Health Facts Boot Camp

March 13, 2017

Clinical Terminologies from the Research Perspective

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Learning objectives

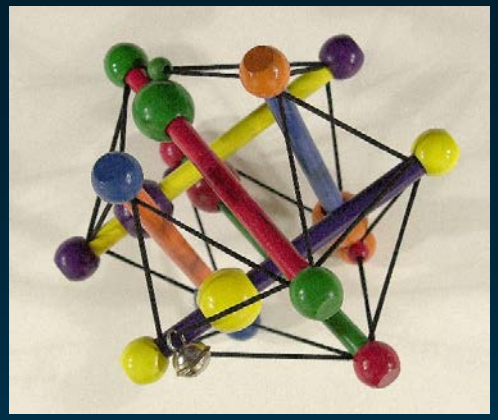
- Describe the history of biomedical ontologies
- List and describe the main biomedical ontologies used in 21st century healthcare
- Discuss the purpose of biomedical ontologies in knowledge management, clinical decision support and analytics

References Review articles

- Bodenreider O, Stevens R. Bio-ontologies: current trends and future directions. Brief Bioinform. 2006 Sep;7(3):256-74.
- Cimino JJ, Zhu X. The practical impact of ontologies on biomedical informatics. Yearb Med Inform. 2006:124-35.
- Bodenreider O. Biomedical ontologies in action: role in knowledge management, data integration and decision support. Yearb Med Inform. 2008:67-79.

Additional references

- Cimino JJ. Desiderata for controlled medical vocabularies in the twenty-first century. Methods Inf Med. 1998 Nov;37(4-5):394-403.
- Bodenreider O. The Unified Medical Language System (UMLS): integrating biomedical terminology. Nucleic Acids Res. 2004 Jan 1;32(Database issue):D267-70.



Medical Ontology Research

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Outline

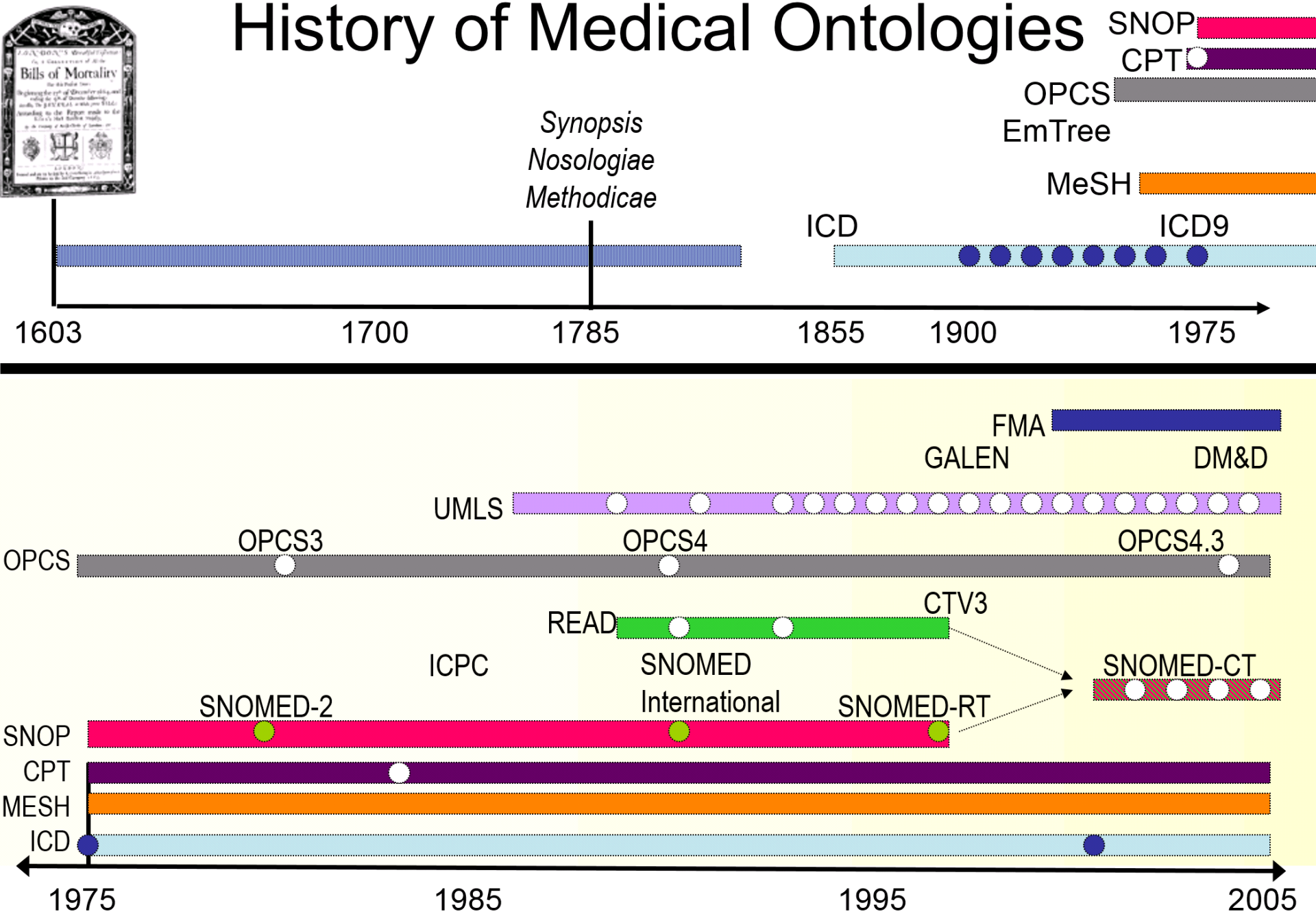
- (Brief) Historical perspective
- "High-Impact" Biomedical Ontologies
  - Structural perspective
- Biomedical Ontologies "in Action"
  - Functional perspective

Clinical Terminologies
Part 1

Historical perspective

[Bodenreider, BIB 2006]

History of Medical Ontologies



Clinical Terminologies
Part 2

"High-Impact" Biomedical Ontologies

A Structural Perspective

Overview

- Structural perspective
  - What are they (vs. what are they for)?
- "High-impact" biomedical ontologies [J. Cimino, YBMI 2006]
  - International Classification of Diseases (ICD)
  - Logical Observation Identifiers, Names and Codes (LOINC)
  - SNOMED Clinical Terms
  - Foundational Model of Anatomy
  - Gene Ontology
  - RxNorm
  - Medical Subject Headings (MeSH)
  - NCI Thesaurus
  - Unified Medical Language System (UMLS)

International Classification of Diseases

ICD Characteristics (1)

- Current version: ICD-10 (2016)
  - Annual updates
- Type: Classification
- Domain: Disorders
- Developer: World Health Organization (WHO)
- Funding: WHO
- Publicly available: Yes
- Used for: Mortality and morbidity statistics worldwide
- URL: http://www.who.int/classifications/icd/en/

ICD Characteristics (2)

- Number of
  - Concepts: 12,320 (ICD-10, 2004)
  - Terms: 1 per concept (tabular)
- Major organizing principles:
  - Tree (single inheritance hierarchy)
  - No explicit classification criteria
    - Idiosyncratic inclusion/exclusion mechanism
  - .8 slots for *Not elsewhere classified* (NEC)
  - .9 slots for *Not otherwise specified* (NOS)
- Specific coding rules
- Distribution: Proprietary format

http://apps.who.int/classifications/icd10/browse/

ICD Top level

ICD-10 Version:2016

- I Certain infectious and parasitic diseases
- II Neoplasms
- III Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
- IV Endocrine, nutritional and metabolic diseases
- V Mental and behavioural disorders
- VI Diseases of the nervous system
- VII Diseases of the eye and adnexa
- VIII Diseases of the ear and mastoid process
- IX Diseases of the circulatory system
- X Diseases of the respiratory system
- XI Diseases of the digestive system
- XII Diseases of the skin and subcutaneous tissue
- XIII Diseases of the musculoskeletal system and connective tissue
- XIV Diseases of the genitourinary system
- XV Pregnancy, childbirth and the puerperium
- XVI Certain conditions originating in the perinatal period
- XVII Congenital malformations, deformations and chromosomal abnormalities
- XVIII Symptoms, signs and abnormal clinical and laboratory findings, not elsewhere classified
- XIX Injury, poisoning and certain other consequences of external causes
- XX External causes of morbidity and mortality
- XXI Factors influencing health status and contact with health services
- XXII Codes for special purposes

ICD Example

- Idiosyncratic inclusion/exclusion criteria

E10 **Type 1 diabetes mellitus**

[See before E10 for subdivisions]

Incl.: diabetes (mellitus):
- brittle
- juvenile-onset
- ketosis-prone

Excl.: diabetes mellitus (in):
- malnutrition-related (E12.-)
- neonatal (P70.2)
- pregnancy, childbirth and the puerperium (O24.-)

glycosuria:
- NOS (R81)
- renal (E74.8)

impaired glucose tolerance (R73.0)

postsurgical hypoinsulinaemia (E89.1)

ICD Example

- *Not elsewhere classified* (NEC)
- *Not otherwise specified* (NOS)

E84	**Cystic fibrosis**
	Incl.: mucoviscidosis
E84.0	**Cystic fibrosis with pulmonary manifestations**
E84.1	**Cystic fibrosis with intestinal manifestations**
	Distal intestinal obstruction syndrome
	Meconium ileus in cystic fibrosis† (P75*)
	Excl.: meconium obstruction (ileus) in cases where cystic fibrosis is known not to be present (P76.0)
E84.8	**Cystic fibrosis with other manifestations**
E84.9	**Cystic fibrosis, unspecified**

ICD-10-CM

- Derived from: ICD-10
  - Finer-grained (both clinically and administratively)
- Type: Classification
  - 92,042 codes (2015)
  - Terms: 1.2 per concept
- Domain: Disorders
- Developer: National Center for Health Statistics (NCVHS)
- Funding: U.S. Government
- Publicly available: Yes
- Used for: Billing
- URL: http://www.cdc.gov/nchs/icd/icd10cm.htm

ICD-10 vs. ICD-10-CM

E72 **Other disorders of amino-acid metabolism**

Excl.: abnormal findings without manifest disease (R7
disorders of:
- aromatic amino-acid metabolism (E70.-)
- branched-chain amino-acid metabolism (
- fatty-acid metabolism (E71.3)
- purine and pyrimidine metabolism (E79.-

gout (M10.-)

E72.0 **Disorders of amino-acid transport**

Cystine storage disease† (N29.8*)
Cystinosis
Cystinuria
Fanconi(-de Toni)(-Debré) syndrome
Hartnup disease
Lowe syndrome

Excl.: disorders of tryptophan metabolism (E70.8)

x6

E72 **Other disorders of amino-acid metabolism**

Excludes1: disorders of:
aromatic amino-acid metabolism (E70.-)
branched-chain amino-acid metabolism (E71.0-E71.2)
fatty-acid metabolism (E71.3)
purine and pyrimidine metabolism (E79.-)
gout (M1A.-, M10.-)

E72.0 **Disorders of amino-acid transport**

Excludes1: disorders of tryptophan metabolism (E70.5)

E72.00 **Disorders of amino-acid transport, unspecified**

E72.01 **Cystinuria**

E72.02 **Hartnup's disease**

E72.03 **Lowe's syndrome**

Use additional code for associated glaucoma (H42)

E72.04 **Cystinosis**
Fanconi (-de Toni) (-Debré) syndrome with cystinosis

Excludes1: Fanconi (-de Toni) (-Debré) syndrome wit

E72.09 **Other disorders of amino-acid transport**
Fanconi (-de Toni) (-Debré) syndrome, unspecified

ICD-10 vs. ICD-10-CM

W58	Bitten or struck by crocodile or alligator

W58 Contact with crocodile or alligator

The appropriate 7th character is to be added to each code from category W58
- A - initial encounter
- D - subsequent encounter
- S - sequela

W58.0 Contact with alligator

- **W58.01 Bitten by alligator**
- **W58.02 Struck by alligator**

W58.01A Bitten by alligator, initial encounter
W58.01D Bitten by alligator, subsequent encounter
W58.01S Bitten by alligator, sequela

- **W58.03 Crushed by alligator**
- **W58.09 Other contact with alligator**

W58.1 Contact with crocodile

- **W58.11 Bitten by crocodile**
- **W58.12 Struck by crocodile**
- **W58.13 Crushed by crocodile**
- **W58.19 Other contact with crocodile**

x24

Logical Observation Identifiers, Names and Codes (LOINC)

LOINC®
from Regenstrief

LOINC Characteristics (1)

- Current version: 2.59 (Feb. 2017)
  - 2 annual releases
- Type: Controlled terminology*
- Domain: Laboratory and clinical observations
- Developer: Regenstrief Institute
- Funding: NLM and other sources
- Publicly available: Yes
- Used for: information exchange
- URL: https://loinc.org/

LOINC Characteristics (2)

- Number of
  - Concepts: 73,958 active codes (2.52, June 2015)
  - Terms: 1 per concept ("long name")
- Major organizing principles:
  - No hierarchical structure among the main codes
  - 6 axes
    - Component (analyte [+ challenge] [+ adjustments])
    - Property
    - Timing
    - System
    - Scale
    - [Method]
- Distribution: proprietary database format

LOINC Example

- *Sodium [Moles/volume] in Serum or Plasma* [the molar concentration of sodium is measured in the plasma (or serum), with quantitative result]

Axis	Value
Component	Sodium
Property	SCnc – Substance Concentration (per volume)
Timing	Pt – Point in time (Random)
System	Ser/Plas – Serum or Plasma
Scale	Qn – Quantitative
Method	--

2951-2 Sodium [Moles/volume] in Serum or Plasma

NAME

Fully-Specified Name:

Component	Property	Time	System	Scale	Method
Sodium	SCnc	Pt	Ser/Plas	Qn	

PART DEFINITION/DESCRIPTION(S)

Sodium is an essential nutrient that regulates blood volume, blood pressure, osmotic equilibrium and electrolyte balance. Sodium chloride is the principal source of sodium in the diet, and is used for seasoning and as a preservative. Increased levels of sodium intake can cause hypertension and reportedly leads to 7.6 million premature deaths worldwide. Sodium is also important in neuron function and osmoregulation between cells and the extracellular fluid.

Source: Wikipedia, **URL:** Sodium (Wikipedia)

BASIC ATTRIBUTES

Class/Type:	CHEM/Lab
CDISC Lab Test:	Y
Common Lab Results Rank:	#5
Common SI Lab Results Rank:	#5
Common Orders Rank:	#107
Last Updated in Version:	2.34
Order vs. Obs.:	Both
Status:	Active

EXAMPLE UNITS

Unit	Source Type
mmol/L	EXAMPLE UCUM UNITS
mmol/L	REGENSTRIEF
mmol/L	eCHN

UNITS AND RANGE

Range	Units Type
mmol/L:[136,145]	

SNOMED Clinical Terms

ihtsdo Leading healthcare terminology, worldwide

SNOMED CT Characteristics (1)

- Current version: January 31, 2017
  - 2 annual releases
- Type: Reference terminology / ontology
- Domain: Clinical medicine
- Developer: IHTSDO
- Funding: IHTSDO member countries
- Publicly available: Yes*
- Used for: clinical documentation, information exchange, analytics
- URL: http://www.ihtsdo.org/

SNOMED CT Characteristics (2)

- Number of
  - Concepts: 320,912 active concepts (Sept. 2016)
  - Terms: 2.6 per concept (“descriptions”)
- Major organizing principles:
  - Polyhierarchy
  - Rich set of associative relationships
  - Logical definitions (incomplete: many primitives)
  - Built using description logics (EL++)
- Distribution: RF2 (proprietary)

http://browser.ihtsdotools.org/

SNOMED CT Top level

- SNOMED CT Concept
  - Body structure (body structure)
  - Clinical finding (finding)
  - Environment or geographical location (environment / location)
  - Event (event)
  - Observable entity (observable entity)
  - Organism (organism)
  - Pharmaceutical / biologic product (product)
  - Physical force (physical force)
  - Physical object (physical object)
  - Procedure (procedure)
  - Qualifier value (qualifier value)
  - Record artifact (record artifact)
  - Situation with explicit context (situation)
  - SNOMED CT Model Component (metadata)
  - Social context (social concept)
  - Special concept (special concept)
  - Specimen (specimen)
  - Staging and scales (staging scale)
  - Substance (substance)

SNOMED CT Example

Parents

- Operation on appendix (procedure)
- Partial excision of large intestine (procedure)

Appendectomy (procedure)

SCTID: 80146002

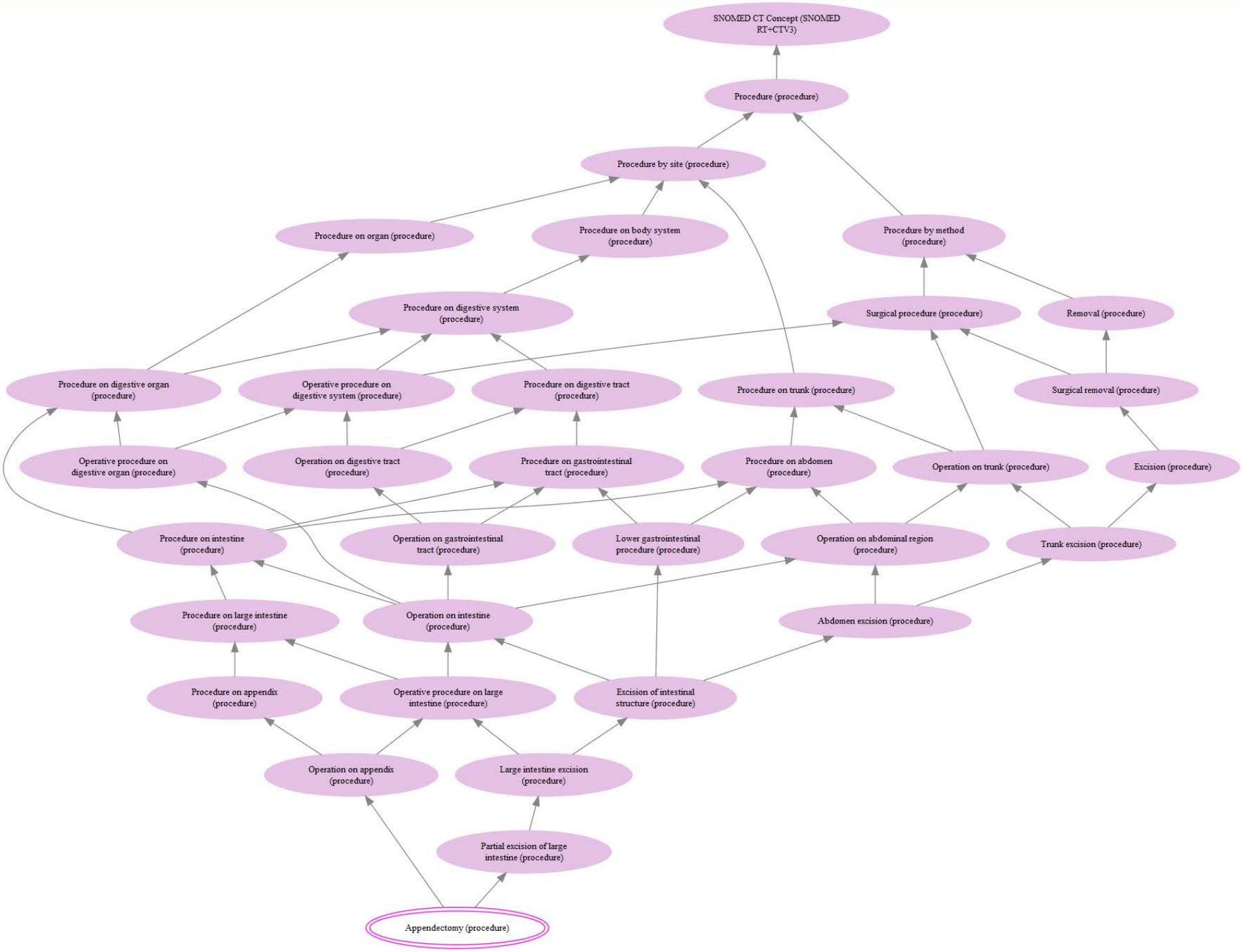
80146002 | Appendectomy (procedure) |

Appendectomy
Excision of appendix
Appendicectomy
Appendectomy (procedure)

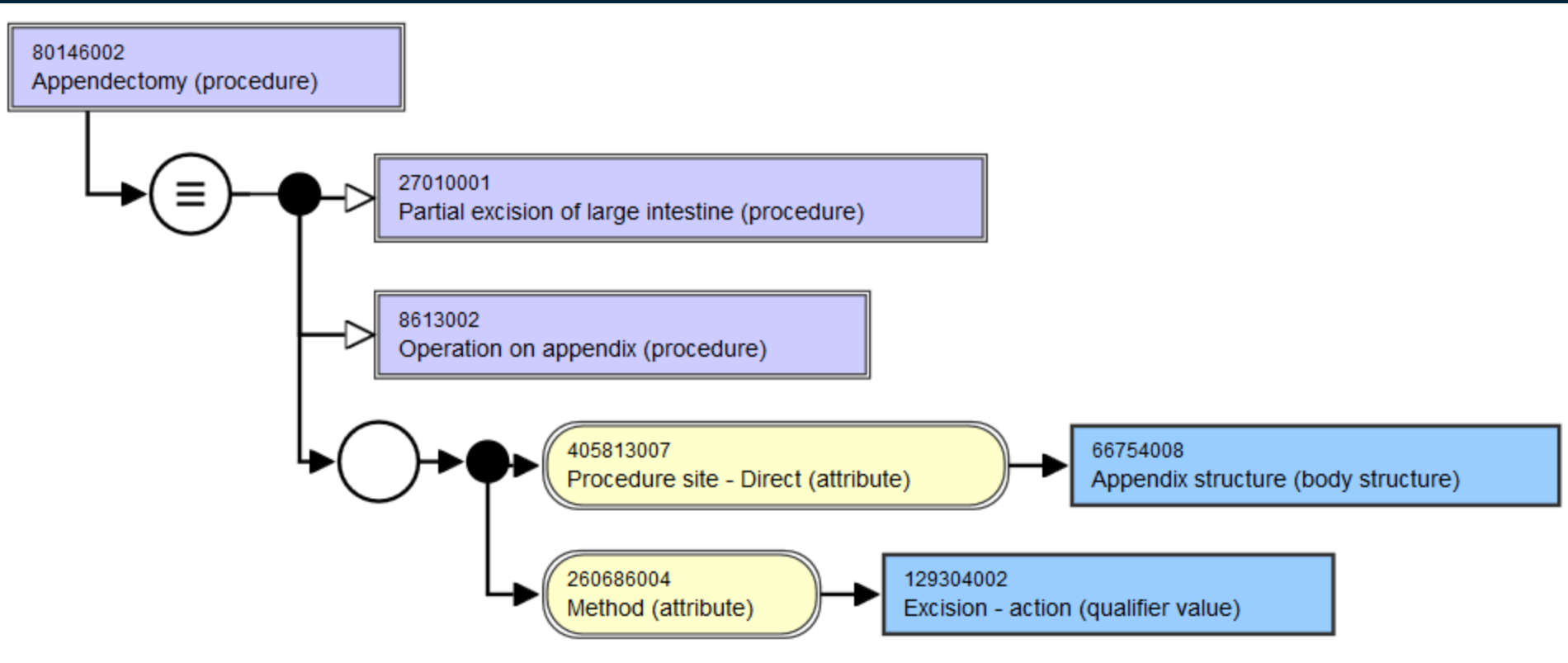
Procedure site - Direct → Appendix structure
Method → Excision - action

Children (8)

- Appendectomy with drainage (procedure)
- Emergency appendectomy (procedure)
- Excision of appendiceal stump (procedure)
- Excision of ruptured appendix by open approach (procedure)
- Incidental appendectomy (procedure)
- Interval appendectomy (procedure)
- Laparoscopic appendectomy (procedure)
- Non-emergency appendectomy (procedure)


SNOMED CT Concept (SNOMED RT+CTV3)
Procedure (procedure)
Procedure by site (procedure)
Procedure on organ (procedure)
Procedure on body system (procedure)
Procedure by method (procedure)
Procedure on digestive system (procedure)
Surgical procedure (procedure)
Removal (procedure)
Procedure on digestive organ (procedure)
Operative procedure on digestive system (procedure)
Procedure on digestive tract (procedure)
Procedure on trunk (procedure)
Surgical removal (procedure)
Operative procedure on digestive organ (procedure)
Operation on digestive tract (procedure)
Procedure on gastrointestinal tract (procedure)
Procedure on abdomen (procedure)
Operation on trunk (procedure)
Excision (procedure)
Procedure on intestine (procedure)
Operation on gastrointestinal tract (procedure)
Lower gastrointestinal procedure (procedure)
Operation on abdominal region (procedure)
Trunk excision (procedure)
Procedure on large intestine (procedure)
Operation on intestine (procedure)
Abdomen excision (procedure)
Procedure on appendix (procedure)
Operative procedure on large intestine (procedure)
Excision of intestinal structure (procedure)
Operation on appendix (procedure)
Large intestine excision (procedure)
Partial excision of large intestine (procedure)
Appendectomy (procedure)

SNOMED CT Example



RxNorm

RxNorm Characteristics (1)

- Current version: March 2017
  - Monthly releases (+weekly updates)
- Type: Controlled terminology
- Domain: Drug names
- Developer: NLM
- Funding: NLM
- Publicly available: Yes*
- Used for: e-prescribing, information exchange, analytics
- URL: http://www.nlm.nih.gov/research/umls/rxnorm/

RxNorm Characteristics (2)

- Number of
  - Concepts: 117,774 (March 2016)
  - Terms: 1.5 per concept
- Major organizing principles:
  - Generic vs. brand
  - Ingredient + Strength + Dose form
  - No hierarchical structure; rich graph of associative relations
  - Integrates all major US drug information sources
  - No clinical information
- Distribution: similar to UMLS RRF format

RxNorm Normalized form

Strength **Ingredient** **Dose form**

4mg/ml Fluoxetine Oral Solution

Strength **Ingredient**

Semantic clinical drug component

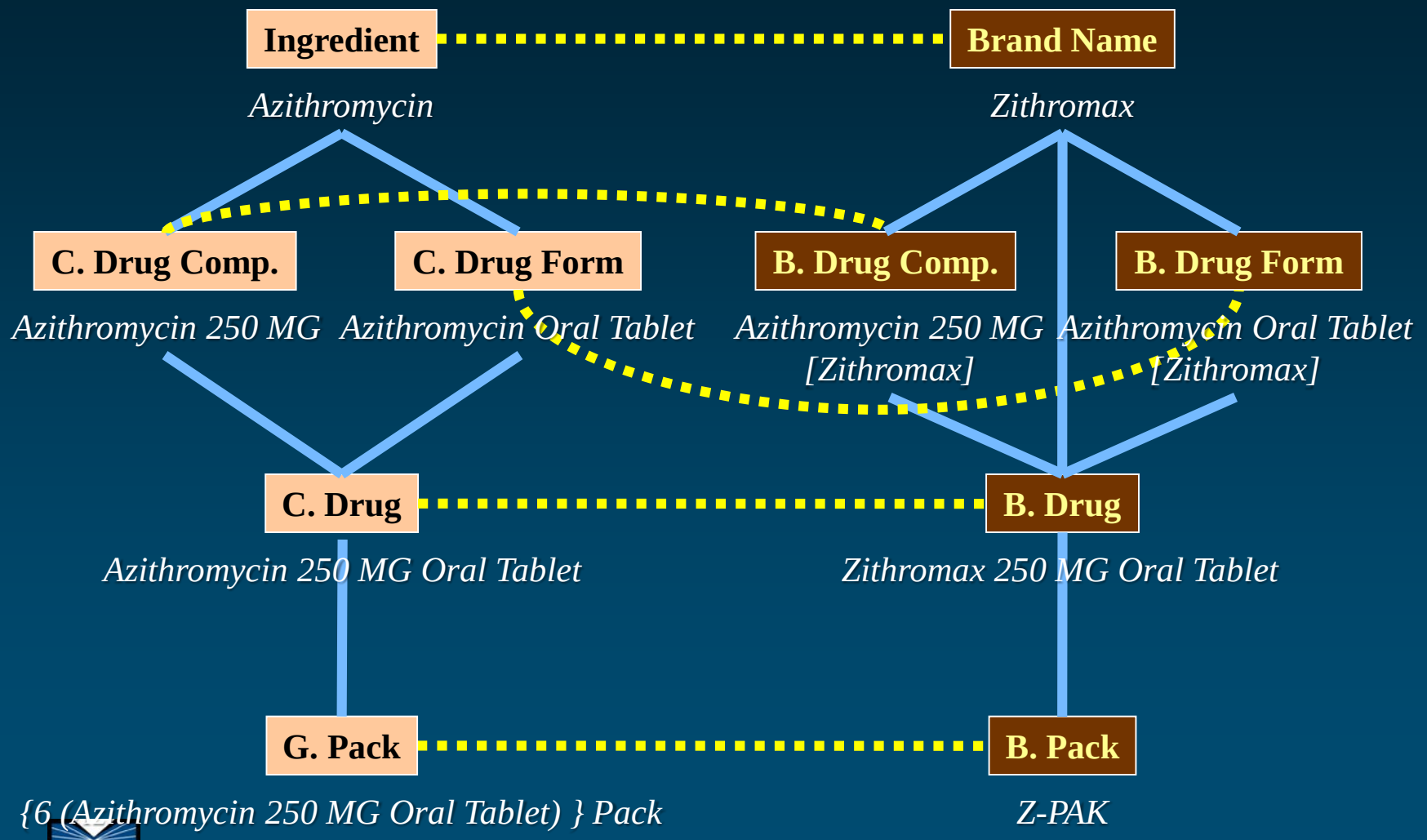
Ingredient **Dose form**

Semantic clinical drug form

Strength **Ingredient** **Dose form**

Semantic clinical drug

RxNorm Example



https://mor.nlm.nih.gov/RxNav/

https://mor.nlm.nih.gov/RxNav/search?searchBy=String&searchTerm=warfarin 110% Search

NIH U.S. National Library of Medicine About Disclaimer FAQ

RxNav Navigating RxNorm Drugs

String warfarin

Warfarin [RxCUI = 11289]

RxNorm Graph | RxNorm Properties | NDC | RxTerms | NDF-RT | Pill Images | Class View | Interaction View

Views
- Classic
- Simple
- Table

Filters
H V Rx S

Group
Form

Links

Legend
MIN
Pack
Multi

Download

IN/MIN (1)
H Rx S Warfarin

PIN (2)
S Warfarin Potassium
H Rx S Warfarin Sodium

BN (2)
H Rx S Coumadin
H Rx S Jantoven

SCDC (11)
S Warfarin Sodium 0.5 MG
H Rx S Warfarin Sodium 1 MG
H Rx S Warfarin Sodium 10 MG
H Rx S Warfarin Sodium 2 MG

SBDC (19)
H Rx S Warfarin Sodium 1 MG [Coumadin]
H Rx S Warfarin Sodium 1 MG [Jantoven]

SCD/GPCK (11)
S Warfarin Sodium 0.5 MG Oral Tablet
H Rx S Warfarin Sodium 1 MG Oral Tablet
H Rx S Warfarin Sodium 10 MG Oral Tablet
H Rx S Warfarin Sodium 2 MG Oral Tablet

SBD/BPCK (19)
H Rx S Coumadin 1 MG Oral Tablet
H Rx S Coumadin 10 MG Oral Tablet
H Rx S Coumadin 2 MG Oral Tablet
S Coumadin 2 MG/ML Injectable

SCDG (3)
S Warfarin Injectable Product
H Rx S Warfarin Oral Product
H Rx S Warfarin Pill

DFG (3)
HV Rx S Injectable Product
HV Rx S Oral Product
HV Rx S Pill

SBDG (5)
S Coumadin Injectable Product
H Rx S Coumadin Oral Product
H Rx S Coumadin Pill
H Rx S Jantoven Oral Product

Product Identification: NDCs

- National Drug Codes
  - Product identification system
  - Three components
    - Manufacturer
    - Product
    - Packaging
- Introduced in 1972 by FDA
- Only format permitted by NCPDP
- Mandated by HIPAA regulations for drug transactions

Source: Dan Malone

NDC Elements: 3 segments

XXXXX-XXXX-XX

Manufacturer Product Packaging

NDC Forms

Warfarin Sodium 1 MG Oral Tablet

XXXX-XXXX-XX (4-4-2) → 0XXXX XXXX XX

0555-0831-02 (Teva Pharmaceuticals USA, Inc.; 100 in 1 BOTTLE) → 00555083102

XXXXX-XXX-XX (5-3-2) → XXXXX 0XXX XX

21695-672-30 (Rebel Distributors Corp; 30 in 1 BOTTLE) → 21695067230

XXXXX-XXXX-X (5-4-1) → XXXXX XXXX 0X

50090-1213-0 (A-S Medication Solutions; 30 in 1 BOTTLE) → 50090121300

Clinical Terminologies
Part 3

Biomedical Ontologies “in Action”

A Functional Perspective

Overview

[Bodenreider, YBMI 2008]

- Functional perspective
  - What are they for (vs. what are they)?
- "High-impact" biomedical ontologies
- 3 major categories of use
  - Knowledge management (indexing and retrieval of data and information, access to information, mapping among ontologies)
  - Data integration, exchange and semantic interoperability
  - Decision support and analytics (data selection and aggregation, decision support, natural language processing applications, knowledge discovery)

Knowledge management

Knowledge management

Annotating data and resources

Terminology in ontology

- Ontology as a source of vocabulary
  - List of names for the entities in the ontology (ontology vs. terminology)
- Most ontologies have some sort of terminological component
- Not all surface forms represented
  - Often insufficient for NLP applications
  - Large variation in number of terms per concept across ontologies

Annotating data

- Gene Ontology
  - Functional annotation of gene products in several dozen model organisms
- Various communities use the same controlled vocabularies
- Enabling comparisons across model organisms
- Annotations
  - Assigned manually by curators
  - Inferred automatically (e.g., from sequence similarity)

GO Annotations across species

ALDH2 aldehyde dehydrogenase 2 family (mitochondrial) [*Homo sapiens* (human)]

Gene ID: 217, updated on 13-Mar-2016

⊟ Gene Ontology Provided by GOA

Function	Evidence Code	Pubs
aldehyde dehydrogenase (NAD) activity	EXP	
aldehyde dehydrogenase (NAD) activity	IDA	PubMed
aldehyde dehydrogenase [NAD(P)+] activity	TAS	PubMed
electron carrier activity	TAS	PubMed

Aldh2 aldehyde dehydrogenase 2, mitochondrial [*Mus musculus* (house mouse)]

Gene ID: 11669, updated on 26-Jan-2016

⊟ Gene Ontology Provided by MGI

Function	Evidence Code	Pubs
NADH binding	ISO	
aldehyde dehydrogenase (NAD) activity	IBA	
aldehyde dehydrogenase (NAD) activity	ISO	
identical protein binding	ISO	
oxidoreductase activity	IEA	
oxidoreductase activity, acting on the aldehyde or oxo group of donors, NAD or NADP as acceptor	IEA	
protein binding	IPI	PubMed

Indexing the biomedical literature

- MeSH
  - Used for indexing and retrieval of the biomedical literature (MEDLINE)
- Indexing
  - Performed manually by human indexers
    - With help of semi-automatic systems (suggestions) e.g., Indexing Initiative at NLM
  - Specific indexing rules

MeSH MEDLINE indexing

Anesth Analg. 2008 Jun;106(6):1813-9. doi: 10.1213/ane.0b013e318172fdba.

PubMed.gov
US National Library of Medicine
National Institutes of Health

Free cortisol in sepsis and septic shock.

Bendel S[1], Karlsson S, Pettilä V, Loisa P, Varpula M, Ruokonen E; Finnsepsis Study Group.

Collaborators (26)

Author information

Abstract

BACKGROUND: Severe sepsis activates the hypothalamopituitary axis, increasing cortisol production. In some studies, hydrocortisone substitution based on an adrenocorticotropic hormone-stimulation test or baseline cortisol measurement has improved outcome. Because only the free fraction of cortisol is active, measurement of free cortisol may be more important than total cortisol in critically ill patients. We measured total and free cortisol in patients with severe sepsis and related the concentrations to outcome.

METHODS: In a prospective study, severe sepsis was defined according the American College of Chest Physicians/Society of Critical Care Medicine criteria. Blood samples were drawn within 24 h of study entry. Serum cortisol was analyzed by electrochemiluminescence immunoassay. The Coolens method was used for calculating serum free cortisol concentrations.

RESULTS: Blood samples were collected from 125 patients, of whom 62 had severe sepsis and 63 septic shock. Hospital mortality was 21%. Calculated free serum cortisol correlated well with serum total cortisol ($r = 0.90$, $P < 0.001$). There was no difference in the total cortisol concentrations in patients with sepsis and septic shock (728 +/- 386 nmol/L vs 793 +/- 439 nmol/L, $P = 0.44$). Nonsurvivors had higher calculated serum free (209 +/- 151 nmol/L) and total (980 +/- 458 nmol/L) cortisol concentrations than survivors (119 +/- 111 nmol/L, $P = 0.002$, and 704 +/- 383 nmol/L, $P = 0.002$). Depending on the definition, the incidence of adrenal insufficiency varied from 8% to 54%.

CONCLUSIONS: Clinically, calculation of free cortisol does not provide essential information for identification of patients who would benefit from corticoid treatment in severe sepsis and septic shock.

PMID: 18499615 [PubMed - indexed for MEDLINE]

MeSH MEDLINE indexing

MeSH Terms

Adrenal Cortex Function Tests
Adrenal Insufficiency/blood*
Adrenal Insufficiency/drug therapy
Adrenal Insufficiency/mortality
Adult
Biomarkers/blood
Female
Finland/epidemiology
Hospital Mortality
Humans
Hydrocortisone/blood*
Hydrocortisone/therapeutic use
Kaplan-Meier Estimate
Male
Predictive Value of Tests
Prospective Studies
Sepsis/blood*
Sepsis/drug therapy
Sepsis/mortality
Severity of Illness Index
Shock, Septic/blood*
Shock, Septic/drug therapy
Shock, Septic/mortality
Treatment Outcome

SNOMED CT/ICD Coding clinical data

- SNOMED CT
  - Used for clinical documentation
  - E.g., problem lists
- ICD-10-CM
  - Used for coding clinical data for billing purposes
  - Other uses of ICD
    - Morbidity and mortality reporting worldwide
  - Specific coding rules

Knowledge management

Accessing biomedical information

Resources for biomedical search engines

- Synonyms
- Hierarchical relations
- High-level categorization
- [Co-occurrence information]
- Translation

MeSH "synonyms" MEDLINE retrieval

- MeSH entry terms
  - Used as equivalent terms for retrieval purposes (query expansion)
  - Not always synonymous
- Increase recall without hurting precision

MeSH Heading	Addison Disease
Entry Term	Addison's Disease
Entry Term	Primary Adrenal Insufficiency
Entry Term	Primary Adrenocortical Insufficiency
Entry Term	Primary Hypoadrenalism

MeSH "synonyms" MEDLINE retrieval

NCBI Resources How To Sign in to NCBI

PubMed.gov
US National Library of Medicine
National Institutes of Health

PubMed | primary hypoadrenalism | Search

Create RSS Create alert Advanced Help

Search details

```
"addison disease"[MeSH Terms] OR
("addison"[All Fields] AND
"disease"[All Fields]) OR "addison
disease"[All Fields] OR
("primary"[All Fields] AND
"hypoadrenalism"[All Fields]) OR
"primary hypoadrenalism"[All Fields]
```

Search

See more...

MeSH hierarchies MEDLINE retrieval

- MeSH "explosion"
  - Search for a given MeSH term and all its descendants
  - A search on Adrenal insufficiency also retrieves articles indexed with its descendant, Addison disease

PubMed.gov
US National Library of Medicine
National Institutes of Health

PubMed | adrenal insufficiency [mh] | Search

Create RSS Create alert Advanced Help

Adrenal insufficiency in prolonged critical illness.
145. Wu JY, Hsu SC, Ku SC, Ho CC, Yu CJ, Yang PC.
Crit Care. 2008;12(3):R65. doi: 10.1186/cc6895. Epub 2008 May 8.
PMID: 18466605 **Free PMC Article**
Similar articles

MeSH Terms
Adrenal Insufficiency/blood
Adrenal Insufficiency/drug therapy
Adrenal Insufficiency/mortality*

Addison's disease: a rare cause of hypertransaminasaemia.
146. Ersan O, Demirezer B.
Dig Dis Sci. 2008 Dec;53(12):3269-71. doi: 10.1007/s10620-008-0297-8. Epub 2008 May 9.
PMID: 18465237
Similar articles

MeSH Terms
Addison Disease/blood*
Addison Disease/complications
Addison Disease/diagnosis*

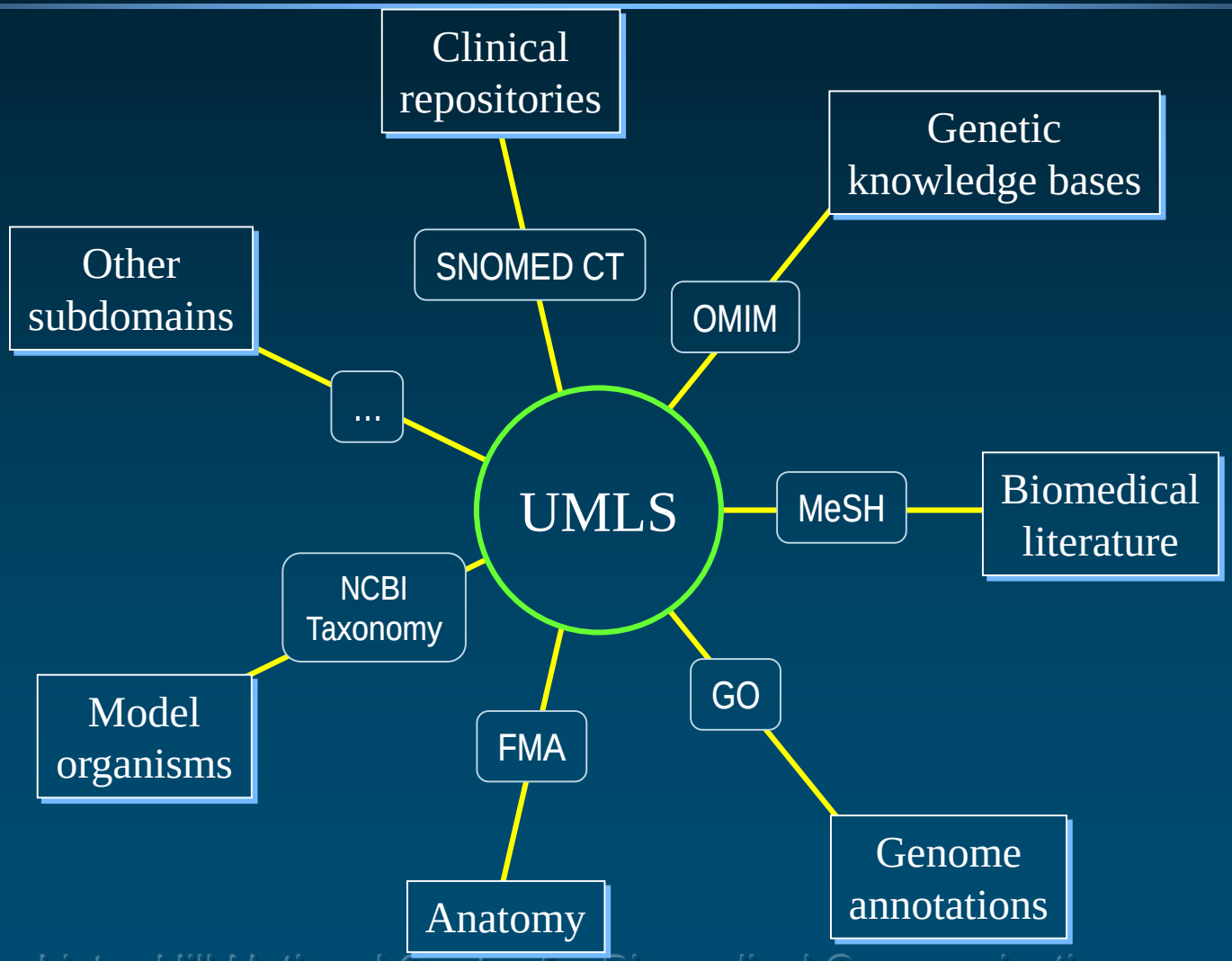
Knowledge management

Mapping across biomedical ontologies

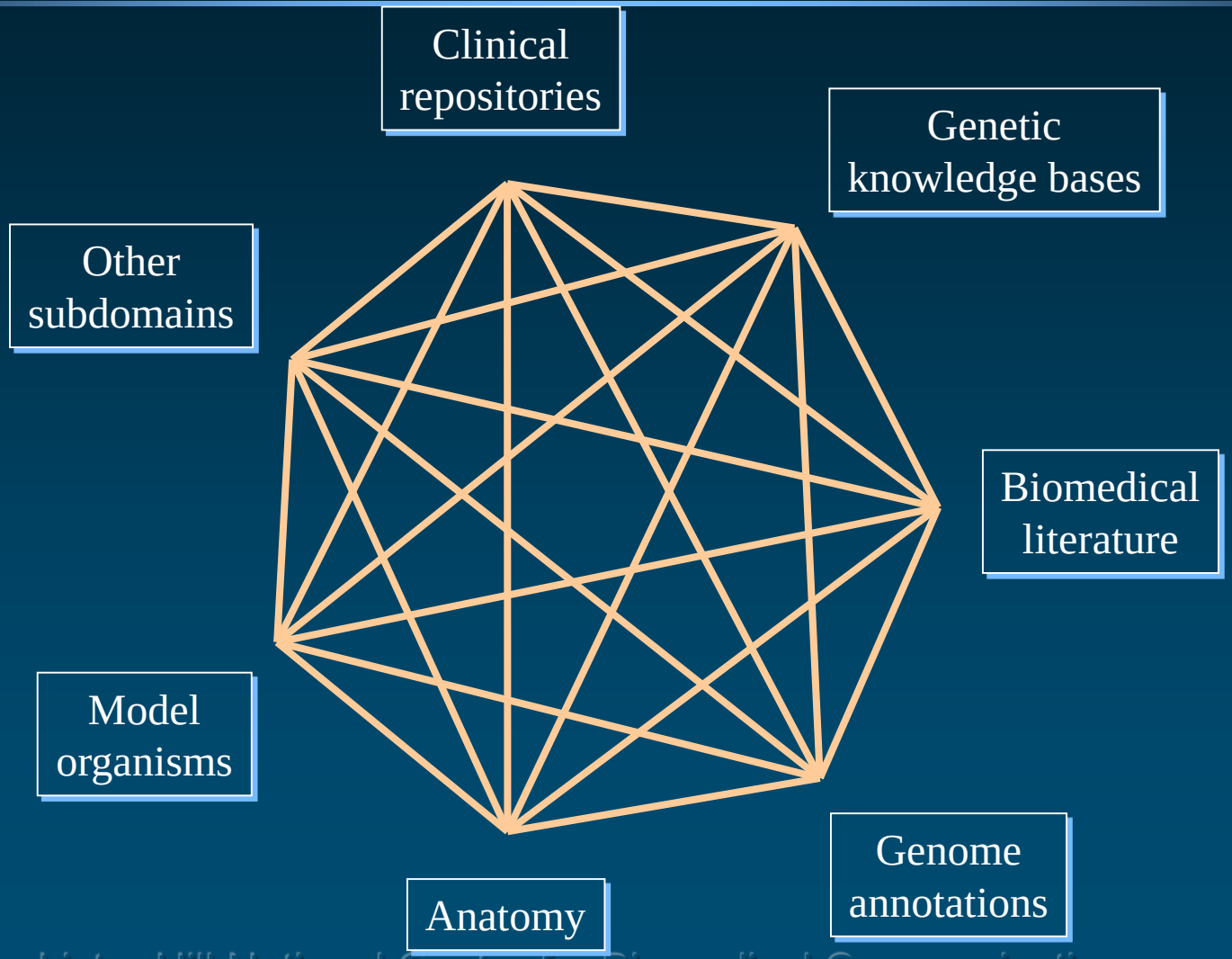
Terminology integration systems

- Terminology integration systems (UMLS, RxNorm) help bridge across vocabularies
- Uses
  - Information integration
  - Ontology alignment
  - Medication reconciliation

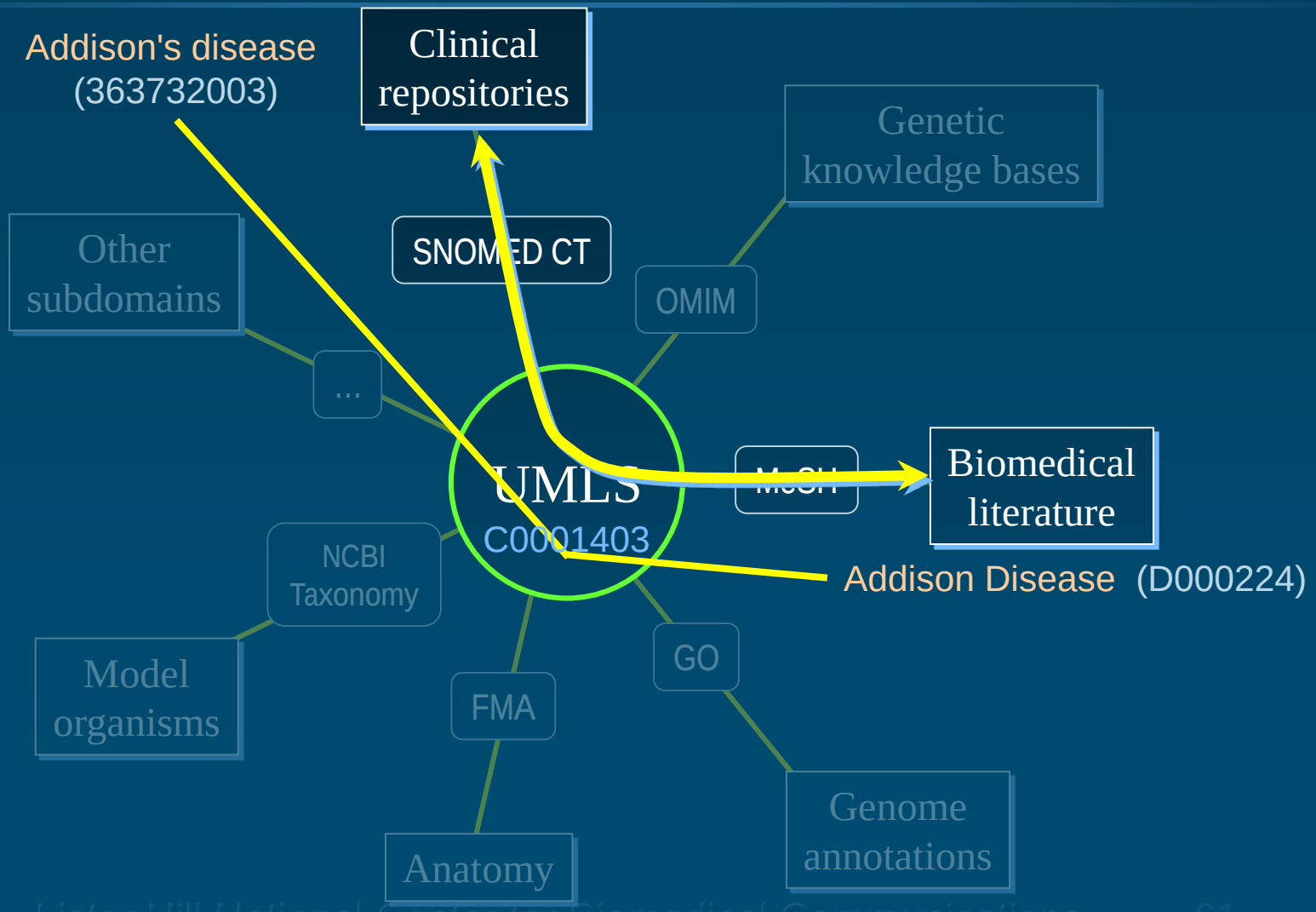
Integrating subdomains



Integrating subdomains



Trans-namespace integration



UMLS Source Vocabularies (2016AB)

- 154 families of source vocabularies
  - Not counting translations
- 25 languages
- Broad coverage of biomedicine
  - 10.3M names (normalized)
  - 3.4M concepts
  - ~13M relations among concepts
- Common presentation

Metathesaurus Basic organization

- Concepts
  - Synonymous terms are clustered into a concept
  - Properties are attached to concepts, e.g.,
    - Unique identifier
    - Definition
- Relations
  - Concepts are related to other concepts
  - Properties are attached to relations, e.g.,
    - Type of relationship
    - Source

Decision support and analytics

Data selection

- The structure of biomedical ontologies helps define groups of values from a high-level value
  - Vs. enumerating all possible values
- Useful for data selection in clinical studies
- ICD is used pervasively for this purpose
  - E.g., Study on supraventricular tachycardia (SVT), based on 2 high-level ICD codes
- Similarity with the definition of value sets for use in the information model

Data aggregation

- Ontologies help partition/aggregate data in data analysis
  - Clinical studies: Study a variable in groups of patients corresponding to the top level categories in ICD
  - Biology studies: Functional characterization of gene expression signatures with high-level concepts from the Gene Ontology
    - Recent trend: co-clustering

Decision support

- Clinical decision support
  - Ontologies help normalize the vocabulary and increase the recall of rules
  - Ontologies provide some domain knowledge and make it possible to create high-level rules (e.g., for a class of drugs rather than for each drug in the class)
- Other forms of decision support
  - Based on automatic reasoning services for OWL ontologies (e.g., grading gliomas with NCIt)

Natural language processing applications

- Ontologies provide background domain knowledge for NLP applications
  - Question answering
  - Document summarization
  - Literature-based discovery
- The UMLS is often used, but other specific resources have been developed

Knowledge discovery

- By standardizing the vocabulary in a given domain, ontologies are enabling resources for knowledge discovery through data mining
- Less frequently, the structure of the ontology is leveraged by data mining algorithms
- Example of available datasets
  - ICD-coded clinical data (in conjunction with non-clinical information, e.g., environmental data)
  - Annotation of gene products to the GO (function prediction)

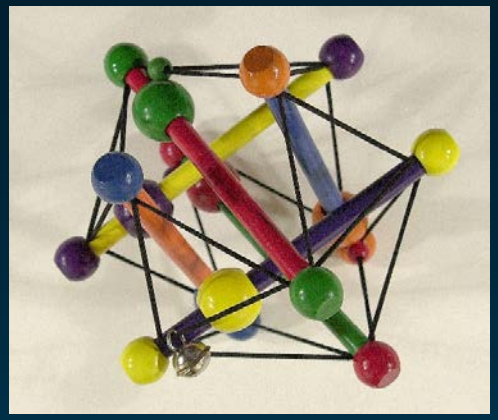
Summary

Summary

- History of biomedical ontologies
- Structure of the main clinical ontologies used
  - ICD, SNOMED CT, LOINC, RxNorm
- Purpose of biomedical ontologies
  - Knowledge management, [health information exchange and semantic interoperability], and clinical decision support and analytics

Topics not discussed

- Semantic Web, URIs, Linked Data
- Ontology creation, Protege
- Accessing terminology resources (APIs)
- Ontology repositories
  - [UMLS], NCBO BioPortal, EBI Ontology Lookup Service
- NLP, named entity recognition, MetaMap
- Mapping local terms to standard terminologies
- VSAC, value sets, common data elements
- OBO ontologies, OBO Foundry
- Coordinated development of ontologies, harmonization
- Boundary between terminology and information model
- […]



Medical Ontology Research

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