Practical Applications of Ontologies in Clinical Systems

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Overview

• Background
  – Clinical System
  – Clinical Ontologies (disclaimer)

• Practical applications
  – Real-life examples from Partners
  – Local curation and management

• Next generation of clinical systems
  – Meaningful use, collaborative care
  – Continuous learning

• Conclusions
Background

Clinical System

Clinical Ontologies (disclaimer)
Clinical System

• “... an automated system with a long term database containing clinical information used for patient care.”

  – Bruce Blum, 1986

• Support (automation) for one or multiple clinical (patient care) functions

• Electronic Health Record system is an integrated suite of clinical systems
Outpatient EHR @ Partners

Document, ordering, results review, messaging, etc.
Clinical Ontologies

• “... a loose definition of Clinical Ontology, which also includes well-organized, but not always formally represented, clinical classifications, nomenclatures and terminologies.”

• “Clinical Ontologies represent clinical phenotypes, diseases, syndromes and many other clinical elements such as medications and personal habits ...”

SNOMED CT

- Systematized Nomenclature of Medicine – Clinical Terms
- Organization: International Health Terminology Standards Development Organisation (IHTSDO)
  - SNOMED Terminology Solutions - College of American Pathologists
- Purpose: Encoding of multiple clinical domains
- Content: Comprehensive (diseases, signs, symptoms, living organisms, chemicals, body parts, morphology, occupations, modifiers, etc.)
- Information:
  - http://www.ihtsdo.org/
CliniClue Xplore

http://www.clininfo.co.uk/clinicue_xplore/concepts/browserLayoutBrowser.html
• Logical Observation Identifiers Names and Codes
• Organization: LOINC Committee
• Purpose: identification of laboratory and clinical observations (HL7 messages)
• Content: laboratory tests, clinical measurements, documents, etc.
• Information:
  – http://loinc.org/
<table>
<thead>
<tr>
<th>Score</th>
<th>LOINC</th>
<th>Component</th>
<th>Property</th>
<th>Timing</th>
<th>System</th>
<th>Scale</th>
<th>Method</th>
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<td>Qn</td>
<td>Per age</td>
<td></td>
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<td>Per age and gender</td>
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<td>Qn</td>
<td>US.estimated from AC</td>
<td>g</td>
</tr>
</tbody>
</table>
Many others (incomplete list)

- **RxNorm**: clinical drugs and drug delivery devices (NLM)
- **ICNP**: International Classification For Nursing Practice (ICN)
- **NDF-RT**: National Drug File - Reference Terminology (VA)
- **CVX**: Vaccines Administered (CDC)
- **ICD-9-CM, ICD-10-CM/ICD-10-PCS**: International Classification of Diseases
- **CPT-4**: Current Procedural Terminology (AMA)
- **HL7 Vocabulary domains** (messaging, documents, services)
Practical applications

Examples from Partners HealthCare:
(1) Problem Lists; (2) Bedside Documentation

Local curation and management
1st Example: Problem List

- Management of **patient-specific problems** (as a list):
  - All active (and inactive) problems associated with a patient
  - Detailed “provenance” (source, onset, changes, status, etc.)
  - Associate problems with encounters, orders, medications, notes, etc.
  - Order (filter) the problem list

- Problems correspond to chronic conditions, diagnoses, symptoms, functional limitations, and visit-specific conditions
  - Managed over time (e.g., single visit, life of a patient)
  - Documentation of historical information
  - Tracking the changing character of problems and their priority

- **Multiple disciplines** can contribute to the problem list

Adapted from HL7 Electronic Health Record - System Functional Model, Release 1 February 2007; Chapter Three: Direct Care Functions.
### Problem List @ Partners (1)

<table>
<thead>
<tr>
<th>Problem Description</th>
<th>Additional Details</th>
</tr>
</thead>
<tbody>
<tr>
<td>Depression</td>
<td>Type: Acute Severity: Major Condition: Worse. Comments: no changes pt still depressed…</td>
</tr>
<tr>
<td>Brain cancer</td>
<td>Comments: inoperable</td>
</tr>
<tr>
<td>Diabetes mellitus</td>
<td>Onset: 04/04/2011 Comments: must reduce their stress an… Type: Chronic Onset: 11/30/2007</td>
</tr>
<tr>
<td>Hypertension</td>
<td>Type: Acute Onset: 09/11/2001 Comments: updating a problem that was… Type: Acute Severity: Minor Condition: Unchanged Location: Right Onset: 03/02/2011 Comments: hurts</td>
</tr>
<tr>
<td>R/O Depression</td>
<td></td>
</tr>
<tr>
<td>Asthma</td>
<td>Type: Acute Onset: 09/11/2001 Comments: updating a problem that was… Type: Acute Severity: Minor Condition: Unchanged Location: Right Onset: 03/02/2011 Comments: hurts</td>
</tr>
<tr>
<td>PR Foot pain</td>
<td></td>
</tr>
<tr>
<td>FH Dyspnea</td>
<td></td>
</tr>
<tr>
<td>H/O Bulimia nervosa</td>
<td></td>
</tr>
<tr>
<td>H/O Verruca plantaris</td>
<td></td>
</tr>
<tr>
<td>Glaucoma</td>
<td>Onset: 04/05/2011</td>
</tr>
<tr>
<td>RSK Melanocytic nevus of skin</td>
<td>Onset: 06/15/2011</td>
</tr>
<tr>
<td>RSK Arteriosclerotic vascular disease</td>
<td></td>
</tr>
</tbody>
</table>
Problem List @ Partners (2)

1. Problem: pain

2. Favorites: Term
   - H/O ankle pain
   - Results: Term
     - Pain
     - Abdominal pain
     - Knee pain
     - Chest pain
     - Neck pain
     - Shoulder pain
     - Joint pain
     - Foot pain
     - Ankle pain
     - Muscle pain
     - Flank pain
     - Musculoskeletal pain
     - Hip pain

3. Problem: Chest pain
   - Modifier:
     - Type:
     - Severity:
     - Conditions:
     - Status:
     - Location:
     - Question of:
     - History of:
     - Probable:
     - Negative:
     - Risk of:
     - Rule out:
     - Negative History of:

   Onset Date: T 4/12/13 Resolution Date: T 4/12/13

Add to Favorites for: My List Practice
Problem List @ Partners (3)

1. Problem: ketogenic

2. No Match Found. Take as typed or choose another Problem. ketogenic [Uncoded problem - will NOT be used for clinical decision support]

3. Problem Description: Ketogenic [uncoded]
Problem list concepts @ Partners

- **Initial phase (+1,200 terms)**
  - Controlled (limited) list of terms developed by Partners
  - Physician-centered
  - Limited synonyms; no classification or editorial policy

- **Current phase (+1,600 concepts)**
  - Terms have been mapped to **SNOMED CT** concepts
  - Additional synonyms
  - Concepts manually aggregated into reusable clinical states (classification subsets)
  - Evolving editorial policy (concept granularity)

- **Ongoing expansion – multidisciplinary**
  (currently +3,500 concepts)
Adoption of SNOMED CT

• Local terms were manually mapped to SNOMED CT
  – Based on the “VA/KP Problem Lists subset”
    ▪ Almost all successfully mapped – few ‘local’ concepts remain
  – Local identifiers were preserved for backwards compatibility
  – Ongoing maintenance with semi-annual SNOMED updates
  – Extensive use of the SNOMED hierarchies to create classification subsets used in decision support rules
Examples of Problem concepts

- Chronic Renal Dysfunction
  - Chronic renal failure syndrome
- Nephropathy
  - Kidney disease
- Cardiac bypass graft surgery
  - Coronary artery bypass graft
- Coronary artery disease
  - Coronary arteriosclerosis
- Diabetes mellitus
  - Diabetes mellitus
- Diabetes of pregnancy
  - Gestational diabetes mellitus
- G6PD deficiency
  - Deficiency of glucose-6-phosphate dehydrogenase
- Low platelets
  - Thrombocytopenic disorder
- Bright red blood per rectum
  - Hematochezia
- Lower GI bleeding
  - Lower gastrointestinal hemorrhage
- Unspecified GI bleed
  - Gastrointestinal hemorrhage
- Hypotension
  - Low blood pressure
- Peptic ulcer disease
  - Peptic ulcer
- Angioplasty
  - Percutaneous transluminal coronary angioplasty
- Pregnancy
  - Patient currently pregnant
- Unwanted fertility
  - Unplanned pregnancy
Classification subsets

- Grouping and filtering concepts (not in use)
  - User-interface, reporting and analytics

- Clinical **decision support rules**
  - Enable simple inferences that decrease the complexity of rules (maintenance)

- **Difficult** to create and maintain without more formal semantic representation
  - SNOMED hierarchies provide a starting point
  - Frequently require validation (local relevance)

- Ideally maintained at a national (international) level to ensure **shared understanding**
  - Collaborative development and maintenance
Management of subsets @ Partners
Management of subsets @ Partners
Benefits of SNOMED CT

- Detailed representation of clinical problems
  - **Consistent** set of concepts (enterprise view)
  - **Compositional** and fine-grained
- Broad **coverage** of clinical domains
- Improved term search (hierarchical views)
- Rich set of **relationships**: inference
- Active **maintenance** by international organization
- Mappings to billing classifications (ICD-9/10)
Implementation challenges

• Difficult reconciliation with pre-existing terms
  – Local ambiguity, redundancy, length restrictions
• Legacy codes ‘hard-coded’ into applications and decision support rules
  – Recently able to discontinue the generation of legacy identifiers
• SNOMED limitations
  – Terms are frequently not clinician-friendly
  – Inconsistencies caused by conflicting intents (over time)
  – Ongoing changes compromise stability (early adoption)
  – Lack of reference implementations (best practices)
Capturing relevant clinical details

- **Problem concept**
  - [Location]
    - Body site, Laterality, ...
  - [Etiology]
  - [Severity]
  - [Chronicity]
  - [...]  
  - [Modifiers]
    - History of, Family History of, Probable, Risk of, Rule out, Question of, Status Post, Negative Family History, ...

- **What is displayed** to the clinician (or patient)?
  - Simple keyword search that returns a list to terms
  - Form with multiple fields (multiple searches)

- **What is stored** in the patient problem list?
  - Single code representing a pre-coordinated concept
  - Multiple codes representing a concept expression

- **Current limitations:**
  - Clinical systems (free text?)
  - Clinical ontologies
Problem List information models

Alignment of Clinical Ontologies with models used by Clinical Systems is critical!
2nd Example: Bedside documentation

- Documentation of Care, Measurements and Results
  - Manage clinical **measurements**: document and annotate measurements of physiologic parameters and clinical conditions (e.g., vital signs, height, weight, I&O, pain severity, size of wound, etc.)
  - Manage clinical **documents** and **notes**: create, modify, and sign **unstructured** (narrative) and **structured** (templates with coded fields) documents and notes, including details about exams and procedures, assessments, and patient-specific care plans and instructions
  - **Medication** administration: list of medications (including vaccines) to be administered and administration details
  - Manage **results**: review, annotate, and communicate test results from ancillary departments or performed at the bedside

- **Multiple disciplines** contribute to bedside documentation
  - Large variety of clinical details typically represented in **narrative form**

Adapted from HL7 Electronic Health Record - System Functional Model, Release 1 February 2007; Chapter Three: Direct Care Functions.
### Bedside documentation @ Partners

**Initial Nursing Assessment**

- **Patient Profile**
  - Information source: Son-Michael
  - Preferred language: Spanish
  - Preferred name: Steven

- **Cognitive/Perceptual**
  - Sensory aides: Contact lenses, Eye glasses, Hearing aids
  - Other: Phone amplifier

- **Memory deficit**
  - Appointments, Completing tasks, Dates, Directions, Medication, Times

- **Orient to room**

- **Interpreter source**
  - Hospital interpreter
  - Telephonic service

- **Other**
  - Any additional information

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**Note:**

- **Show sessions log**
- **Include error sessions**

- **Date and Time:** 07/19/2011 11:22 AM (Saved at: 07/19/2011 11:40 AM) by [User Name]
Bedside documentation process

- **Concurrent** authoring for multiple disciplines
  - Overlap? Multiple ways to capture the same data?

- **Multiple restrictions** imposed by the documentation system
  - Very limited support for synonyms and reference clinical ontologies
  - Prevalence of pre-coordinated concepts (clinician-friendly)
  - Underlying information models not explicitly defined (no reuse)
    - Relations between data elements exist within data entry template (UI)
    - Context largely defined by data entry template (UI)

- Lack of **reference models** to inform what should be captured in coded format
  - Significant portions captured as free text within discrete fields
  - Compromises reporting and computerized decision support

- Alignment with reference standards **after** content is defined
  - Limited expertise to search and use reference clinical ontologies
Content definition

• Iteratively define content with stakeholders
  – Start with existing paper & electronic forms
  – Define what will be documented and how, including:
    ▪ Coded elements and their respective values
    ▪ Formulas and calculations
    ▪ Sequencing and disposition of elements
    ▪ Required vs. optional elements
  – *(Crosswalk with previously defined content - as needed)*
  – Iterate until reach consensus
Examples of Documentation concepts

- Easy Bruising
- Change in appetite
- Difficulty in Walking
- Heart Murmur
- Hearing Loss
- Ambulating
- Depressed
- Constipation
- Stool Consistency

- Reflexes: Babinski, right
- Motor strength: elbow extension, right
- Nephrostomy tube (right) insertion site
- Head of bed elevation
- Polyuria or polydipsia
- Rash/pruritus
- Redness
- Tolerating orals

Data types: true/false, free text, numeric, enumerated, etc.
Content modeling

- Extract **data elements & data values** from approved content
  - Name data elements using defined naming conventions
    - Preserve clinician-friendly labels
  - Classify data elements using defined categories (strict assignments)
  - **Index** (tag) data elements using applicable reference clinical ontologies: SNOMED, LOINC, ICNP, ... (enable subsequent retrieval)
  - **Map** coded data values to applicable reference clinical ontologies
  - *(Additional crosswalk with previously defined data elements – as needed)*
  - Iterate until all data elements and values are properly defined

- Update (import) data elements within documentation system
  - Current phase of content development: **+6,500 data elements**
Birth Weight: `<number><units>`

**LOINC**
8339-4: Body weight at birth
Mass; Pt; Patient; Qn; Measured

**Data Element:** numeric measurement with unit

**Topic**

**Value set**

SNOMED CT (or UCUM)
258681007: Units of mass (SI)

**Value (concept)**

SNOMED CT
258682000: gram, g

**Linking to reference clinical ontologies**
Indexing & Mapping sources

• LOINC
  – Data elements (1\textsuperscript{st} choice)
  – Documents and notes (1\textsuperscript{st} choice)

• SNOMED CT
  – Data values (1\textsuperscript{st} choice)
  – Data elements (2\textsuperscript{nd} choice)

• ICNP
  – Nursing problems, outcomes, interventions

• Others (Nutrition)
Benefits of Indexing and Mappings

• Availability of structured and coded data
  – Consistency across sites and disciplines
  – Identify (prevent) data redundancy (streamline workflow)
  – External confirmation that data content is relevant
  – Simplify data reporting (across clinical systems)
  – Enables advanced computerized decision support
  – Quality of the resulting clinical data (analysis & research)

• Compliance with efforts to promote interoperability
  – Data exchange and reporting
  – Import (adopt) templates and forms developed by others

• Contribute to the development and improvement of existing clinical ontologies
Terminology teams @ Partners

• Terminology engineers (4.0 FTE)
• Clinical Informaticians (2.6 FTE)
• Subject Matter Experts (domain specific)

• Software engineers (3.0 FTE)
• Project Manager (1.5 FTE)
Clinical Ontologies: advantages (1)

- Provide guidance (basis) for:
  - Concepts
  - Synonyms & Codes (‘Designations’)
  - Hierarchies & Classes
  - Mappings & Decompositions
  - Translations to other languages

- Required platform for data & knowledge interoperability
Clinical Ontologies: advantages (2)

- Contribute computable underpinnings for content **maintenance**
  - Advanced inference leveraging logic-based knowledge (e.g., SNOMED CT)

- Reduce local maintenance burden
  - Assuming **compatible** rate of change
Clinical Ontologies: limitations

• Must support local **customizations**
  – Concepts, designations, additional relationships

• Must accommodate **changes**
  – Reconcile concepts added locally with eventual availability in reference clinical ontologies
  – Reference clinical ontologies might evolve at incompatible speeds (too fast/slow)

• Must support concepts composed from **different sources**
  – Most clinical systems require concurrent/integrated use of multiple reference clinical ontologies
Core Principle @ Partners

• All reference clinical ontologies (e.g., LOINC, SNOMED, FDB, RxNorm, etc.) will be used by clinical systems through local Partners concepts
  – Concepts used by clinical systems and knowledge content are always local Partners concepts
  – Local concepts can be mapped to reference concepts in clinical ontologies
Core Principle: Motivation

- Local concepts will be created for all domains
  - Overcome **content coverage limitations** of clinical ontologies
  - Support research activities that require highly specialized content
  - Commitment to **submit** local extensions to organizations maintaining the reference clinical ontologies

- Local concepts will be **customized** as needed
  - Including ‘granularity’, designations, and associations
  - Consistent metadata and lifecycle management (unified *metamodel*)

- Local concepts will have stable **identifiers**
  - Internally defined and long-lived
  - Appropriate versioning and mappings to/from reference concepts

- Mappings to external concepts will occur as needed (**parsimonious**)
  - Enable resolution of overlapping content from different clinical ontologies

- Curation will follow KM lifecycle and collaboration best practices
Core Principle: Challenges

• Local and reference concepts must be complementary
  – Adopt semantic technologies for effective maintenance and inference
  – Manage local extensions, restrictions, and replacements (overrides)
  – No intent to replicate all reference designations and associations

• Adoption of compositional identifiers
  – Support for versioning and namespaces
  – Consistent with other knowledge assets (e.g., models, templates, rules, etc.)

• Mechanism to identify specific designations and associations
  – Proper support for classification (grouping) and contextual constraints

• Long-term stability and overall consistency outweigh maintenance
  – Recognize that local ontology maintenance ‘never ends’
  – Knowledge maintenance and software maintenance will be streamlined, while enabling interoperability and extensibility (innovation)
Next generation of clinical systems

Meaningful use

“Medical Home”

Continuous learning
Meaningful use of EHRs

- Universal use of EHRs by 2014
- Transformation of the healthcare system – improvements to outcomes and efficiency
- Requires “meaningful use” of EHRs, not just installation of the software
  - Incentive payments totaling up to $27 billion over 10 years
  - As much as $44,000 (through Medicare) and $63,750 (through Medicaid) per clinician
  - Incentives encourage early adoption; no incentives after 2014; Penalties begin in 2015

Meaningful use components

- Use of a certified EHR in a **meaningful manner**, such as e-prescribing
- Use of certified EHR technology for **electronic exchange of health information** to improve quality of health care
- Use of certified EHR technology to submit **clinical quality** and other measures

[https://www.cms.gov/EHRIncentivePrograms/30_Meaningful_Use.asp](https://www.cms.gov/EHRIncentivePrograms/30_Meaningful_Use.asp)
MU Stage 1 and Clinical Ontologies

- Problems: ICD-9-CM or SNOMED CT
- Procedures: ICD-9-CM (volume 3), Health Care Financing Administration Common Procedure Coding System (HCPCS), CPT-4
- Laboratory test results: LOINC
- Medications: RxNorm, or any source vocabulary that is included in RxNorm
- Immunizations: CVX
- Race and Ethnicity: OMB Directive No. 15
- HL7 Continuity of Care Document (CCD) & Messages
Meaningful Use stages

More sophisticated clinical systems, requiring an ever increasing variety (and amount) of structured and coded data

https://www.cms.gov/EHRIncentivePrograms
Patient-centered medical homes

• “… highly integrated, team-based practices that promote patient centered care through routine patient feedback and better access … also promote improved clinical quality and efficiency through increased care coordination.”

• Critical improvements in EHRs:
  – Clinical decision support, registries, team care, care transitions, personal health records, telehealth technologies, and measurement

• Information exchange with integration of inpatient and outpatient EHRs

• More data on the aggregate and individual patient/provider level

By 2020, ninety percent of clinical decisions will be supported by accurate, timely, and up-to-date clinical information, and will reflect the best available evidence and informed personal preference.
Conclusions

Challenges & Opportunities
Opportunities

• Government providing exceptional incentives for Healthcare IT adoption
  – IT identified as a key enabler of a more effective healthcare system

• Proposed healthcare delivery models require high levels of integration within and across institutions
  – Moving towards seamless collaboration where patients are active contributors

• Opportunity for a new generation of clinical systems beyond efficient record storage and communication
  – New paradigm with pervasive computerized data analysis and decision support
  – Widespread use of interoperable services and data, with advanced functions that enable team-based care
Challenges

• Cost-effective **semantic interoperability**
  – Existing standards make data exchange possible, but not simple or efficient (projects take months or years)
  – Data exchanged in a structured and coded format still represents a small portion of the electronic record

• Clinical systems that can seamlessly represent and process a **complete electronic patient care record**
  – Current systems frequently rely on legacy data architectures that limit the use of clinical ontologies
  – Slow adoption of technologies that can help overcome the current data representation limitations

• Clinical ontologies with proper **domain coverage** and **extensibility**
  – Existing methods and tools to use clinical ontologies are not accessible to typical clinicians
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