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
  - 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)

[J. Cimino, YBMI 2006]
Biomedical ontologies

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

[Bodenreider, YBMI 2008]
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](http://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]

<table>
<thead>
<tr>
<th>Axis</th>
<th>Value</th>
</tr>
</thead>
<tbody>
<tr>
<td>Component</td>
<td>Sodium</td>
</tr>
<tr>
<td>Property</td>
<td>SCnc – Substance Concentration (per volume)</td>
</tr>
<tr>
<td>Timing</td>
<td>Pt – Point in time (Random)</td>
</tr>
<tr>
<td>System</td>
<td>Ser/Plas – Serum or Plasma</td>
</tr>
<tr>
<td>Scale</td>
<td>Qn – Quantitative</td>
</tr>
<tr>
<td>Method</td>
<td>--</td>
</tr>
</tbody>
</table>
SNOMED CT

- 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

http://browser.ihtsdotools.org/

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)

Parents
- Congenital anomaly of the kidney (disorder)

- Congenital hypoplasia of kidney (disorder)
  - 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)
- Oligomeganephric 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
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, …
UMLS Example

Addison's disease (363732003)

Other subdomains

Clinical repositories

SNOMED CT

OMIM

Biomedical literature

Addison Disease (D000224)

Genetic knowledge bases

Model organisms

FMA

NCBI Taxonomy

Anatomy

Genome annotations

GO
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

<table>
<thead>
<tr>
<th>eMeasure Title</th>
<th>Venous Thromboembolism Prophylaxis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Description</td>
<td>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.</td>
</tr>
</tbody>
</table>

$\text{Medication} \text{VTE Prophylaxis} =$

- 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"

Metadata

- Name: Low Dose Unfractionated Heparin for VTE Prophylaxis
- OID: 2.16.840.1.113762.1.4.1045.39

<table>
<thead>
<tr>
<th>Code</th>
<th>Descriptive</th>
<th>Code System</th>
</tr>
</thead>
<tbody>
<tr>
<td>1361568</td>
<td>heparin sodium, porcine 2000 UNT/ML Injectable Solution</td>
<td>RXNORM</td>
</tr>
<tr>
<td>1361574</td>
<td>heparin sodium, porcine 20000 UNT/ML Injectable Solution</td>
<td>RXNORM</td>
</tr>
<tr>
<td>1361577</td>
<td>heparin sodium, porcine 2500 UNT/ML Injectable Solution</td>
<td>RXNORM</td>
</tr>
<tr>
<td>1361615</td>
<td>heparin sodium, porcine 5000 UNT/ML Injectable Solution</td>
<td>RXNORM</td>
</tr>
<tr>
<td>1361853</td>
<td>0.5 ML heparin sodium, porcine 10000 UNT/ML Prefilled Syringe</td>
<td>RXNORM</td>
</tr>
<tr>
<td>1362831</td>
<td>heparin sodium, porcine 10000 UNT/ML Injectable Solution</td>
<td>RXNORM</td>
</tr>
<tr>
<td>1362837</td>
<td>heparin sodium, porcine 12500 UNT/ML Injectable Solution</td>
<td>RXNORM</td>
</tr>
</tbody>
</table>

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. **Biomedical ontologies in action: role in knowledge management, data integration and decision support.** Yearb Med Inform. 2008:67-79.
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