Module 4: Ontologies

Topics:

BioMedical Ontologies (knowledge graphs)

Ontology Tools

Learning Objectives:

Describe what an ontology is, how and why they are used in biomedicine.

Explain how ontologies help achieve semantic integration.

Know how to use an ontology look-up service.

Ontology Overview

- Ontology Introduction
- Ontology Background
  1. BioMed Domain – Health care and Life Science
  2. Reference and Application
  3. Ontology Granularity and Layout
- Examples
  1. Reference Ontology Examples
     1. BioPAX – Mid level – biological pathways
     2. Gene Ontology ("GO") – Gene annotation
  2. Application Ontology Examples
     1. Influenza Ontology
     2. Translational Medicine Ontology
- Best Practices
  - Development Process: Start with Use Case, develop prototype, Evaluation
  - Standards: BioMedical Ontology Best practices (BioPortal, BFO, SIO)

Introduction to Biomedical Ontologies #1: What is an Ontology?

https://www.youtube.com/watch?v=3EUaurjK7u8

Ontologies – knowledge graphs

Semantic Web Technology

Ontologies are how we structure our metadata and data to bring meaning into our data

- Define entities explicitly
- Define meaningful relationships
- Make queries and assertions about relationships
- When used with a reasoner, make inferences

Ontology editing tools

- Use the Web Ontology Language (OWL) to **define the ontology** (taxonomy) to organize a set of data types
- Use Resource Description Framework (RDF) to **apply ontology rules** to data sets
- Use SPARQL protocol and RDF Query Language to **make queries** against the dataset.

https://www.youtube.com/watch?v=k7kbkDMOQFs

Health Care and Life Science (HCLS) Domain

The Open Biological and Biomedical Ontologies

A suite of orthogonal interoperable reference ontologies

http://www.obofoundry.org

Barry Smith
NCOR, U Buffalo NY

Table 1

Coverage of initial Foundry ontologies

Granularity	Continuant: Independent	Continuant: Independent	Continuant: Dependent		Occurrent
Organ and organism	Organism (NCBI taxonomy or similar)	Anatomical entity (FMA, CARO)	Organ function (Physiology ontology, to be determined)	Phenotypic quality (PATO)	Organism-level process (GO)
Cell and cellular component	Cell (CL, FMA)	Cellular component (FMA,GO)	Cellular function (GO)		Cellular process (GO)
Molecule	Molecule (ChEBI, SO, RnaO, PRO)		Molecular function (GO)		Molecular process (GO)

Down the left column is the granularities (spatial scales) of the entities represented in the ontologies; along the top is a d[illegible]ion corresponding to the ways these entities exist in time'Continuants' endure through time. 'Occurrents (p[illegible]nfold through time in successive stages. Continuants are divided into physical things, on the one

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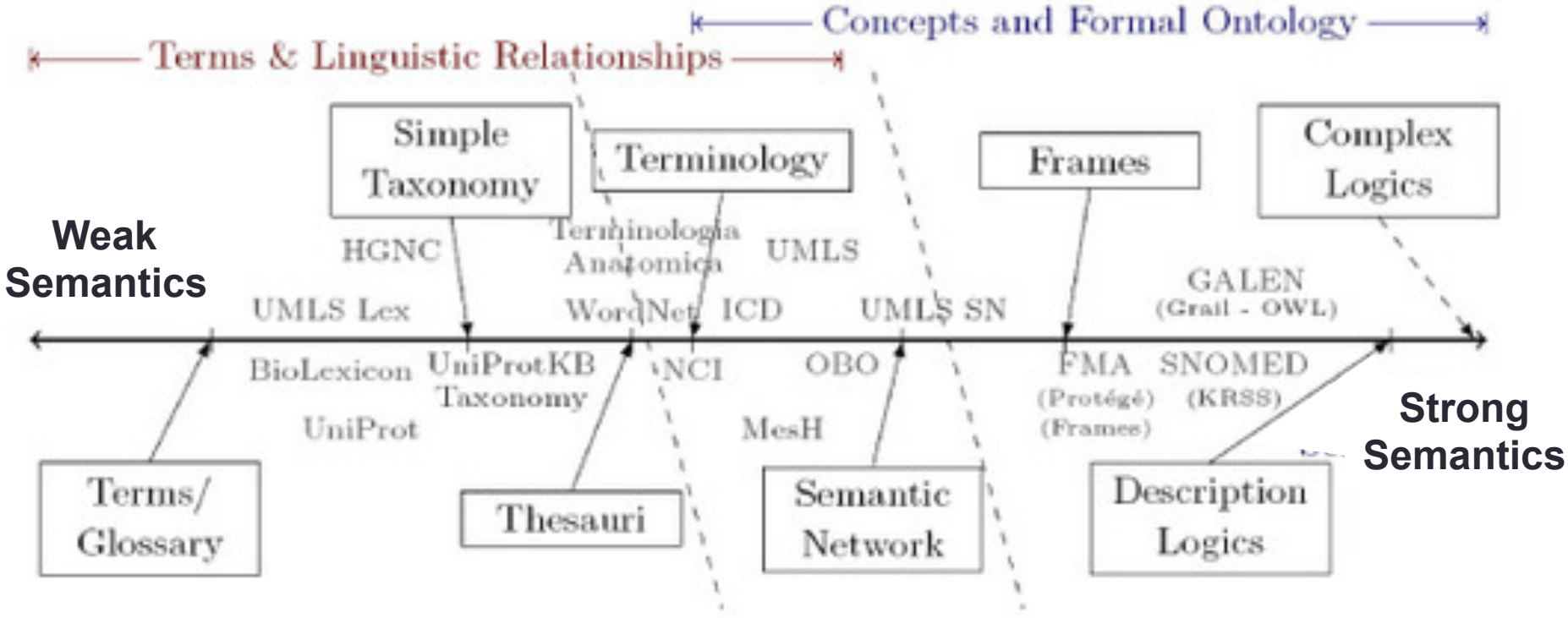
From: Nat Biotechnol. 2007 November; 25(11): 1251.
doi: 10.1038/nbt1346

http://ontology.buffalo.edu/smith/

Ontology Spectrum

Existing formalisms



engineering in Life Sciences

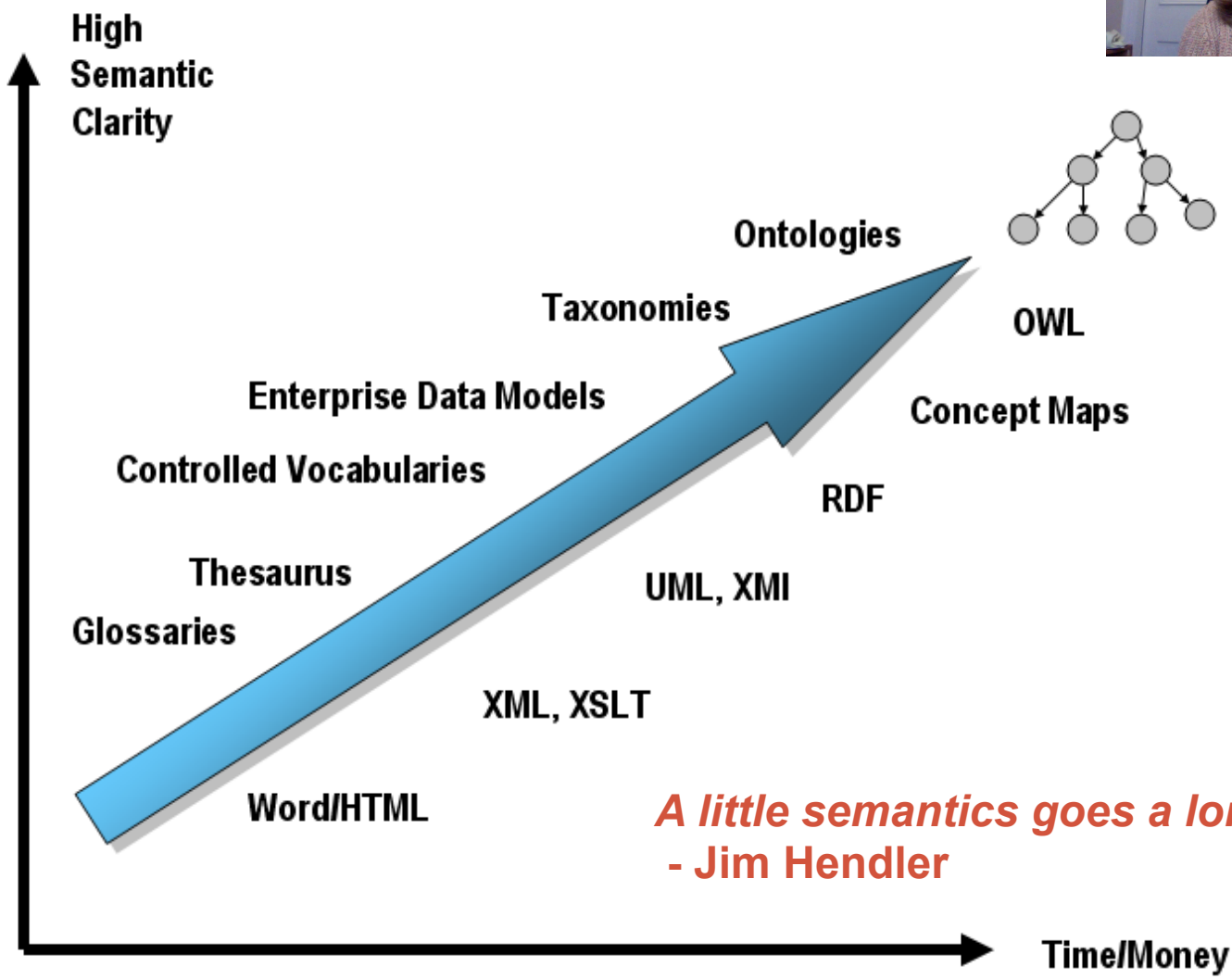
by Jimeno-Yepes, Antonio; Jiménez-Ruiz, Ernesto; Berlanga-Llavori, Rafael; Rebholz-Schuhmann, Dietrich

Journal: BMC Bioinformatics **Vol.** 10 **Issue** Suppl 10

DOI: 10.1186/1471-2105-10-S10-S4

http://www.mkbergman.com/wp-content/themes/ai3v2/images/2007Posts/070501d_SemanticSpectrum.png

Ontology Spectrum



A little semantics goes a long way.
- Jim Hendler

http://www.mkbergman.com/wp-content/themes/ai3v2/images/2007Posts/070501d_SemanticSpectrum.png

Application vs. Reference Ontology

Reference Ontology

- Intended as an authoritative source
- True to the limits of what is known
- Used by other applications, and, used by other ontologies

Application Ontology

- Built to support a particular application (use case)
- Reused rather than define terms
- Skeleton structure to support application
- Terms defined refine or create new concepts directly or through new classes based on inference

http://www.nlm.nih.gov/research/umls/presentations/2004-medinfo_tut.pdf