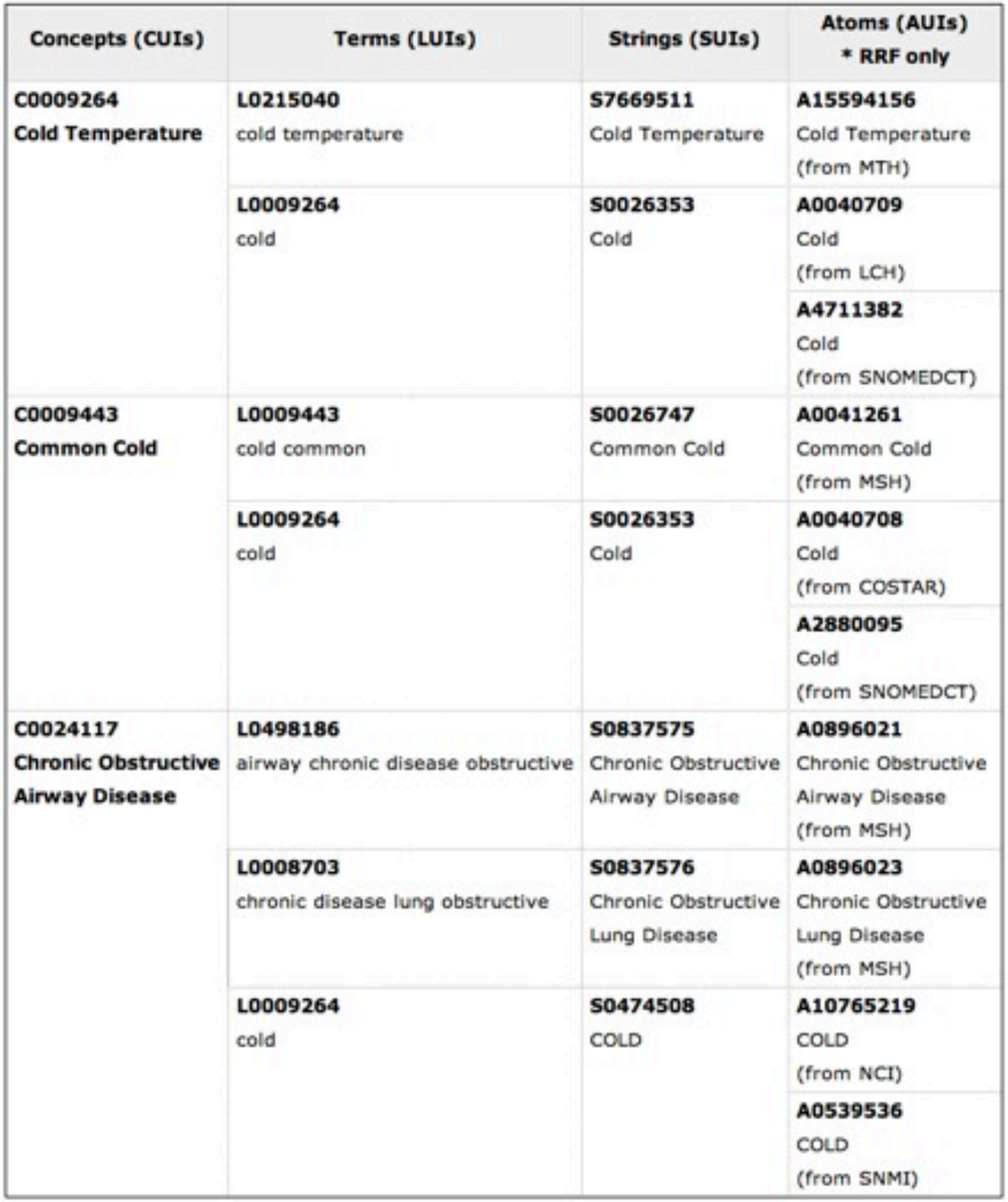
- Each concept has only one preferred term chosen from all linked terms based on order of precedence of source vocabularies. With a few exceptions, MeSH is number one.
- Each concept is linked to semantic types in the semantic network.
- NOT a controlled vocabulary – or is it?



Concept (CUI)	Terms (LUIs)	Strings (SUIs)	Atoms (AUIs) * RRF Only
C0004238 Atrial Fibrillation (preferred) Atrial Fibrillations Auricular Fibrillation Auricular Fibrillations	**L0004238** Atrial Fibrillation (preferred) Atrial Fibrillations	**S0016668** Atrial Fibrillation (preferred)	**A0027665** Atrial Fibrillation (from MSH) **A0027667** Atrial Fibrillation (from PSY)
		S0016669 (plural variant) Atrial Fibrillations	**A0027668** Atrial Fibrillations (from MSH)
	L0004327 (synonym) Auricular Fibrillation Auricular Fibrillations	**S0016899** Auricular Fibrillation (preferred)	**A0027930** Auricular Fibrillation (from PSY)
		S0016900 (plural variant) Auricular Fibrillations	**A0027932** Auricular Fibrillations (from MSH)

http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=nlmumls&part=ch02

From UMLS Reference Manual, National Library of Medicine, 2009.



Concepts (CUIs)	Terms (LUIs)	Strings (SUIs)	Atoms (AUIs) * RRF only
C0009264 **Cold Temperature**	**L0215040** cold temperature	**S7669511** Cold Temperature	**A15594156** Cold Temperature (from MTH)
	L0009264 cold	**S0026353** Cold	**A0040709** Cold (from LCH)
			A4711382 Cold (from SNOMEDCT)
C0009443 **Common Cold**	**L0009443** cold common	**S0026747** Common Cold	**A0041261** Common Cold (from MSH)
	L0009264 cold	**S0026353** Cold	**A0040708** Cold (from COSTAR)
			A2880095 Cold (from SNOMEDCT)
C0024117 **Chronic Obstructive Airway Disease**	**L0498186** airway chronic disease obstructive	**S0837575** Chronic Obstructive Airway Disease	**A0896021** Chronic Obstructive Airway Disease (from MSH)
	L0008703 chronic disease lung obstructive	**S0837576** Chronic Obstructive Lung Disease	**A0896023** Chronic Obstructive Lung Disease (from MSH)
	L0009264 cold	**S0474508** COLD	**A10765219** COLD (from NCI)
			A0539536 COLD (from SNMI)

From UMLS Reference Manual, National Library of Medicine, 2009.

Semantic Network – Structure

- Small vocabulary that attempts to implement an ideal vocabulary
- Terms defined with free text definitions and by linkage.

UMLS Semantic Network

"T001" "Organism" "A1.1"

"Generally, a living individual, including all plants and animals."

"Homozygote; Radiation Chimera; Sporocyst“

"T002" "Plant" "A1.1.1"

"An organism having cellulose cell walls, growing by synthesis of inorganic substances, generally distinguished by the presence of chlorophyll, and lacking the power of locomotion. Plant parts are included here as well."

"Pollen; Potatoes; Vegetables"

"T003" "Alga" "A1.1.1.1"

"A chiefly aquatic plant that contains chlorophyll, but does not form embryos during development and lacks vascular tissue."

"Chlorella; Laminaria; Seaweed“

… 188 terms

UMLS Sem Net: Relations

H: isa

R: associated_with

R1: physically_related_to

R1.1: part_of

R1.2: consists_of

R1.3: contains

R1.4: connected_to

R1.5: interconnects

R1.6: branch_of

R1.7: tributary_of

R1.8: ingredient_of

R2: spatially_related_to

R2.1: location_of

R2.2: adjacent_to

R2.3: surrounds

R2.4: traverses

R3: functionally_related_to

R3.1: affects

R3.1.1: manages

R3.1.2: treats

R3.1.3: disrupts

R3.1.4: complicates

R3.1.5: interacts_with

R3.1.6: prevents

R3.2: brings_about

R3.2.1: produces

R3.2.2: causes

R3.3: performs

R3.3.1: carries_out

R3.3.2: exhibits

R3.3.3: practices

R3.4: occurs_in

R3.4.1: process_of

R3.5: uses

R3.6: manifestation_of

R3.7: indicates

R3.8: result_of

R4: temporally_related_to

R4.1: co-occurs_with

R4.2: precedes

R5: conceptually_related_to

R5.1: evaluation_of

R5.10: method_of

R5.11: conceptual_part_of

R5.12: issue_in

R5.2: degree_of

R5.3: analyzes

R5.3.1: assesses_effect_of

R5.4: measurement_of

R5.5: measures

R5.6: diagnoses

R5.7: property_of

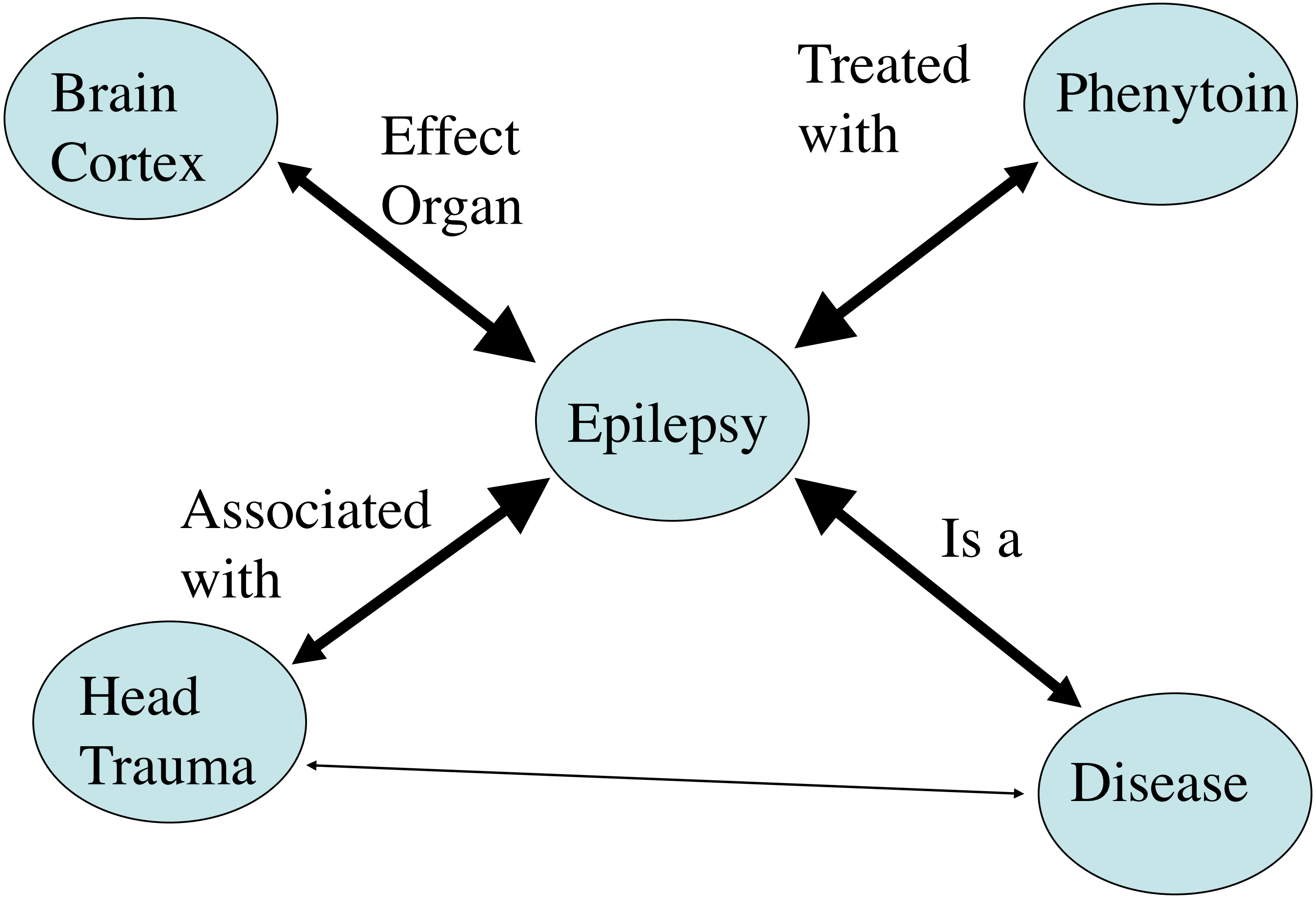
R5.8: derivative_of

R5.9: developmental_form_of

Definition of relations

```
("RL" "T132" "physically_related_to" "R1"
 "Related by virtue of some physical attribute or characteristic." "" "" ""
 "PR" "physically_related_to")
("RL" "T133" "part_of" "R1.1"
 "Composes, with one or more other physical units, some larger whole.  This includes component of,
      division of, portion of, fragment of, section of, and layer of."
 "" "" "" "PT" "has_part")
("RL" "T134" "contains" "R1.3"
 "Holds or is the receptacle for fluids or other substances.  This includes is filled with, holds, and is
      occupied by."
 "" "" "" "CT" "contained_in")
("RL" "T135" "location_of" "R2.1"
 "The position, site, or region of an entity or the site of a process." "" ""
 "" "LO" "has_location")
("RL" "T136" "temporally_related_to" "R4"
 "Related in time by preceding, co-occuring with, or following." "" "" "" "TR"
 "temporally_related_to")
("RL" "T137" "co-occurs_with" "R4.1"
 "Occurs at the same time as, together with, or jointly.  This includes is co-incident with, is concurrent
      with, is contemporaneous with, accompanies, coexists with, and is concomitant with."
 "" "" "" "CW" "co-occurs_with")
```

Semantic Network



State of the Art

- UMLS is not sufficient.
  - META is not complete. Still weak for clinical terms (sign and symptom terms).
  - META has superficial organization. Links between vocabularies is based primarily on lexical matches. Inter-vocabulary links growing slower than total size.
  - Ambiguous sources mean META is ambiguous.

State of the Art

- Semantic typing does scale up so META and the semantic network can form a starting point.
- Semantic rules are being added to SNOMED which may remove its ambiguity problem. This would greatly strengthen SNOMED and META.
- Who “owns” the rules?

State of the Art

- Semantic tools are being developed to provide end user management of vocabularies. The same tools would allow users to add (or nominate) new terms and help the user understand the semantic definition of existing terms.
- Links back to META allow institutions to “own” a vocabulary while complying with other organizations’ requirements.

Representation Languages

- Capabilities
  - Naming: intensional as well as extensional
    - E.g., “people who provide me healthcare”
  - Definitions
  - Assertions
- Examples
  - K-Rep
  - GALEN and GRAIL
  - W3C: RDF, OWL, …

Communication Standards

- HL7, DICOM, CorbaMED, XML-based …
- HL7:
  - Messages (unit of transfer, type=“purpose”)
    - Segments (e.g., header, event-type, pat. id, …)
      - Fields (character string)
- HL7 Details
  - Optional and repeated fields
  - Character encoding:
    - <cr> Segment separator
    - | Field separator
    - ^ Component separator
    - & Subcomponent separator
    - ~ Repetition separator

HL7 Examples

- AD – Address

Components: <street address (ST)> ^ < other designation (ST)> ^ <city (ST)> ^ <state or province (ST)> ^ <zip or postal code (ST)> ^ <country (ID)> ^ <address type (ID)> ^ <other geographic designation (ST)>

|10 ASH LN^#3^LIMA^OH^48132-|

- CE – Coded Element

Components: <identifier (ST)> ^ <text (ST)> ^ <name of coding system (ST)> ^ <alternate identifier (ST)> ^ <alternate text (ST)> ^ <name of alternate coding system (ST)>

|F-11380^CREATININE^I9^2148-5^CREATININE^LN|

HL7 (3.0) Reference Implementation Model

- Attempt at complete specification of health-care related *communications* (and, by derivation, *records*)
- Top-level types:
  - Entities
  - Roles
  - Acts
  - Infrastructure (communications)
  - Other
- http://www.hl7.org/ (see RIM documents)

HST.950J / 6.872 Biomedical Computing
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