

Framing the Conversation: Ontologies within Semantic Interoperability Ecosystems

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Ontologies support semantic interoperability in healthcare

Two use cases in information exchange and analytics



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Outline

- ◆ The ecosystem of ontologies for healthcare
 - Clinical ontologies
 - Ontology integration systems
- ◆ Two use cases
 - Information exchange
 - Value sets for Clinical Quality Measures
 - Analytics
 - Analysis of large distributed clinical data warehouses

The ecosystem of ontologies for healthcare

Biomedical ontologies

◆ Structural perspective

[J. Cimino, YBMI 2006]

- What are they (vs. what are they for)?

◆ “High-impact” biomedical ontologies

- 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)



Biomedical ontologies

◆ Functional perspective

[Bodenreider, YBMI 2008]

- 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 reasoning** (data selection and aggregation, decision support, natural language processing applications, knowledge discovery).



LOINC



- ◆ Type: Controlled terminology*
- ◆ Domain: Laboratory and clinical observations
- ◆ Developer: Regenstrief Institute
- ◆ Availability
 - Publicly available: Yes
 - Repositories: UMLS, BioPortal
- ◆ Size: > 50k codes
- ◆ Uses: information exchange (e.g., HL7 messages)
- ◆ URL: www.regenstrief.org/loinc/loinc.htm



LOINC Example

- ◆ *Sodium:SCnc:Pt:Ser/Plas:Qn (2951-2)*
[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	--

SNOMED CT

SNOMED CT

The Global Language of Healthcare

- ◆ Type: Reference terminology / ontology
- ◆ Domain: Clinical medicine
- ◆ Developer: IHTSDO
- ◆ Availability
 - Publicly available: Yes* (in member countries)
 - Repositories: UMLS, BioPortal
- ◆ Size: > 300k concepts
- ◆ Uses: clinical documentation, analytics
- ◆ URL: <http://www.ihtsdo.org/>



SNOMED CT Example

The screenshot displays the IHTSDO SNOMED CT Browser interface. At the top, the title is "IHTSDO SNOMED CT Browser". Navigation buttons include "Release: us-edition v20150901", "Perspective: Full", "Feedback", "About", and a language selector (US flag). The main content area is divided into a left sidebar and a right main panel. The sidebar shows a "Taxonomy" view with a tree structure of SNOMED CT concepts. The main panel shows "Concept Details" for "Congenital hypoplasia of kidney (disorder)".

Left Sidebar: Taxonomy

- 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)

Main Panel: Concept Details

Parents

- Congenital anomaly of the kidney (disorder)

Selected Concept: Congenital hypoplasia of kidney (disorder)

SCTID: 32659003
32659003 | Congenital hypoplasia of kidney (disorder) |

Renal hypoplasia
Congenital hypoplasia of kidney
Congenital hypoplasia of kidney (disorder)

Occurrences:
Occurrence → Congenital
Associated morphology → Hypoplasia
Finding site → Kidney structure

Children (7)

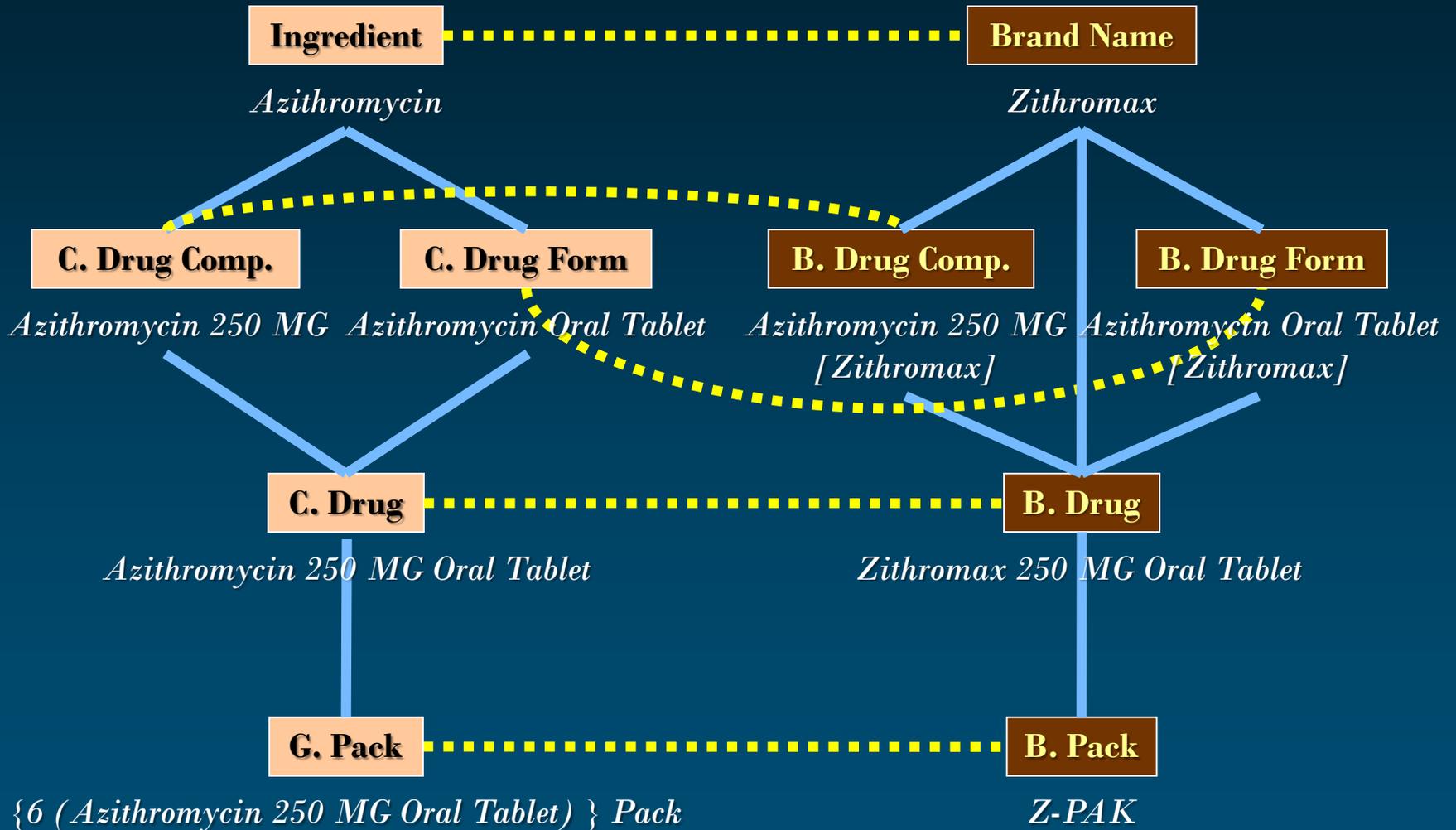
- Ask-Upmark kidney (disorder)
- Bilateral renal hypoplasia (disorder)
- Congenital hypoplasia of renal papilla (disorder)
- Congenital hypoplasia of renal pelvis (disorder)
- Congenital small renal papilla (disorder)
- Familial hypoplastic, glomerulocystic kidney (disorder)
- Oligomeganephronic hypoplasia of kidney (disorder)

RxNorm

- ◆ Type: Controlled terminology
- ◆ Domain: Drug names
- ◆ Developer: NLM
- ◆ Availability
 - Publicly available: Yes*
 - Repositories: UMLS, BioPortal
- ◆ Size: > 10k ingredients; 19k clinical drugs
- ◆ Uses: e-prescription, information exchange
- ◆ URL: <http://www.nlm.nih.gov/research/umls/rxnorm/>



RxNorm Example

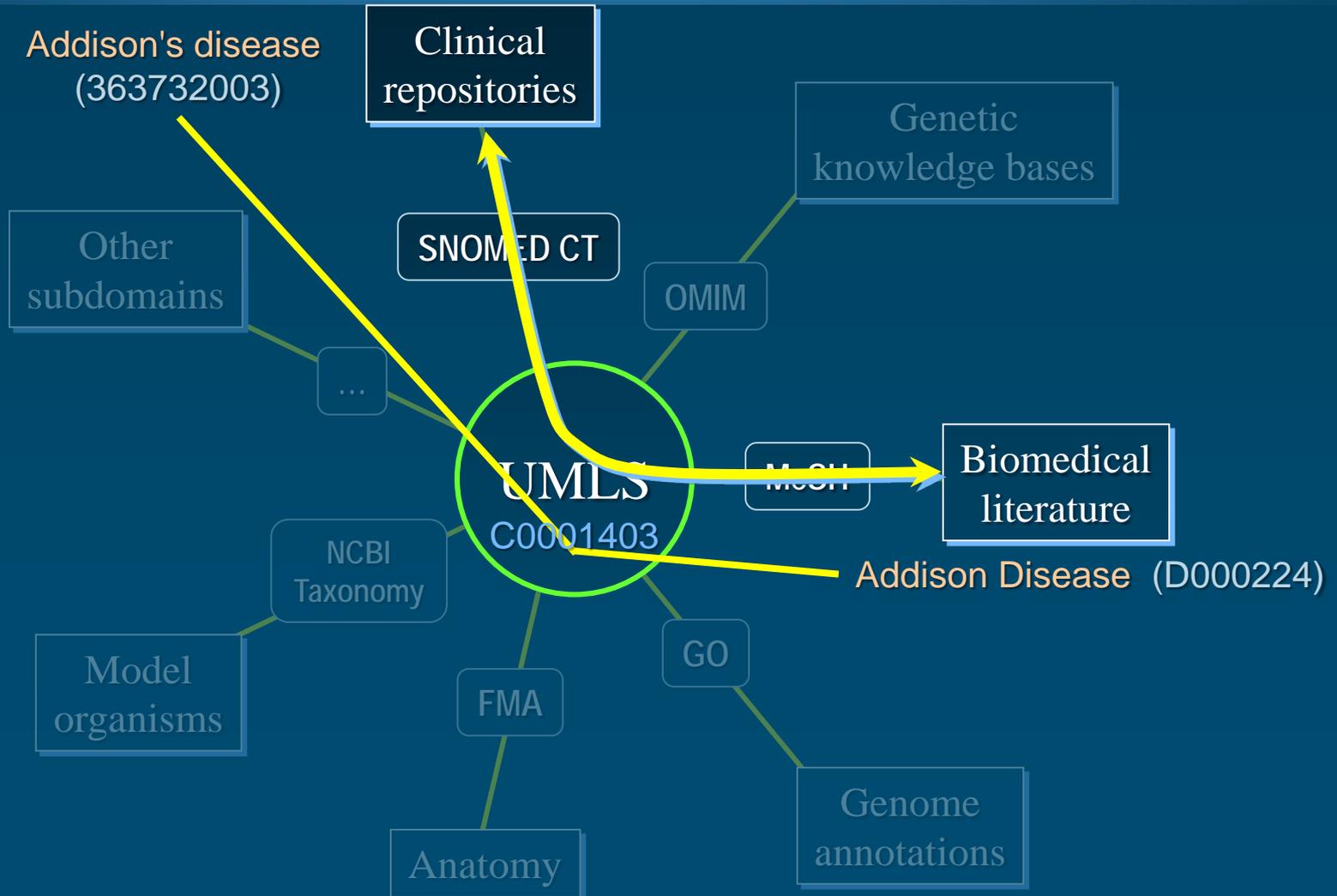


Unified Medical Language System

- ◆ Type: Terminology integration system
- ◆ Domain: Biomedicine
- ◆ Developer: NLM
- ◆ Availability
 - Publicly available: Yes*
 - Repositories: also available through BioPortal
- ◆ Size: 140 source vocabularies; 3.1M concepts
- ◆ Uses: annotation, NLP, cross-walk, ...
- ◆ URL: <http://www.nlm.nih.gov/research/umls/>



UMLS Example



Use case #1
Information exchange

Value sets for Clinical Quality Measures

Information exchange

- ◆ “Meaningful Use” incentive program
 - Use of certified electronic health record (EHR) systems
 - Requires use of select biomedical terminologies
 - For information exchange (e-prescribing, lab results)
 - For quality purposes (clinical quality measures)
 - Rely on reference value sets



Reference value sets

eMeasure Title	Venous Thromboembolism Prophylaxis
Description	This measure assesses the number of patients who received VTE prophylaxis or have documentation why no VTE prophylaxis was given the day of or the day after hospital admission or surgery end date for surgeries that start the day of or the day after hospital admission.

- **\$MedicationVTEProphylaxis =**
 - Union of:
 - "Medication, Administered: **Low Dose Unfractionated Heparin for VTE Prophylaxis**"
 - "Medication, Administered: Low Molecular Weight Heparin for VTE Prophylaxis"
 - "Medication, Administered: Injectable Factor Xa Inhibitor for VTE Prophylaxis"
 - "Medication, Administered: Warfarin"

Search Results
Value Set Details

Value Set Information Available Updates: Approved By Steward Expansion Versions: 2 MU2 Update 2015-05-01

Metadata

Name:
Low Dose Unfractionated Heparin for VTE Prophylaxis

OID:
2.16.840.1.113762.1.4.1045.39

Code	Descriptor	Code System
1361568	heparin sodium, porcine 2000 UNT/ML Injectable Solution	RXNORM
1361574	heparin sodium, porcine 20000 UNT/ML Injectable Solution	RXNORM
1361577	heparin sodium, porcine 2500 UNT/ML Injectable Solution	RXNORM
1361615	heparin sodium, porcine 5000 UNT/ML Injectable Solution	RXNORM
1361853	0.5 ML heparin sodium, porcine 10000 UNT/ML Prefilled Syringe	RXNORM
1362831	heparin sodium, porcine 10000 UNT/ML Injectable Solution	RXNORM
1362837	heparin sodium, porcine 12500 UNT/ML Injectable Solution	RXNORM



<https://vsac.nlm.nih.gov/>

Use case #2

Analytics

*Analysis of large distributed clinical data
warehouses*

Analytics

- ◆ Clinical data warehouses
 - Distinct from EHR systems
- ◆ “ETL” (extract – transform – load) processes
 - Data normalized to “standards”
 - Local data mapped to ontologies
 - Facilitated by ontology integrations systems (e.g., UMLS)
- ◆ Analysis leverages hierarchical and other relations
 - Transitive closures
- ◆ Facilitates analysis of large-scale data repositories
 - Including distributed repositories across institutions

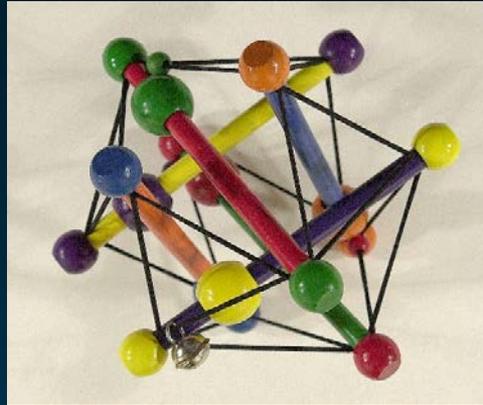


- ◆ “Multi-stakeholder, interdisciplinary collaborative to bring out the value of health data through large-scale analytics”
- ◆ OMOP Common Data Model
 - Standard vocabularies (e.g., LOINC, SNOMED CT, RxNorm)
- ◆ Investigation of treatment pathways
 - For 3 chronic diseases (3 year-follow up)
 - > 1M patients with hypertension
 - Across multiple clinical institutions
 - In several countries

References Review articles

- ◆ Bodenreider O, Stevens R. Bio-ontologies: current trends and future directions. *Brief Bioinform.* 2006 Sep;7(3):256-74.
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- ◆ Bodenreider O. Biomedical ontologies in action: role in knowledge management, data integration and decision support. *Yearb Med Inform.* 2008:67-79.





Medical Ontology Research

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