



College of  
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## Biomedical ontology *in action*



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U.S. National Library of Medicine



# Outline

- ◆ Introduction to biomedical terminologies through an example
- ◆ “High-Impact” Biomedical Ontologies
  - Structural perspective
- ◆ Biomedical Ontologies “in Action”
  - Functional perspective
- ◆ Terminology research at NLM

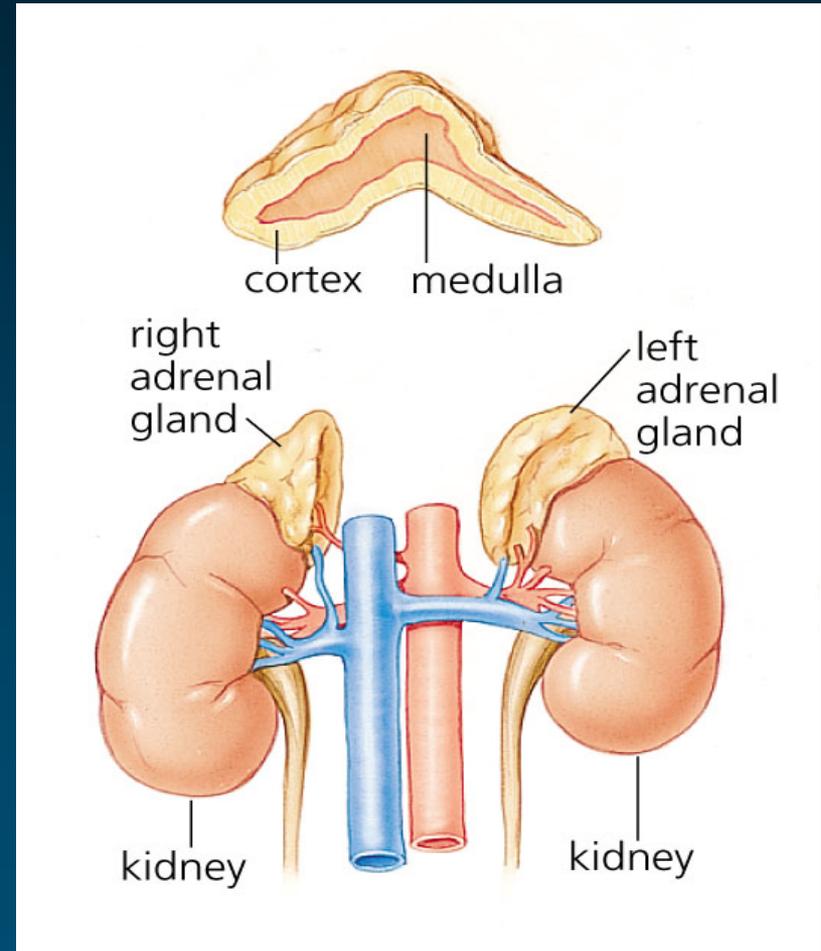


*Biomedical ontology in action*  
*Part 1*

Introduction to biomedical  
terminologies through an example

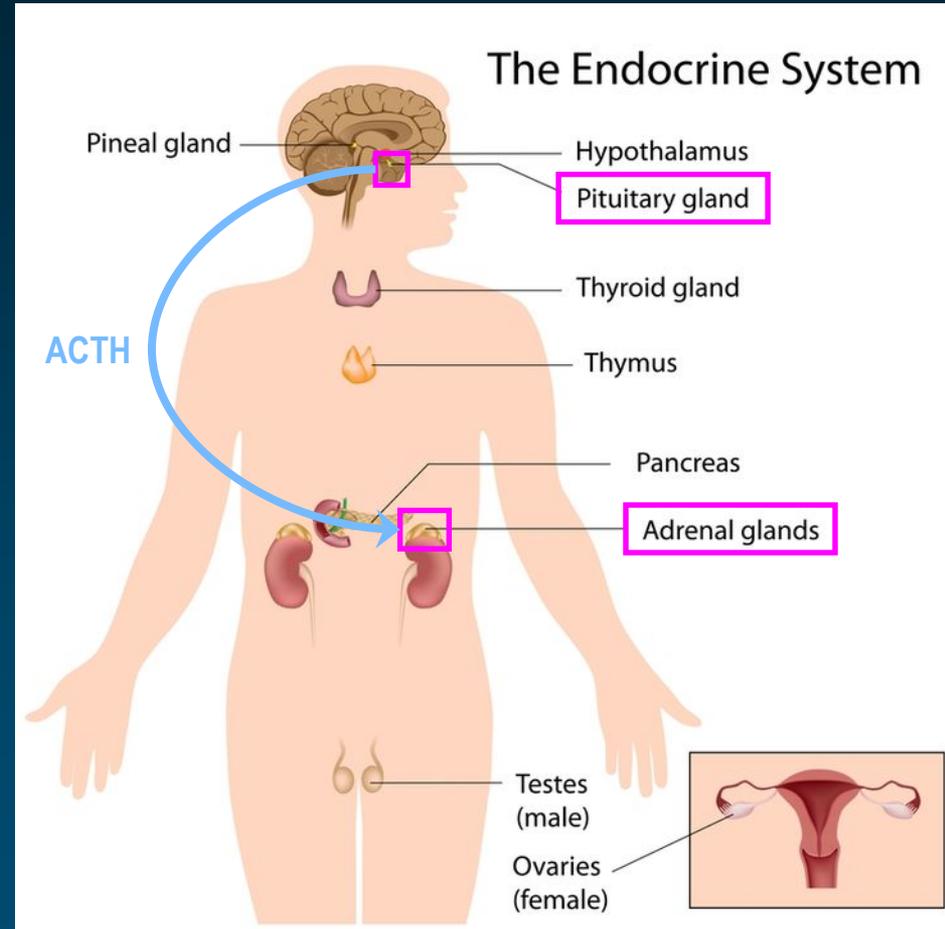
# Addison's disease

- ◆ Addison's disease is a rare endocrine disorder
- ◆ Addison's disease occurs when the adrenal glands do not produce enough of the hormone cortisol
- ◆ For this reason, the disease is sometimes called chronic adrenal insufficiency, or hypocortisolism



# Adrenal insufficiency Clinical variants

- ◆ Primary / Secondary
  - Primary: lesion of the adrenal glands themselves
  - Secondary: inadequate secretion of ACTH by the pituitary gland
- ◆ Acute / Chronic
- ◆ Isolated / Polyendocrine deficiency syndrome



# Addison's disease: Symptoms

- ◆ Fatigue
- ◆ Weakness
- ◆ Low blood pressure
- ◆ Pigmentation of the skin (exposed and non-exposed parts of the body)
- ◆ ...

# AD in medical vocabularies

## ◆ Synonyms: different terms

- Addisonian syndrome
  - Bronzed disease
  - Addison melanoderma
  - Asthenia pigmentosa
  - Primary adrenal deficiency
  - Primary adrenal insufficiency
  - Primary adrenocortical insufficiency
  - Chronic adrenocortical insufficiency
- )} eponym  
)} symptoms  
)} clinical variants

## ◆ Contexts: different hierarchies



# Internal Classification of Diseases

- ▼ **IV Endocrine, nutritional and metabolic diseases**
  - ▶ E00-E07 Disorders of thyroid gland
  - ▶ E10-E14 Diabetes mellitus
  - ▶ E15-E16 Other disorders of glucose regulation and pancreatic internal secretion
  - ▼ **E20-E35 Disorders of other endocrine glands**
    - ▶ E20 Hypoparathyroidism
    - ▶ E21 Hyperparathyroidism and other disorders of parathyroid gland
    - ▶ E22 Hyperfunction of pituitary gland
    - ▶ E23 Hypofunction and other disorders of pituitary gland
    - ▶ E24 Cushing syndrome
    - ▶ E25 Adrenogenital disorders
    - ▶ E26 Hyperaldosteronism
    - ▼ **E27 Other disorders of adrenal gland**
      - E27.0 Other adrenocortical overactivity
      - E27.1 Primary adrenocortical insufficiency
      - E27.2 Addisonian crisis
      - E27.3 Drug-induced adrenocortical insufficiency
      - E27.4 Other and unspecified adrenocortical insufficiency
      - E27.5 Adrenomedullary hyperfunction
      - E27.8 Other specified disorders of adrenal gland
      - E27.9 Disorder of adrenal gland, unspecified
    - ▶ E28 Ovarian dysfunction
    - ▶ E29 Testicular dysfunction
    - ▶ E30 Disorders of puberty, not elsewhere classified
    - ▶ E31 Polyglandular dysfunction
    - ▶ E32 Diseases of thymus
    - ▶ E34 Other endocrine disorders
    - ▶ E35 Disorders of endocrine glands in diseases classified elsewhere

## **E27 Other disorders of adrenal gland**

### **E27.0 Other adrenocortical overactivity**

Overproduction of ACTH, not associated with Cushing disease  
Premature adrenarche

**Excl.:** Cushing syndrome ([E24.-](#))

### **E27.1 Primary adrenocortical insufficiency**

Addison disease  
Autoimmune adrenalitis

**Excl.:** amyloidosis ([E85.-](#))  
tuberculous Addison disease ([A18.7](#))  
Waterhouse-Friderichsen syndrome ([A39.1](#))

### **E27.2 Addisonian crisis**

Adrenal crisis  
Adrenocortical crisis

### **E27.3 Drug-induced adrenocortical insufficiency**

Use additional external cause code (Chapter XX), if desired, to identify drug.

### **E27.4 Other and unspecified adrenocortical insufficiency**

Adrenal:

- haemorrhage
- infarction

Adrenocortical insufficiency NOS  
Hypoadosteronism

**Excl.:** adrenoleukodystrophy [Addison-Schilder] ([E71.3](#))  
Waterhouse-Friderichsen syndrome ([A39.1](#))

### **E27.5 Adrenomedullary hyperfunction**

Adrenomedullary hyperplasia  
Catecholamine hypersecretion

### **E27.8 Other specified disorders of adrenal gland**

Abnormality of cortisol-binding globulin

### **E27.9 Disorder of adrenal gland, unspecified**



# Medical Subject Headings

## MeSH Tree Structures

### [Endocrine System Diseases \[C19\]](#)

#### [Adrenal Gland Diseases \[C19.053\]](#)

##### [Adrenal Insufficiency \[C19.053.500\]](#)

▶ [Addison Disease \[C19.053.500.263\]](#)

[Adrenoleukodystrophy \[C19.053.500.270\]](#)

[Hypoadosteronism \[C19.053.500.480\]](#)

[Waterhouse-Friderichsen Syndrome \[C19.053.500.740\]](#)

### [Immune System Diseases \[C20\]](#)

#### [Autoimmune Diseases \[C20.111\]](#)

▶ [Addison Disease \[C20.111.163\]](#)

[Anemia, Hemolytic, Autoimmune \[C20.111.175\]](#)

[Anti-Glomerular Basement Membrane Disease \[C20.111.190\]](#)

[Anti-Neutrophil Cytoplasmic Antibody-Associated Vasculitis \[C20.111.193\] +](#)

[Antiphospholipid Syndrome \[C20.111.197\]](#)

[Arthritis, Juvenile \[C20.111.198\]](#)

[Arthritis, Rheumatoid \[C20.111.199\] +](#)

[Autoimmune Diseases of the Nervous System \[C20.111.258\] +](#)

[...]



# SNOMED CT

Concept Details

Concept Details

Summary

Details

Diagram

Expression

Re

**Parents**

- ▶ ● Abdominal organ finding (finding)
- ▶ ● Disorder of abdomen (disorder)
- ▶ ● Disorder of endocrine system (disorder)
- ▲ ● Disorder of adrenal gland (disorder)
- ▲ ● Hypoadrenalism (disorder)
- ▲ ● Adrenal hypofunction (disorder)
- ▶ ● Disorder of adrenal gland (disorder)
- ▲ ● Disorder of adrenal cortex (disorder)
- ▲ ● Adrenal cortical hypofunction (disorder)

```

graph BT
    Addison[Addison's Disease] --> AdrenalCort[Adrenal cortical hypofunction]
    AdrenalCort --> AdrenalHypo[Adrenal hypofunction]
    AdrenalCort --> DisorderCort[Disorder of adrenal cortex]
    AdrenalHypo --> Hypoadrenalism[Hypoadrenalism]
    DisorderCort --> DisorderGland[Disorder of adrenal gland]
    Hypoadrenalism --> DisorderGland
    DisorderGland --> DisorderEndo[Disorder of endocrine system]
            
```

● Addison's disease (disorder) ☆ ↗

SCTID: 363732003

363732003 | Addison's disease (disorder) |

- Addison disease
- Addison's disease
- Addison's disease (disorder)

Finding site → Adrenal cortex structure

**Children (4)**

- ● Addison's disease due to autoimmunity (disorder)
- ● Addison's disease with adrenoleucodystrophy (disorder)
- ● Polyglandular autoimmune syndrome, type 1 (disorder)
- ● Tuberculous Addison's disease (disorder)

10

*Biomedical ontology in action*  
*Part 2*

**“High-Impact” Biomedical  
Ontologies**

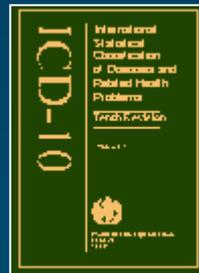
*A Structural Perspective*

# 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 (2017)
  - 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



# 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
<a href="#">[See before E10 for subdivisions]</a>	
<b>Incl.:</b> diabetes (mellitus):	
<ul style="list-style-type: none"><li>• brittle</li><li>• juvenile-onset</li><li>• ketosis-prone</li></ul>	
<b>Excl.:</b> diabetes mellitus (in):	
<ul style="list-style-type: none"><li>• malnutrition-related (<a href="#">E12.-</a>)</li><li>• neonatal (<a href="#">P70.2</a>)</li></ul>	
<ul style="list-style-type: none"><li>• pregnancy, childbirth and the puerperium (<a href="#">O24.-</a>)</li></ul>	
glycosuria:	
<ul style="list-style-type: none"><li>• NOS (<a href="#">R81</a>)</li><li>• renal (<a href="#">E74.8</a>)</li></ul>	
impaired glucose tolerance ( <a href="#">R73.0</a> )	
postsurgical hypoinsulinaemia ( <a href="#">E89.1</a> )	

# ICD Example

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

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

# 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 (CDC/NCHS)
- ◆ 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 (E71.0-E71.2)  
• 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)



**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 with

**E72.09 Other disorders of amino-acid transport**

Fanconi (-de Toni) (-Debré) syndrome, unspecified

x6

# 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

W58.03 Crushed by alligator

W58.09 Other contact with alligator

W58.01A Bitten by alligator, initial encounter

W58.01D Bitten by alligator, subsequent encounter

W58.01S Bitten by alligator, sequela

### 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 Characteristics (1)

- ◆ Current version: 2.61 (June 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:	<b>Component</b>	<b>Property</b>	<b>Time</b>	<b>System</b>	<b>Scale</b>	<b>Method</b>
	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.

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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

<b>Unit</b>	<b>Source Type</b>
mmol/L	EXAMPLE UCUM UNITS
mmol/L	REGENSTRIEF
mmol/L	eCHN

### UNITS AND RANGE

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

# SNOMED Clinical Terms



# SNOMED CT Characteristics (1)

- ◆ Current version: July 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)



# 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)

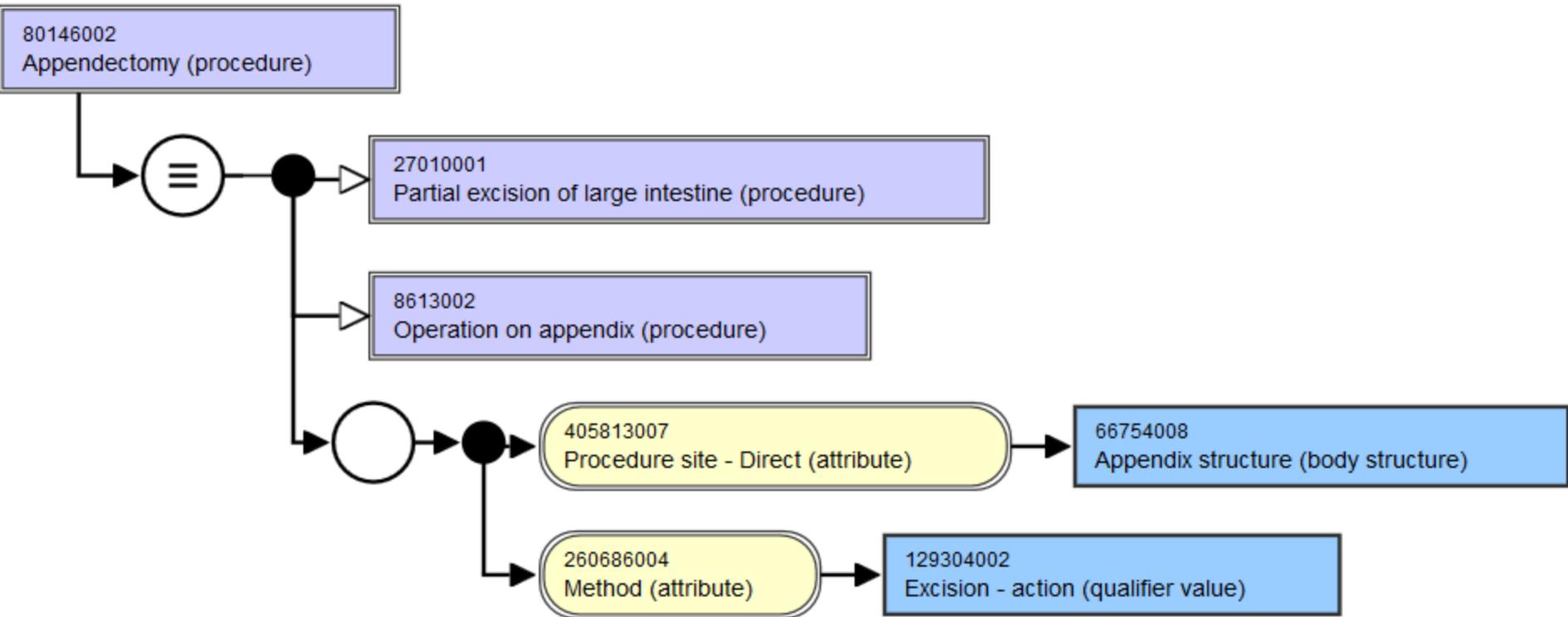
Procedure site - Direct → Appendix structure  
Method → Excision - action

## 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 Example



RxNorm

# RxNorm Characteristics (1)

- ◆ Current version: August 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**

4mg/ml

**Ingredient**

Fluoxetine

**Dose form**

Oral Solution

**Strength**

Semantic clinical drug component

**Ingredient**

**Ingredient**

Semantic clinical drug form

**Dose form**

**Strength**

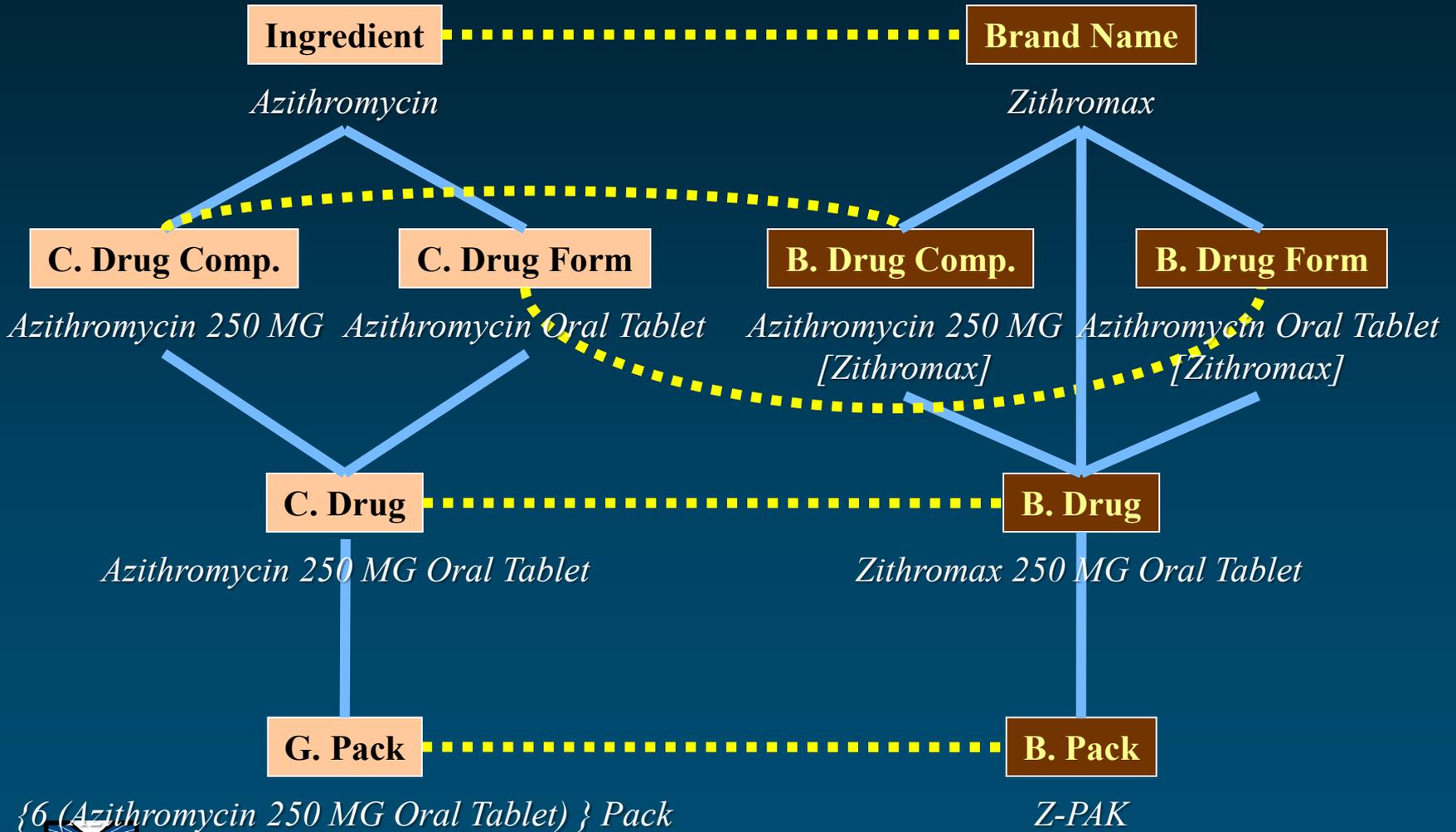
Semantic clinical drug

**Ingredient**

**Dose form**



# RxNorm Example



{6 (Azithromycin 250 MG Oral Tablet) } Pack

Z-PAK





String [dropdown] warfarin [input] [search icon] [refresh icon]



### 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
- MIN
  - Pack
  - Multi
- Download

IN/MIN	Ingredient (1)
H Rx S	Warfarin

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

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

SCDC	Clinical Drug Component (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	Branded Drug Component (18)
H Rx S	Warfarin Sodium 1 MG [Coumadin]
H Rx S	Warfarin Sodium 1 MG [Jantoven]
H Rx S	Warfarin Sodium 10 MG [Coumadin]
H Rx S	Warfarin Sodium 10 MG [Jantoven]

SCD/GPCK	Clinical Drug or Pack (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	Branded Drug or Pack (18)
H Rx S	Coumadin 1 MG Oral Tablet
H Rx S	Coumadin 10 MG Oral Tablet
H Rx S	Coumadin 2 MG Oral Tablet
H Rx S	Coumadin 2.5 MG Oral Tablet

SCDG	Clinical Dose Form Group (3)
S	Warfarin Injectable Product
H Rx S	Warfarin Oral Product
H Rx S	Warfarin Pill

DFG	Dose Form Group (3)
HVRx S	Injectable Product
HVRx S	Oral Product
HVRx S	Pill

SBDG	Branded Dose Form Group (4)
H Rx S	Coumadin Oral Product
H Rx S	Coumadin Pill
H Rx S	Jantoven Oral Product
H Rx S	Jantoven Pill



*Biomedical ontology in action*  
*Part 3*

**Biomedical Ontologies “in Action”**

*A Functional Perspective*

# Overview

- ◆ Functional perspective [Bodenreider, YBMI 2008]
  - What are they for (vs. what are they)?
- ◆ “High-impact” biomedical ontologies
- ◆ 3 major categories of use
  - **Knowledge management**
    - Annotating data and resources
    - Mapping across biomedical ontologies
  - **Decision support and analytics**
    - Value sets and clinical quality measures
  - **Data integration, exchange and semantic interoperability**
    - Common data models
    - Fast Healthcare Interoperability Resources (FHIR)



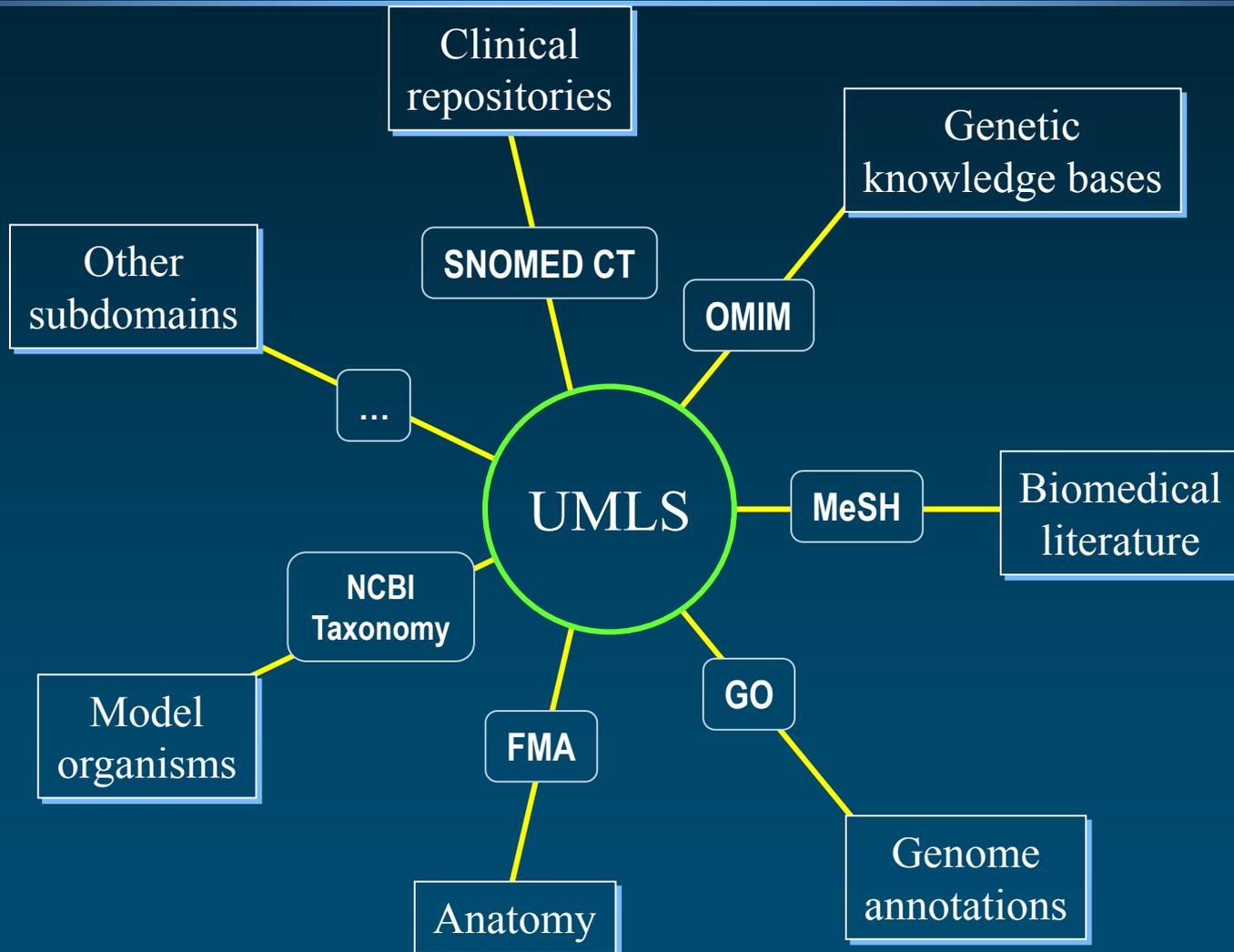
# Knowledge management

*Mapping across biomedical ontologies*

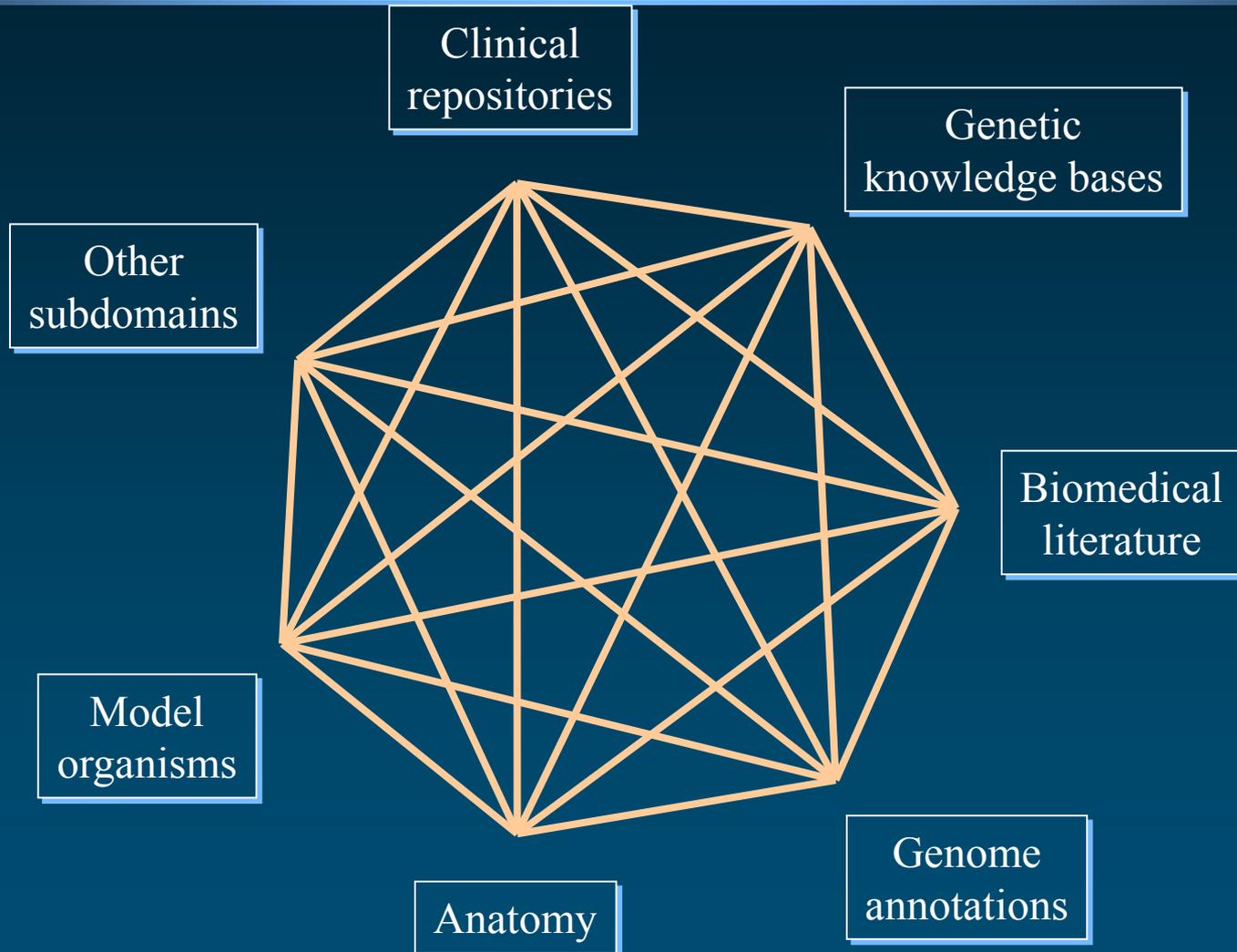
# Terminology integration systems

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

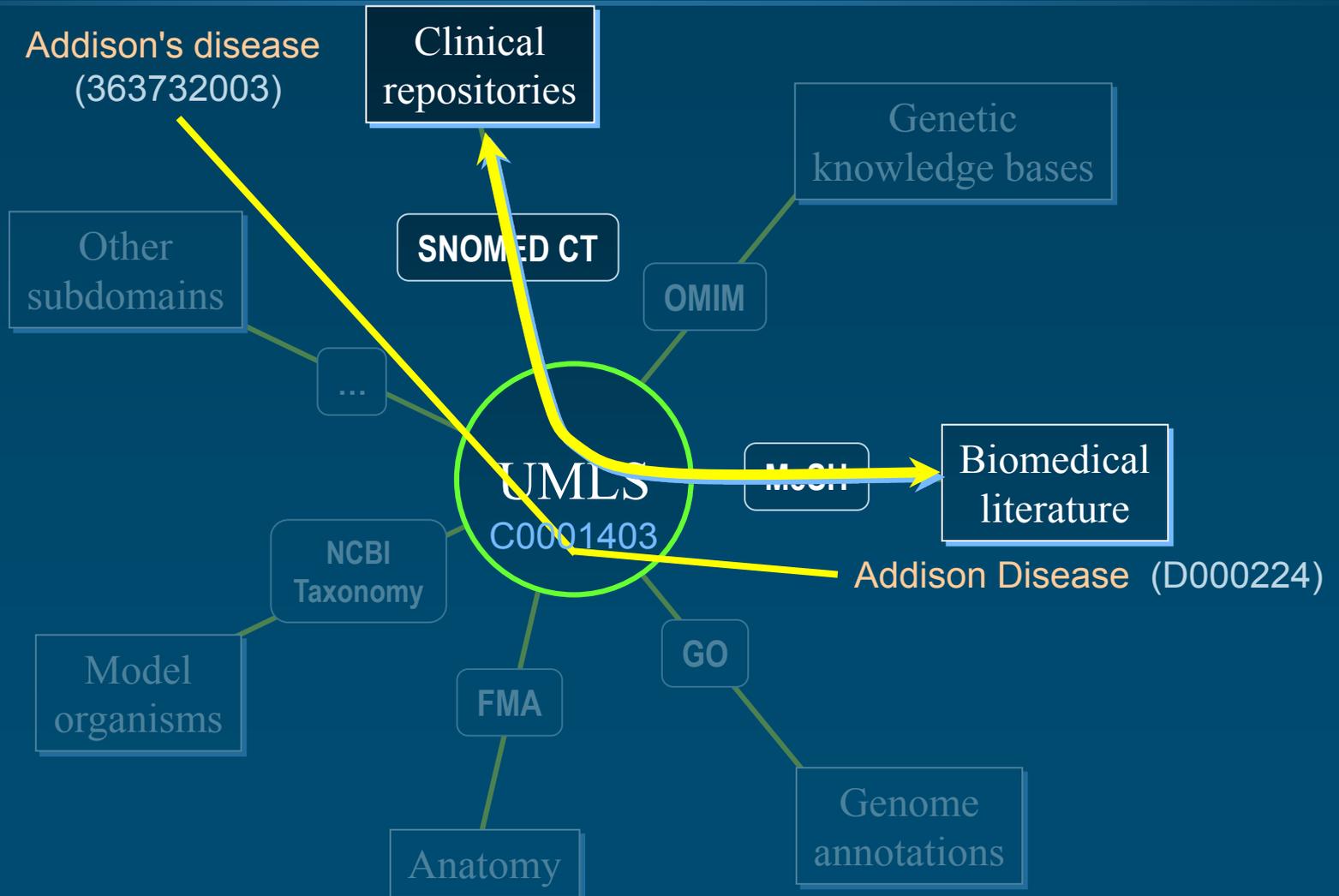
# Integrating subdomains



# Integrating subdomains



# Trans-namespace integration



# UMLS Source Vocabularies

(2015AB)

- ◆ 153 families of source vocabularies
  - Not counting translations
- ◆ 25 languages
- ◆ Broad coverage of biomedicine
  - 9.8M names (normalized)
  - 3.2M 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



# Data integration, exchange and semantic interoperability

*Common data models*

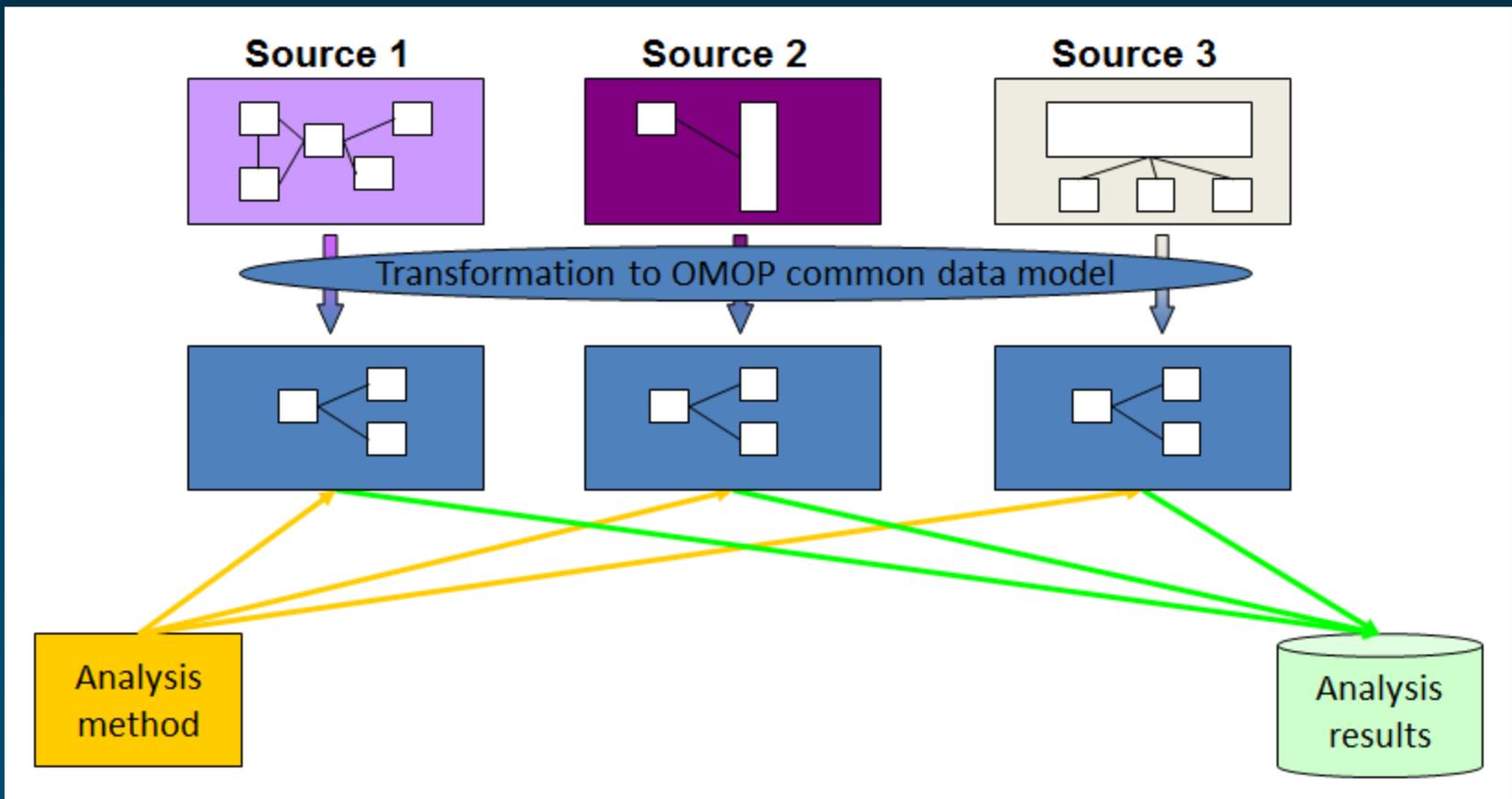
# Clinical data models

- ◆ Used in clinical data warehouses
  - Oriented towards analytics
  - Different from the transactional data models of EHR systems
  - Used to normalize data across EHR systems
- ◆ Multiple “common” data models
  - OMOP
  - i2b2
  - PCORnet
  - Sentinel
  - CDISC

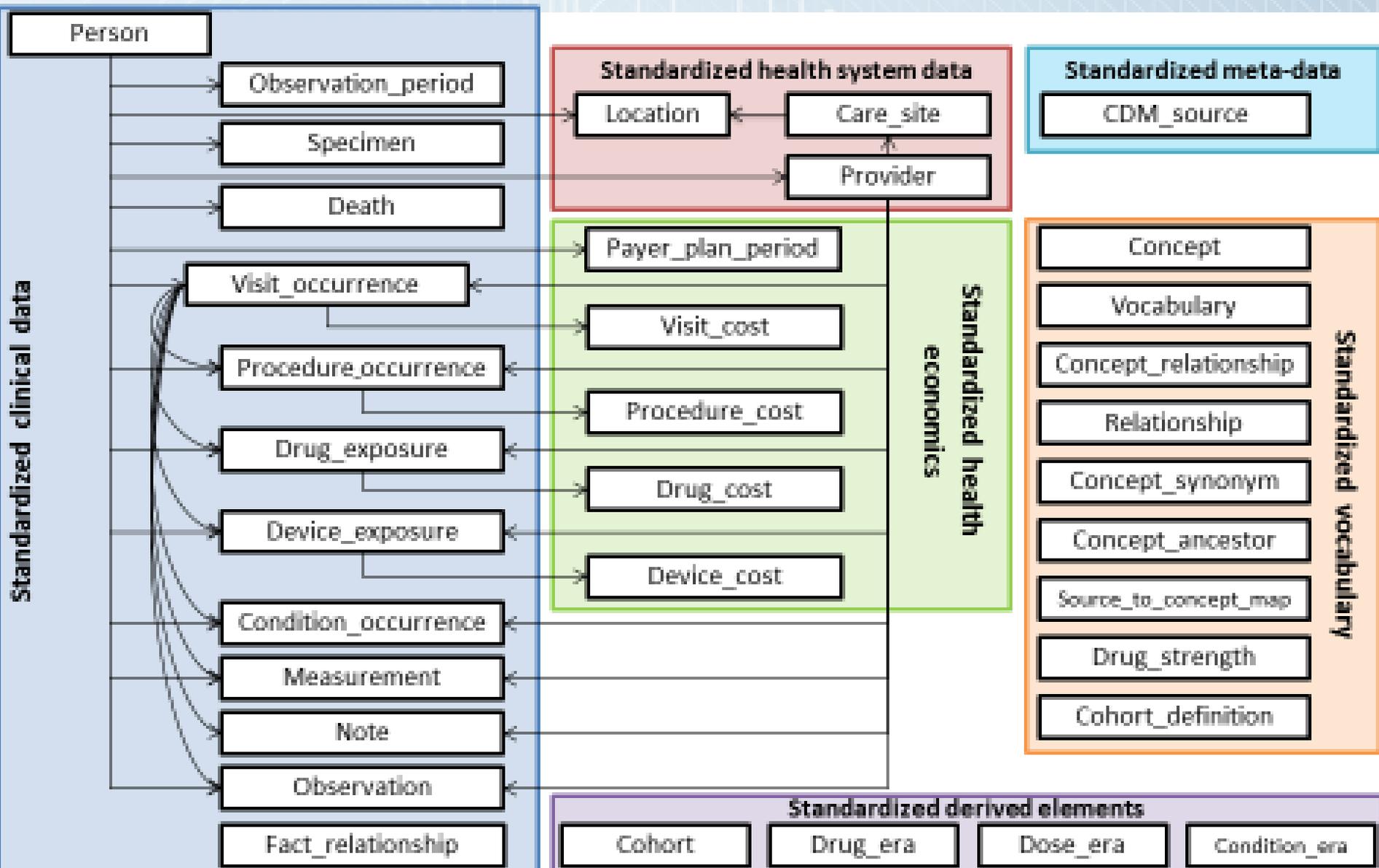


# OMOP

## ◆ OMOP – Observational Medical Outcomes Partnership



# OMOP Common Data Model



- Standardized Clinical Data Tables
  - PERSON
  - OBSERVATION\_PERIOD
  - SPECIMEN
  - DEATH
  - VISIT\_OCCURRENCE
  - PROCEDURE\_OCCURRENCE
  - DRUG\_EXPOSURE
  - DEVICE\_EXPOSURE
  - CONDITION\_OCCURRENCE
  - MEASUREMENT
  - NOTE
  - NOTE\_NLP (V5 5.2)
  - OBSERVATION
  - FACT\_RELATIONSHIP
- Standardized Health System Data Tables
  - LOCATION
  - CARE\_SITE
  - PROVIDER
- Standardized Health Economics Data Tables
  - PAYER\_PLAN\_PERIOD
  - COST (V5.0.1)
  - VISIT\_COST - removed
  - PROCEDURE\_COST - removed
  - DRUG\_COST - removed
  - DEVICE\_COST - removed
- Standardized Derived Elements
  - COHORT
  - COHORT\_ATTRIBUTE
  - DRUG\_ERA
  - DOSE\_ERA
  - CONDITION\_ERA

- Standardized Vocabularies
  - CONCEPT
  - VOCABULARY
  - DOMAIN
  - CONCEPT\_CLASS
  - CONCEPT\_RELATIONSHIP
  - RELATIONSHIP
  - CONCEPT\_SYNONYM
  - CONCEPT\_ANCESTOR
  - SOURCE\_TO\_CONCEPT\_MAP
  - DRUG\_STRENGTH
  - COHORT\_DEFINITION
  - ATTRIBUTE\_DEFINITION
- Standardized meta-data
  - CDM\_SOURCE

# Common data models in action



COLLOQUIUM  
PAPER

## Characterizing treatment pathways at scale using the OHDSI network

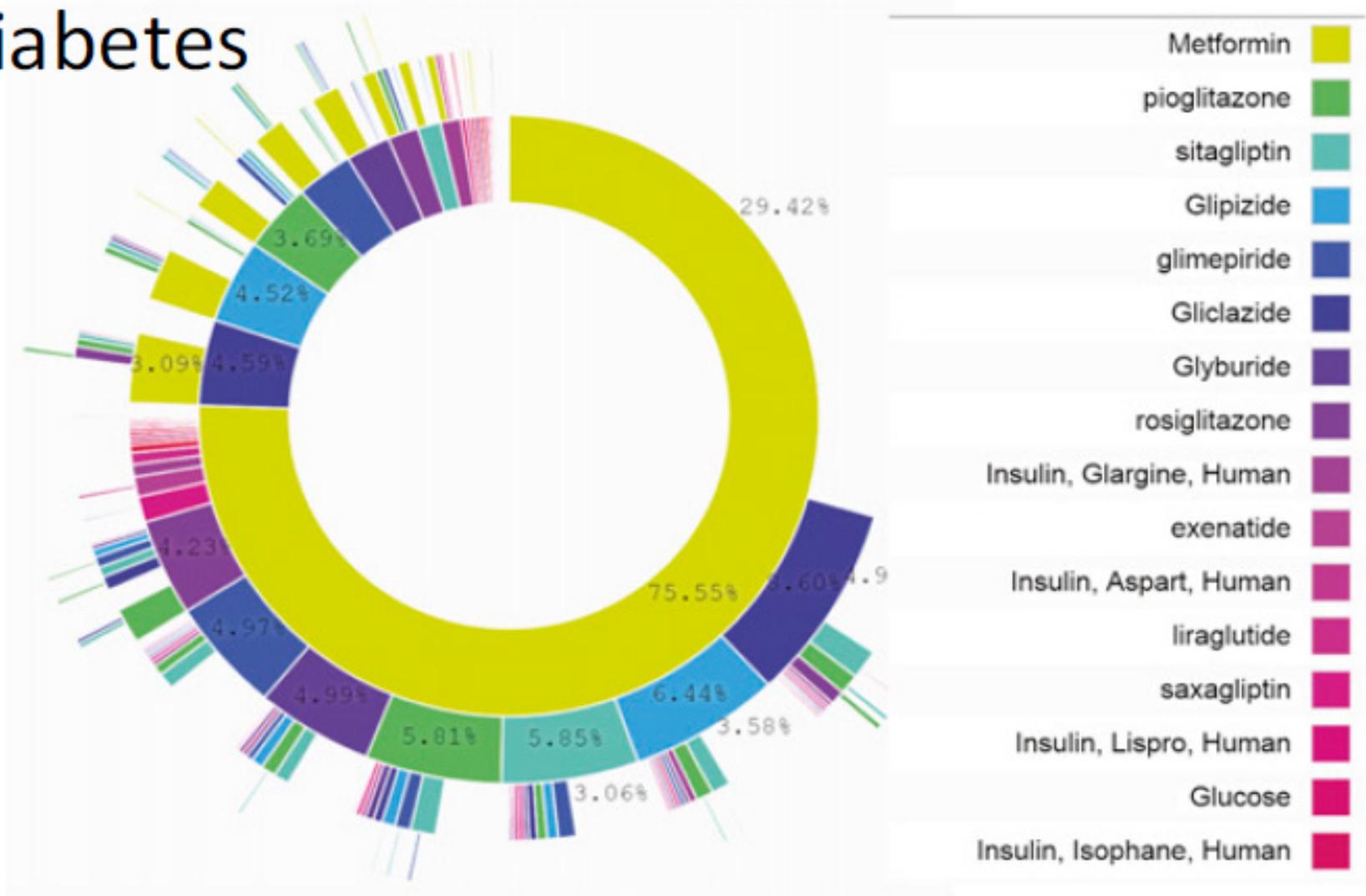
George Hripcsak<sup>a,b,c,1</sup>, Patrick B. Ryan<sup>c,d</sup>, Jon D. Duke<sup>c,e</sup>, Nigam H. Shah<sup>c,f</sup>, Rae Woong Park<sup>c,g</sup>, Vojtech Huser<sup>c,h</sup>, Marc A. Suchard<sup>c,i,j,k</sup>, Martijn J. Schuemie<sup>c,d</sup>, Frank J. DeFalco<sup>c,d</sup>, Adler Perotte<sup>a,c</sup>, Juan M. Banda<sup>c,f</sup>, Christian G. Reich<sup>c,l</sup>, Lisa M. Schilling<sup>c,m</sup>, Michael E. Matheny<sup>c,n,o</sup>, Daniella Meeker<sup>c,p,q</sup>, Nicole Pratt<sup>c,r</sup>, and David Madigan<sup>c,s</sup>

[www.pnas.org/cgi/doi/10.1073/pnas.1510502113](http://www.pnas.org/cgi/doi/10.1073/pnas.1510502113)

PNAS | July 5, 2016 | vol. 113 | no. 27 | 7329–7336



# A Diabetes



*Biomedical ontology in action*  
*Part 4*

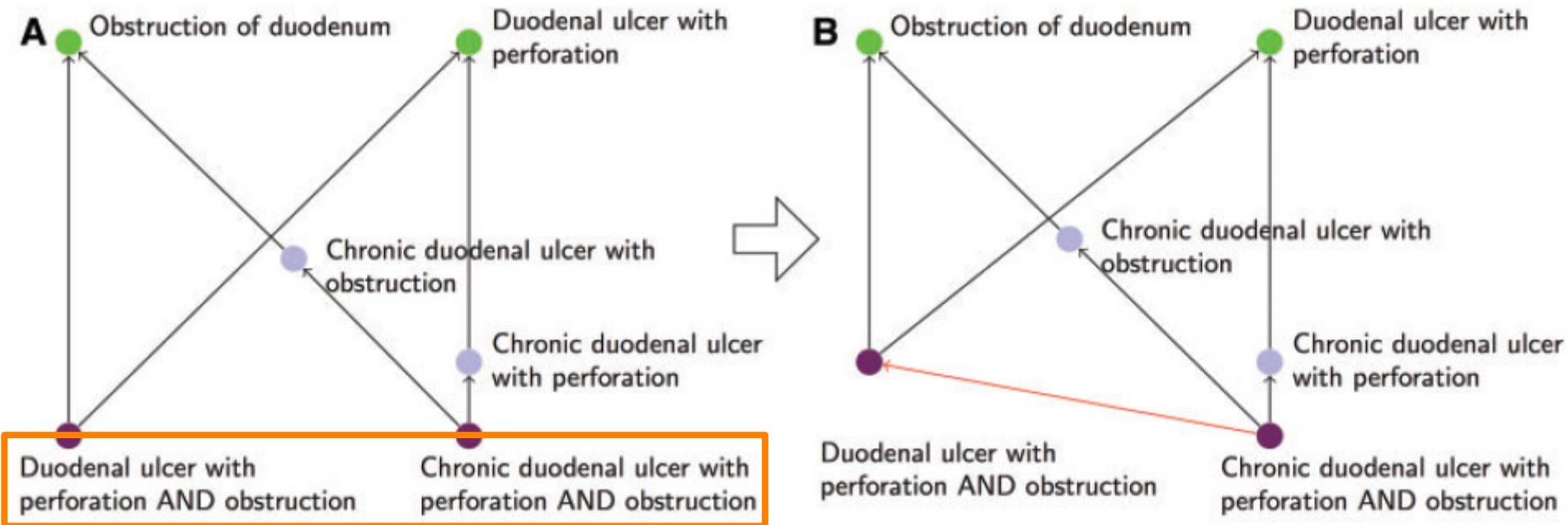
**Terminology research at NLM**

*Examples of terminology-related projects*

# Quality assurance in SNOMED CT

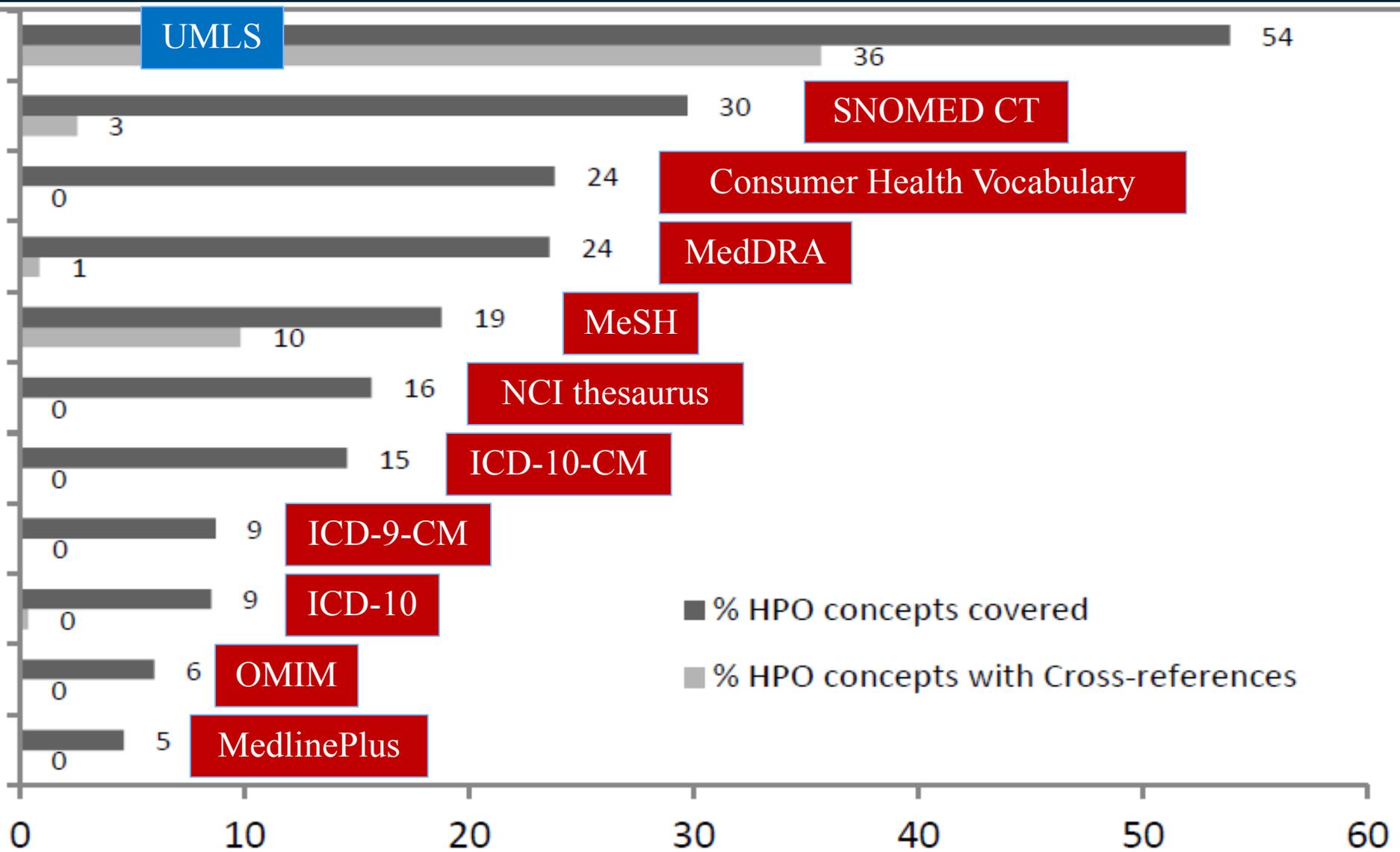
Non-lattice subgraph

Suggested remediation

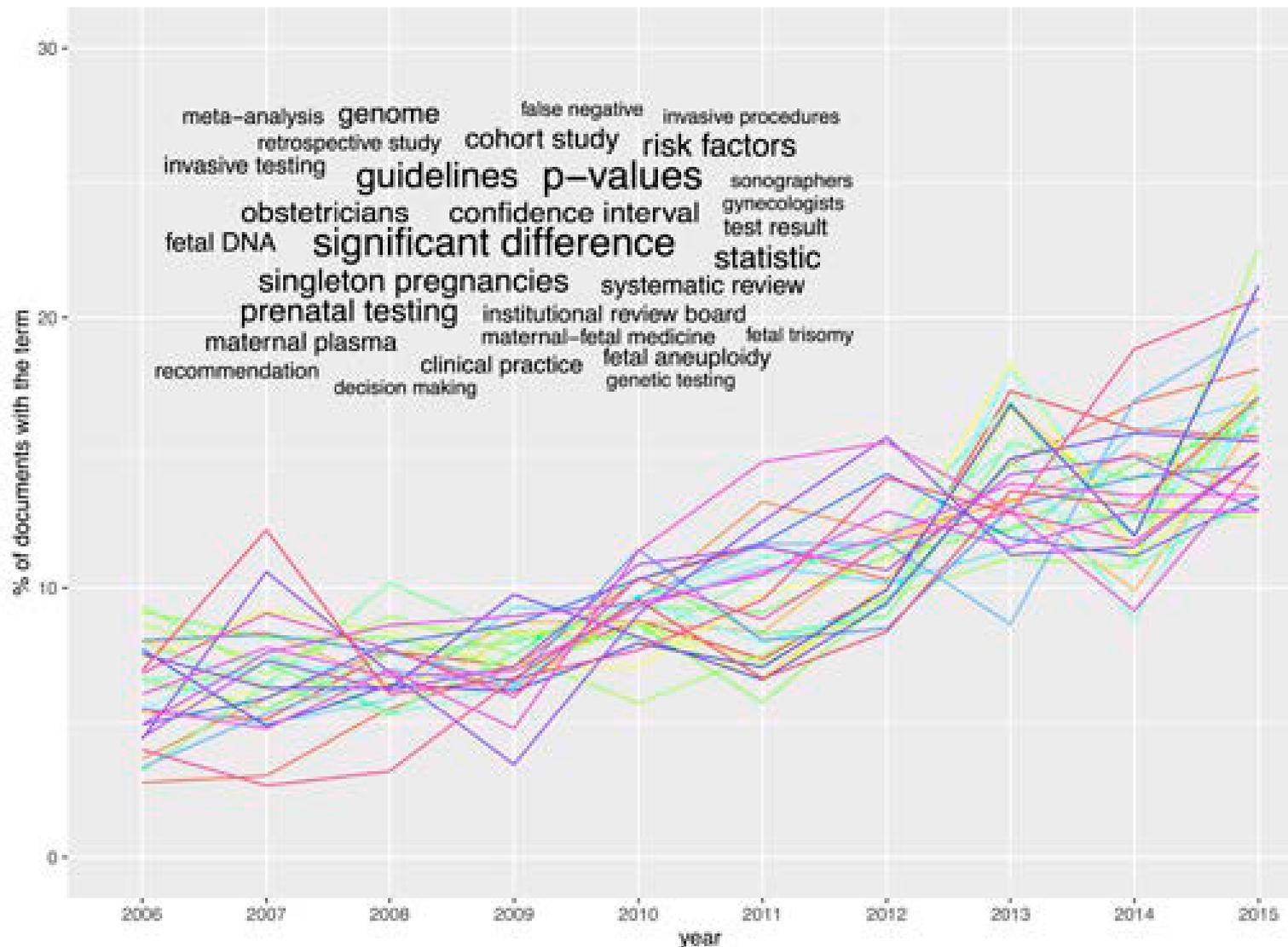


Duodenal ulcer with perforation AND obstruction  $\supset$  Chronic duodenal ulcer with perforation AND obstruction

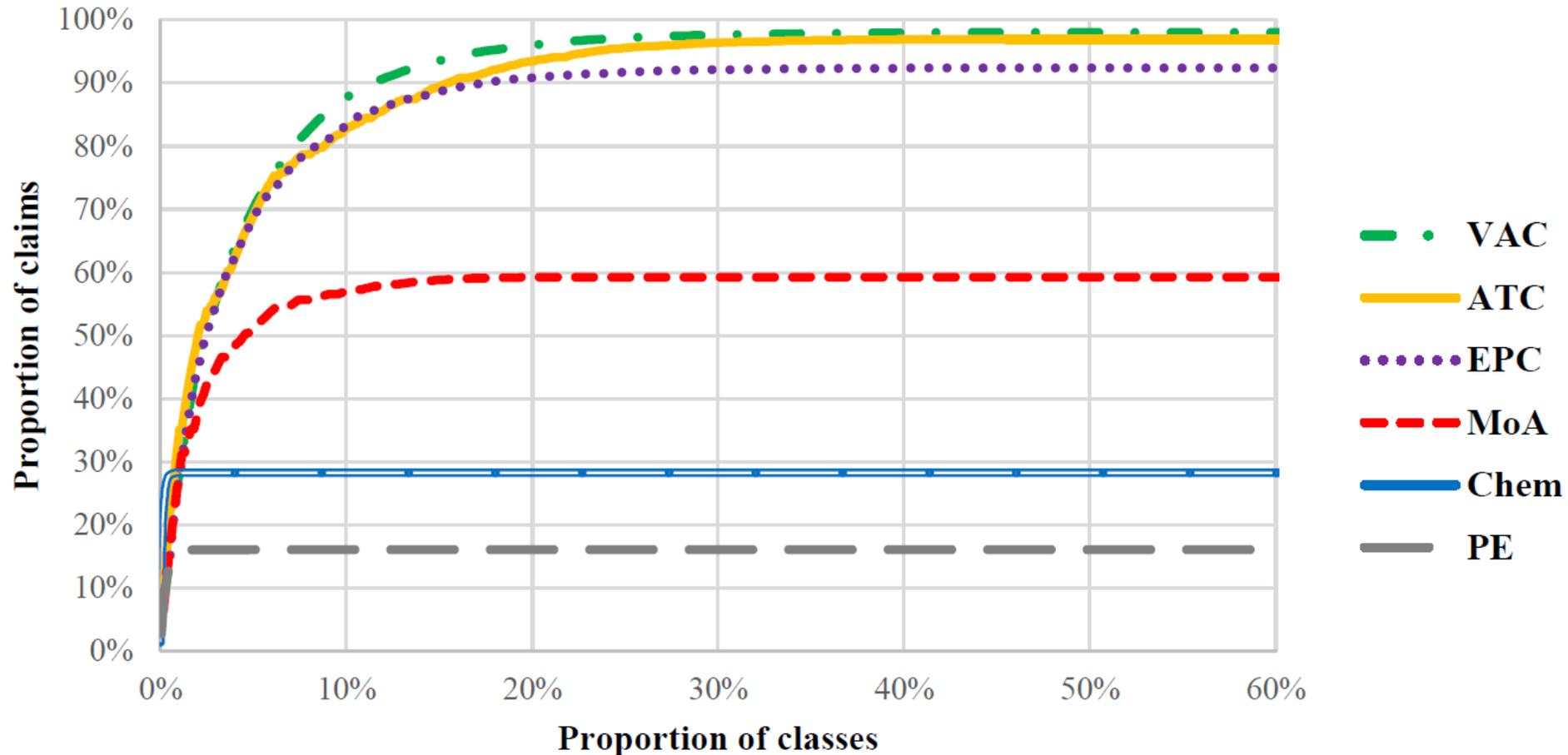
# Coverage of phenotypes



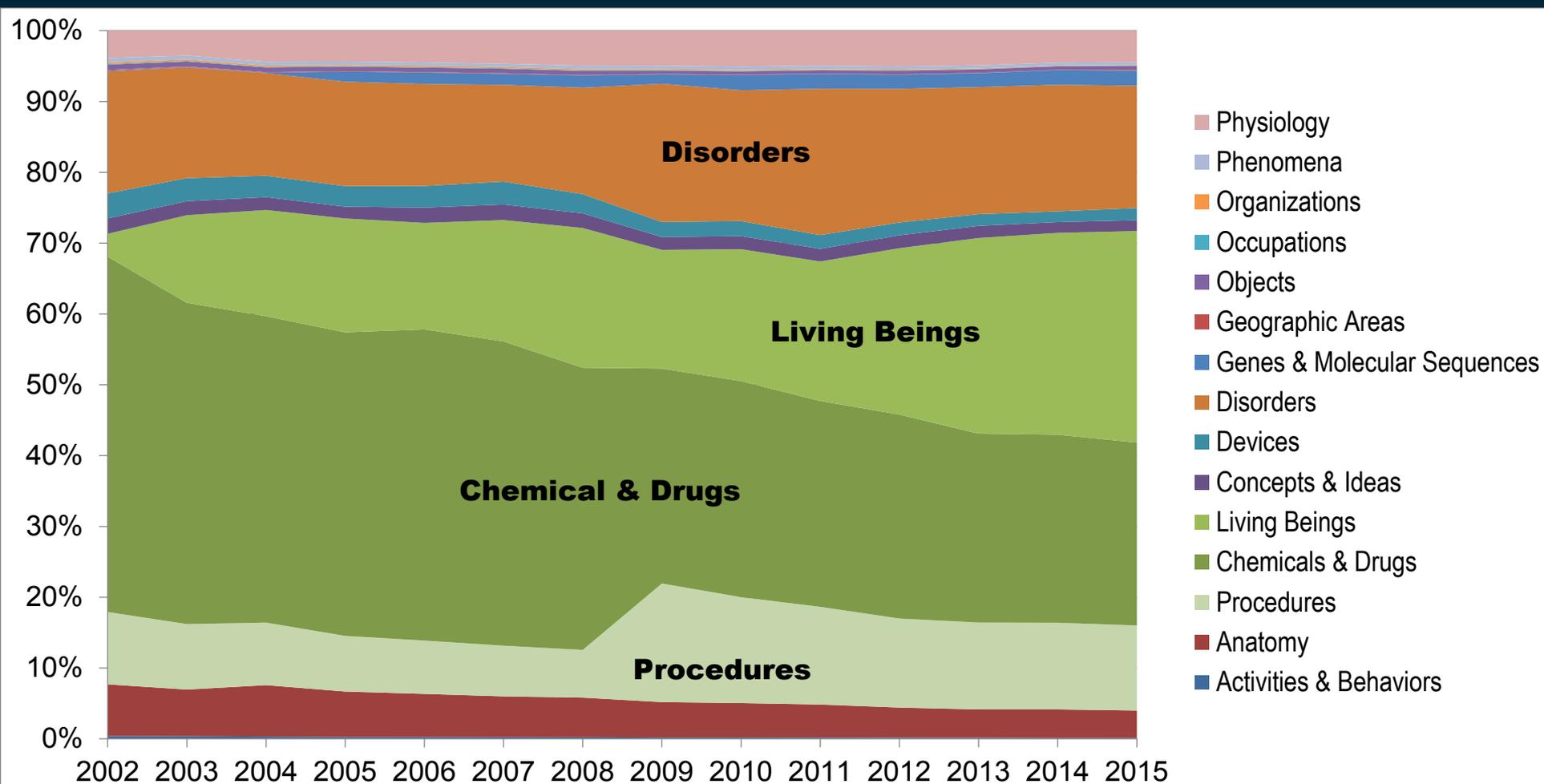
# Identifying terms for Fetal Medicine



# Suitability of drug classification systems



# Evolution of the UMLS Metathesaurus



# References Review articles

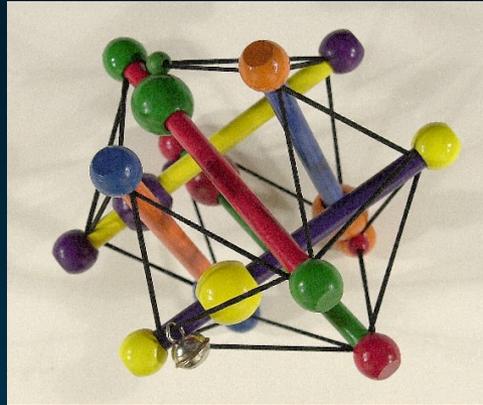
- ◆ Bodenreider O, Stevens R. Bio-ontologies: current trends and future directions. *Brief Bioinform.* 2006 Sep;7(3):256-74.
- ◆ Cimino JJ, Zhu X. The practical impact of ontologies on biomedical informatics. *Yearb Med Inform.* 2006:124-35.
- ◆ Bodenreider O. Biomedical ontologies in action: role in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