An Ontology Ecosystem Approach to Electronic Health Record Interoperability

Barry Smith
Ontology Summit
April 7, 2016
Electronic Health Records – pro

• no more redundant tests
• continuity of care improved
• quality of information improved
  – no more lost charts
  – no more illegible handwriting
  – no more non-standard codes
• charts are accessible from multiple sites simultaneously
Electronic Health Records – con

1. to reep these benefits we have to get the data inside the computer
2. in a form that allows it to be shared

Addressing 1. brings costs/risks in areas such as privacy, safety, clinician distraction,
Addressing 2. requires EHR system interoperability
Interoperability

Definition: Two systems A and B are interoperable if the system A-data can be used by system B in the same way that it is used by system A and vice versa.

- EHR systems in the US (at least) are still (2016) a long way from interoperability.
• Perhaps Epic (Prop: Judy Faulkner) will solve the problem
• slowly, but surely, **everyone** will use Epic
start with the US
tomorrow, the galaxy
even total victory of Epic would not imply interoperability

• RAND Corporation: Epic is a “closed system” that makes it “challenging and costly” for hospitals to interconnect. (New York Times, September 13, 2014)
The Staggering Cost Of An Epic Electronic Health Record Might Not Be Worth It

Duke University Health System will shell out $700 million, so will Boston-based Partners HealthCare;
Why Health Care Tech Is Still So Bad

By ROBERT M. WACHTER  MARCH 21, 2015

… I interviewed Boeing’s top cockpit designers, who wouldn’t dream of green-lighting a new plane until they had spent thousands of hours watching pilots in simulators and on test flights. This principle of user-centered design is part of aviation’s DNA, yet has been woefully lacking in health care software design.
Why is the US stuck with EHR systems which are so clunky and distracting, and which address hardly at all the issue of interoperability?

Why was it all done so quickly, when there were so few talented, trained personnel, with the needed sorts of expertise?

Answer: the **HITECH Act (2009)**: let’s bribe physicians to adopt these Mumps-based EHR systems quickly, and then penalize them if they fail to do so.
Compare: the COMPUTECH Act (1959)
Let’s bribe computer users to use only this new-fangled COBOL language in all the work they do … and then penalize them with bigger and bigger fines each year until they all do so, forever and ever.
To get paid under the Hitech act, you must show “Meaningful Use”

<table>
<thead>
<tr>
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<tbody>
<tr>
<td>1. Capturing health information in a coded format</td>
<td>1. Disease management, clinical decision support</td>
<td>1. Achieving improvements in quality, safety and efficiency</td>
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<tr>
<td>2. Using the information to track key clinical conditions</td>
<td>2. Medication management</td>
<td>2. Focusing on decision support for national high priority conditions</td>
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<td>3. Communicating captured information for care coordination purposes</td>
<td>3. Support for patient access to their health information</td>
<td>3. Patient access to self-management tools</td>
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<td>5. Reporting of clinical quality measures and public health information</td>
<td>5. Quality measurement</td>
<td>5. Improving population health outcomes</td>
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<td>6. Reporting of clinical quality measures and public health information</td>
<td>6. Research</td>
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<td>7. Reporting of clinical quality measures and public health information</td>
<td>7. Bi-directional communication with public health agencies</td>
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Data capture and sharing

Advance clinical processes

Leverage information to improve outcomes
# Meaningful Use and Interoperability

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<tr>
<td>1. Capturing health information in a coded format</td>
<td><strong>Problem lists must be “stored” using codes from SNOMED-CT</strong>*</td>
<td>1. Achieving improvements in quality, safety and efficiency</td>
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<td>2. Using the information to track key clinical conditions</td>
<td><em>Systematized Nomenclature of Medicine – Clinical Terms</em></td>
<td>2. Focusing on decision support for national high priority conditions</td>
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<td>3. Communicating captured information for care coordination purposes</td>
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<td>3. Patient access to self-management tools</td>
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<td>4. Reporting of clinical quality measures and public health information</td>
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<td>4. Access to comprehensive patient data</td>
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<td>5. Improving population health outcomes</td>
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<tr>
<td><strong><a href="https://www.healthit.gov/sites/default/files/meaningfulusetablesseries2_110112.pdf">https://www.healthit.gov/sites/default/files/meaningfulusetablesseries2_110112.pdf</a></strong></td>
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*SNOMED-CT: Systematized Nomenclature of Medicine – Clinical Terms*
Coding with SNOMED-CT still not standard practice among physicians in US

Human coding with SNOMED-CT is unreliable and inconsistent

The organization of SNOMED-CT allows many alternative ways of coding what is medically the same phenomenon
An example of a problem list

• No Allergies
• No Known Allergies

Two problems?
Or one?
Or zero?
What is a problem?

SNOMED: **Solitary leiomyoma (disorder)**
Concept ID: 254769006

SNOMED: **Leiomyoma (morphologic abnormality)**
Concept ID: 44598004

Two problems or one?

http://www.snomedbrowser.com/Codes/Details/254769006
http://www.snomedbrowser.com/Codes/Details/44598004
Introduction to HL7 Standards

HL7 and its members provide a framework (and related standards) for the exchange, integration, sharing, and retrieval of electronic health information. These standards define
What is a problem?

HL7 Glossary

Problem = Def. a clinical statement that a clinician chooses to add to a problem list.

‘Clinical statement’ is not defined
Ontological incoherence

- People don’t know what ‘problem’ means
- Machines will not know what ‘problem’ means either
- And so they will fail to auto-generate SNOMED-conformant problem lists from EHRs in a way that promotes interoperability
Perhaps

FHIR: Fast Healthcare Interoperability Resources can help
What is FHIR?

- **Fast Healthcare Interoperability Resources**
- Pronounced “FIRE”
- Essentially HL7 v4
  - (won’t be marketed that way)
  - New artifacts
  - New methodology
  - New tools
  - New publishing approach
  - Still built on RIM, vocab & datatypes, but more hidden
FHIR: Condition

=Def. Use to record detailed information about conditions, problems or diagnoses recognized by a clinician. There are many uses including: recording a diagnosis during an encounter; populating a problem list or a summary statement, such as a discharge summary.
Something like FHIR will be needed

... come the day when every patient’s genome is sequenced as they walk through the hospital door ...
How ensure that we will have in digital form the needed clinical information onto which this sequence information can smoothly and securely dock?
Personalized medicine needs large cohorts with rich phenotypic data conforming to common standards. How to get there?

1. Everyone uses Epic
2. Government enforces common standards
3. Let’s start again from scratch, using the same approach we should have used from the beginning: rigorous testing-based development of EHRs by leading medical research institutions until we see what technologies will work
Perspective

Published online: 7 November 2007 | doi:10.1038/nbt1346

The OBO Foundry: coordinated evolution of ontologies to support biomedical data integration

Barry Smith¹, Michael Ashburner², Cornelius Rosse³, Jonathan Bard⁴, William Bug⁵, Werner Ceusters⁶, Louis J Goldberg⁷, Karen Eilbeck⁸, Amelia Ireland⁹, Christopher J Mungall¹⁰, The OBI Consortium¹¹, Neocles Leontis¹², Philippe Rocca-Serra⁹, Alan Ruttenberg¹³, Susanna-Assunta Sansone⁹, Richard H Smith¹⁴, and Xi Zhou¹⁵. See references 16 -
<table>
<thead>
<tr>
<th>Granularity</th>
<th>Continuant</th>
<th>Occurrent</th>
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<tbody>
<tr>
<td>Complex of Organisms</td>
<td>Family, Community, Deme, Population</td>
<td>Population Environment (EnvO)</td>
</tr>
<tr>
<td>Organ and Organism</td>
<td>Organism (NCBI Taxonomy)</td>
<td>Anatomical Entity (FMA, CARO)</td>
</tr>
<tr>
<td>Cell and Cellular Component</td>
<td>Cell (CL)</td>
<td>Cellular Component (FMA, GO)</td>
</tr>
<tr>
<td>Molecule</td>
<td>Molecule (CHEBI, SO, RNAO, PRO)</td>
<td>Molecular Function (GO)</td>
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Building Ontologies with Basic Formal Ontology

By Robert Arp, Barry Smith and Andrew D. Spear

Overview

In the era of “big data,” science is increasingly information driven, and the potential for computers to store, manage, and integrate massive amounts of data has given rise to such new disciplinary fields as biomedical informatics. Applied ontology offers a strategy for the organization of scientific information in computer-tractable form, drawing on concepts not only from computer and information science but also from linguistics, logic, and philosophy. This book provides an introduction to the field of applied ontology that is of particular relevance to biomedicine, covering theoretical components of ontologies, best practices for ontology design, and examples of biomedical ontologies in use.

After defining an ontology as a representation of the types of entities in a given domain, the book distinguishes between different kinds of ontologies and taxonomies, and shows how applied ontology builds on the traditional kinds of an ontology. It concludes with an overview of the Basic Formal Ontology (BFO)
BFO-based hub and spokes strategy for developing interoperable ontology modules

<table>
<thead>
<tr>
<th>Basic Formal Ontology (BFO)</th>
<th>Information Artifact Ontology (IAO)</th>
<th>Ontology for Biomedical Investigations (OBI)</th>
<th>Spatial Ontology (BSPO)</th>
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<tbody>
<tr>
<td>Anatomy Ontology (FMA*, CARO)</td>
<td>Environment Ontology (EnvO)</td>
<td>Infectious Disease Ontology (IDO*)</td>
<td>Biological Process Ontology (GO*)</td>
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<tr>
<td>Cell Ontology (CL)</td>
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<td>Subcellular Anatomy Ontology (SAO)</td>
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<td>Sequence Ontology (SO*)</td>
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<td>Protein Ontology (PRO*)</td>
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examples of the BFO/OBO Foundry ontology ecosystem approach extended to other domains

<table>
<thead>
<tr>
<th>NIF Standard</th>
<th>Neuroscience Information Framework</th>
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<tbody>
<tr>
<td>eagle-I / VIVO</td>
<td>Integrated Semantic Framework / CTSA Connect</td>
</tr>
<tr>
<td>Core</td>
<td></td>
</tr>
<tr>
<td>IDO Core / IDO</td>
<td>Infectious Disease Ontology Suite</td>
</tr>
<tr>
<td>extensions</td>
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<tr>
<td>cROP / Planteome</td>
<td>Common Reference Ontologies for Plants</td>
</tr>
</tbody>
</table>
Examples of BFO/OBO Foundry approach extended into yet further domains

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<thead>
<tr>
<th>UNEP Ontology Framework</th>
<th>United Nations Environment Programme</th>
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<tr>
<td>USGS National Map Ontologies</td>
<td>United States Geological Survey</td>
</tr>
<tr>
<td>Joint Doctrine Ontologies</td>
<td>US Air Force Research Labs / Training and Doctrine Command (TRADOC)</td>
</tr>
<tr>
<td>TRIP Ontologies</td>
<td>Federal Highway Administration (FHWA) Transportation Research Informatics Platform (TRIP)</td>
</tr>
</tbody>
</table>
165+ ontologies re-using BFO

from: [http://ifomis.uni-saarland.de/bfo/users](http://ifomis.uni-saarland.de/bfo/users)

- ACGT Master Ontology (ACGT MO)
- Alzheimer Disease Ontology (ADO)
- Adverse Event Ontology (AEO)
- Adverse Event Reporting Ontology (AERO)
- AFO Foundational Ontology
- Actionable Intelligence Retrieval System (AIRS)
- Bank Ontology
- Beta Cell Genomics Application Ontology (BCGO)
- BioAssay Ontology
- Bioinformatics Web Service Ontology
- Biological Collections Ontology (BCO)
- Biomedical Ethics Ontology
- Biomedical Grid Terminology (BiomedGT, retired)
- BioTop: A Biomedical Top-Domain Ontology
- BIRNLex: controlled terminology for annotation of BIRN data sources
- Blood Ontology (BLO)
- Body Fluids Ontology (BFLO)
- Bone Dysplasia Ontology
- Cancer Cell Ontology (OncoCL)
- Cancer Chemoprevention Ontology (CanCo)
- Cardiovascular Disease Ontology (CVDO)
- Cell Behavior Ontology (CBO)
- Cell Cycle Ontology
- Cell Expression, Localization, Development and Anatomy Ontology (CELDA)
- Cell Line Ontology (CLO)
- Cell Ontology (CL)
- Chemical Analysis Ontology (CAO)
- Chemical Entities of Biological Interest (ChEBI)
- Sirtuin Study Framework Ontology (SFFO)
- Environment Ontology (ENVO)
- Epidemiology Ontology (EO)
- Epilepsy and Seizure Ontology (EPSO)
- Evolution Ontology (EO)
- Experimental Factor Ontology (EFO)
- Experimental ACTioins Biomedical Protocol Ontology (EXACT2)
- Exposé: An Ontology for Data Mining Experiments
- Flybase Drosophila Anatomy Ontology (FBbt)
- Fission Yeast Phenotype Ontology (FYPO)
- Flower-Visiting Domain Ontology (FV),
- Flower-Visiting Behavior Application Ontology (FVB)
- Foundational Model of Anatomy (FMA) Ontology
- Gastrointestinal Endoscopy Ontology (GIEO)
- Gene Regulation Ontology (GRO)
- General Information Model (GIM)
- Genomic Feature and Variation Ontology (GFVO)
- Gestalt: Federated Access to Cyber Observables for Detection of Targeted Attack
- Health Data Ontology Trunk (HDOT)
- Human Interaction Network Ontology (HINO)
- Human Physiology Simulation Ontology (HuPSON)
- Infectious Disease Ontology (IDO)
- Information Artifact Ontology (IAO)
- Informed Consent Ontology (ICO)
- Interaction Network Ontology (INO)
- Interdisciplinary Prostate Ontology Project (IPOP)
- Intracranial aneurysm (ICA) Ontology
- Knowledge Base Of Biomedicine (KaBOB)
- Known Flower-Visiting Group Domain Ontology (KFG)
- Lactic Acid
## IDO Core and IDO Extensions

<table>
<thead>
<tr>
<th>IDO</th>
<th>Description</th>
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<tbody>
<tr>
<td>IDO-BRU</td>
<td>Brucellosis Ontology</td>
</tr>
<tr>
<td>IDO-HIV</td>
<td>HIV Ontology</td>
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<tr>
<td>IDO-FLU</td>
<td>Influenza Ontology</td>
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<tr>
<td>IDO-DENGUE</td>
<td>Dengue Ontology</td>
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<tr>
<td>IDO-STAPH</td>
<td>Staph. Aureus Ontology</td>
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<tr>
<td>IDO-PLANT</td>
<td>Plant Infectious Disease Ontology</td>
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<tr>
<td>IDO-MRSA</td>
<td>Methicillin-Resistant Staph. Aureus Ontology</td>
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<tr>
<td>IDO-VECTOR</td>
<td>Vector-Borne Infectious Disease Ontology</td>
</tr>
<tr>
<td>IDO-MAL</td>
<td>Malaria Ontology</td>
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How IDO evolves

CORE and SPOKES: Domain ontologies

SEMI-LATTICE: By subject matter experts in different communities of interest.
How IDO STAPH evolves

- IDOMAL
- IDOFL
- IDU
- IDO STAPH
- IDOMR Sa
- IDOMRSa
- IDORatSa
- IDORatStrep
- IDOSStrep
- IDOAntibioticResistant
- IDOHumanSa
- IDOHumanStrep
- IDOHumanBacterial
- IDOHumanSa
- IDOHumanStrep
- IDOAntibioticResistant
Clinical Terminology Shock and Awe (CTSA)

• Fifth Annual Workshop of the Clinical and Translational Science Ontology Group
• **Date:** September 7-8, 2016
• **Venue:** Ramada Hotel, Amherst, NY
• **Goals:** To explore uses of common ontologies to support sharing and discovery of clinical data