Module 3: Biomedical Ontology

Data Science for Drug Discovery, Health & Translational Medicine

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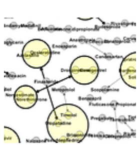
Indiana University School of Informatics and Computing



Module 3: Ontologies

Topics:

BioMedical Ontologies (also known as knowledge graphs)

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 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**
   3. **Best Practices**

Conclusion

1. **Process: Start with Use Case, develop prototype, Evaluation**
2. **Standards: BioMedical Ontology Best practices (BioPortal, BFO, SIO)**

Background

Domain: Health Care, Life Science, and People

1. Times have changed
2. Data Driven Medicine
3. Health Care Singularity

What are you building: Reference vs. Application

1. Ontology Spectrum
2. Reference vs Application Ontology

Why: Function (Use Case)

1. Link, Aggregate, Search, Integrate, etc.

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Scope: HCLS Domain

Health Care & Life Science

The Open Biological and Biomedical Ontologies

http://www.obofoundry.org

Goal: a suite of orthogonal interoperable reference ontologies

Table 1

Coverage of initial Foundry ontologies

	Continuant				Occurrent
Granularity	Independent		Dependent		
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 ion corresponding to the ways these entities exist in time 'Continuants' endure through time. 'Occurrents (p nfold through time in successive stages. Continuants are divided into physical things, on the one

Barry Smith
U Buffalo, NCBO

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

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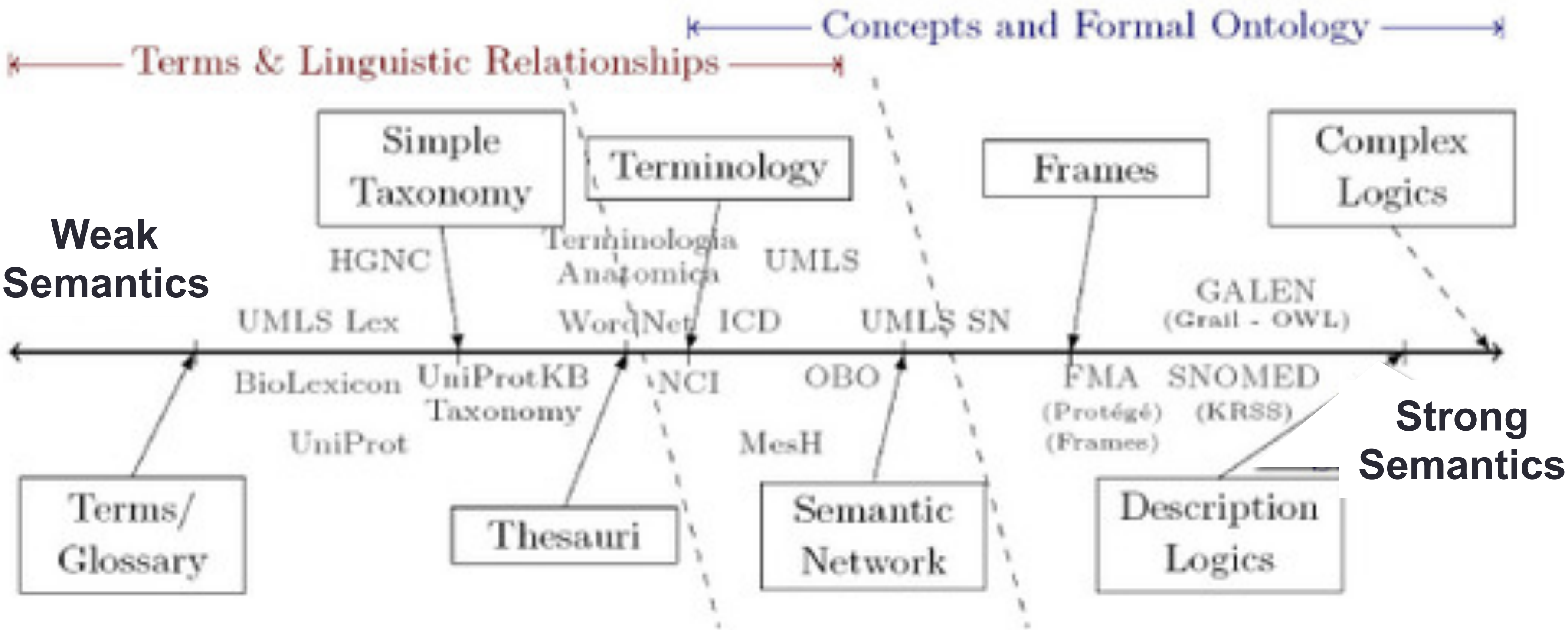
Ontology Spectrum

Existing formalisms



[**Reuse of terminological resources for efficient ontological 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

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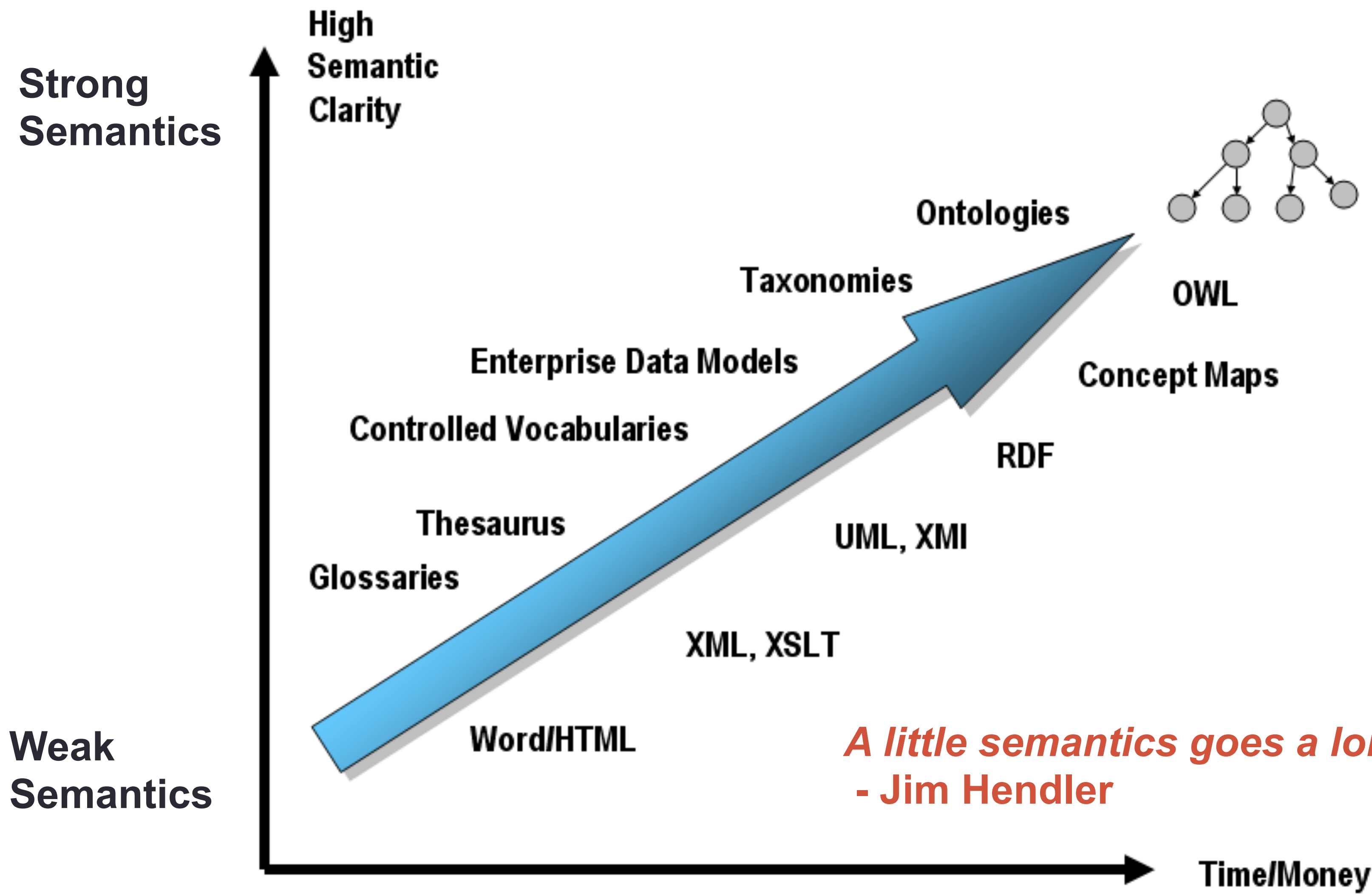
Ontology Spectrum



A little semantics goes a long way.
- Jim Hendler

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

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Application vs. Reference Ontology

Reference Ontology
- Intended as an authoritative source
- True to the limits of what is known
- Used by others

- 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

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Examples

2 Reference Ontology Examples

- BioPAX – Mid level – biological pathways
- Gene Ontology ("GO") – Gene annotation

2 Application Ontology Example

- Influenza Ontology
- Translational Medicine Ontology

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The Open Biological and Biomedical Ontologies

http://www.obofoundry.org

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

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Overview

Introduction (10 minutes)

1. Background
   1. BioMed Domain – Health care and Life Science
   2. Reference and Application
   3. Ontology Granularity and Layout
2. **Examples: (40 minutes)**
   1. Reference Ontology Examples
      1. **BioPAX – Mid level – biological pathways (10)**
      2. Gene Ontology ("GO") – Gene annotation (5)
   2. Application Ontology Examples
      1. Influenza Ontology (5)
      2. Best Practices (10)
3. Conclusion (5 minutes)
   1. Process: Start with Use Case, develop prototype, Evaluation
   2. Standards: BioMedical Ontology Best practices (BioPortal, BFO, SIO)
   3. Conferences

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Examples

2 Reference Ontology Examples

- **BioPAX – Mid level – biological pathways**
- Gene Ontology (“GO”) – Gene annotation

2 Application Ontology Example

- Influenza Ontology
- Translational Medicine Ontology

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The Open Biological and Biomedical Ontologies

http://www.obofoundry.org

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

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BioPAX

Biological PAthway eXchange

An abstract data model for biological pathway *integration*

Initiative ***arose from the community***

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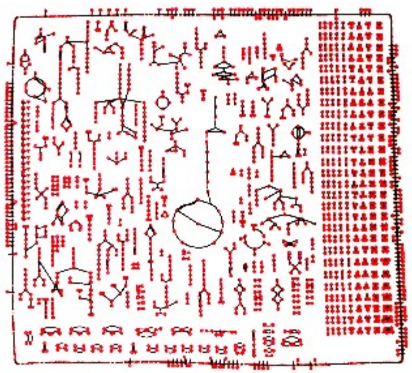
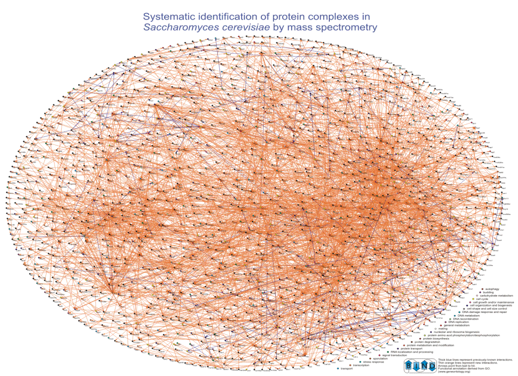
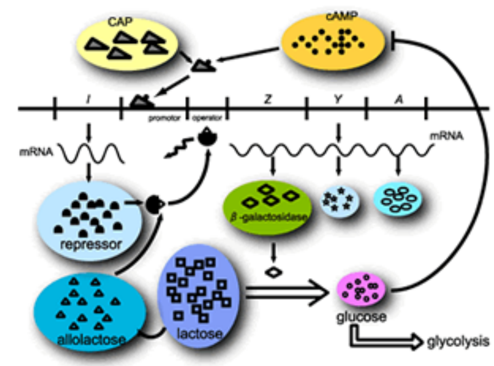
Biological Pathways of the Cell

What's a pathway?
Depends on who you ask!

BioPAX







Metabolic Pathways	**Molecular Interaction Networks**	**Signaling Pathways**	**Gene Regulation**
BioPAX Level 1	**BioPAX Level 2**	**BioPAX Level 3**	**BioPAX Level 4**

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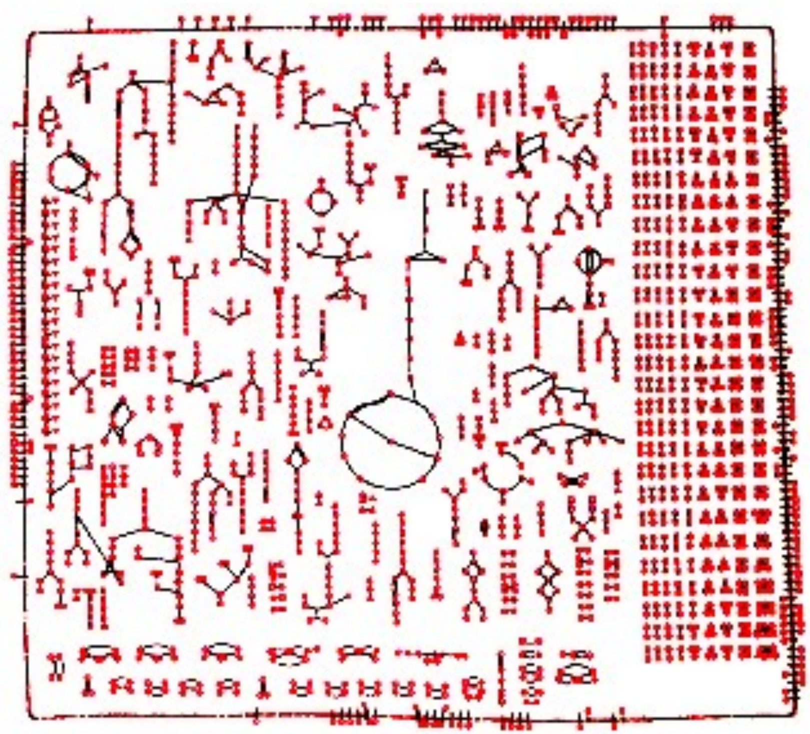
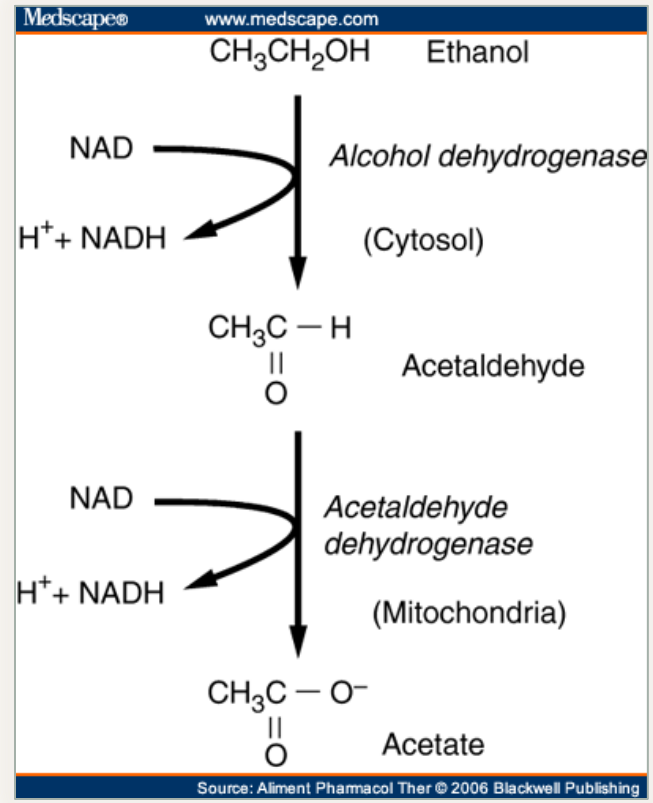
Biological Pathways of the Cell

BioPAX

BioPAX Level 1

Metabolic Pathways





A series of **chemical reactions**, **catalyzed** by **enzymes**

The **products** of one are the **reactants** of the next

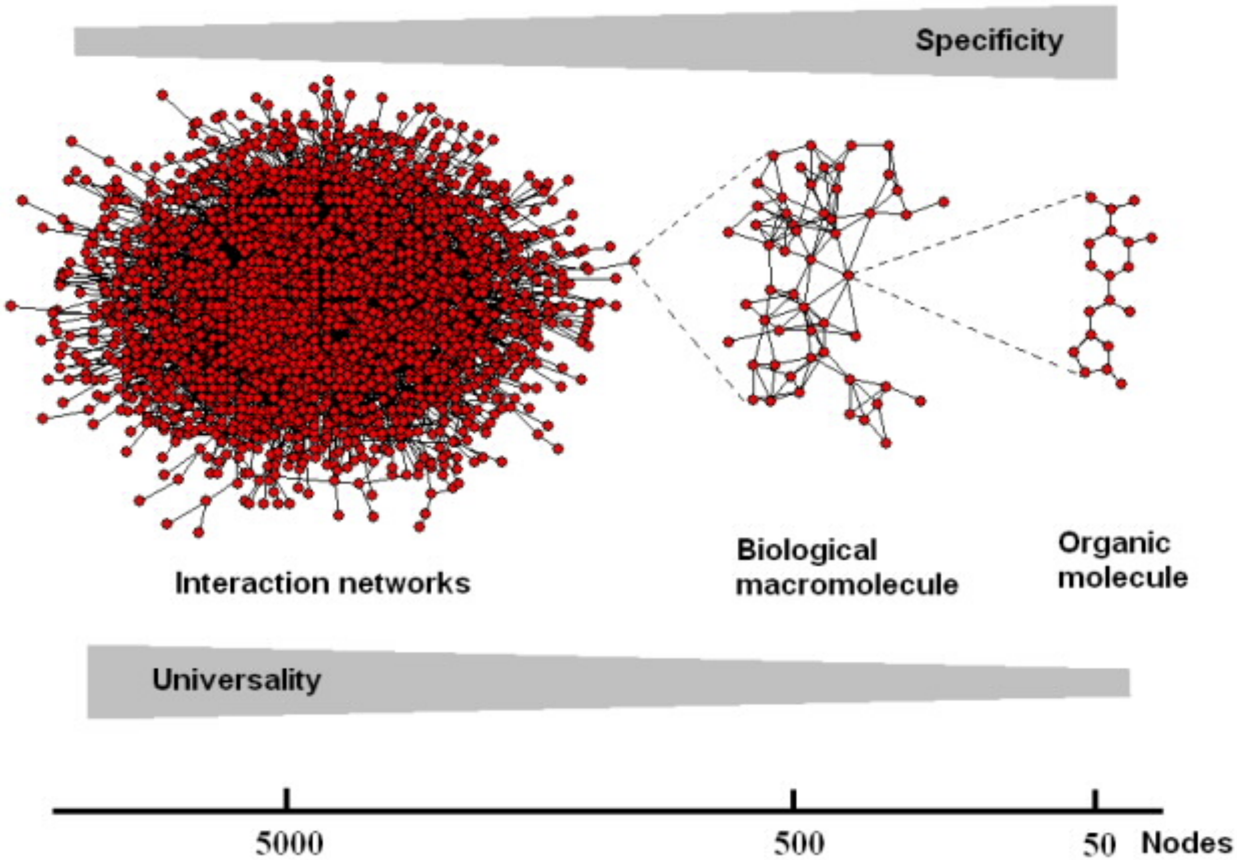
e.g. Conversion, Transport

Biological Pathways of the Cell

BioPAX

BioPAX Level 2

Molecular Interaction Networks



http://www.estradalab.org/research/

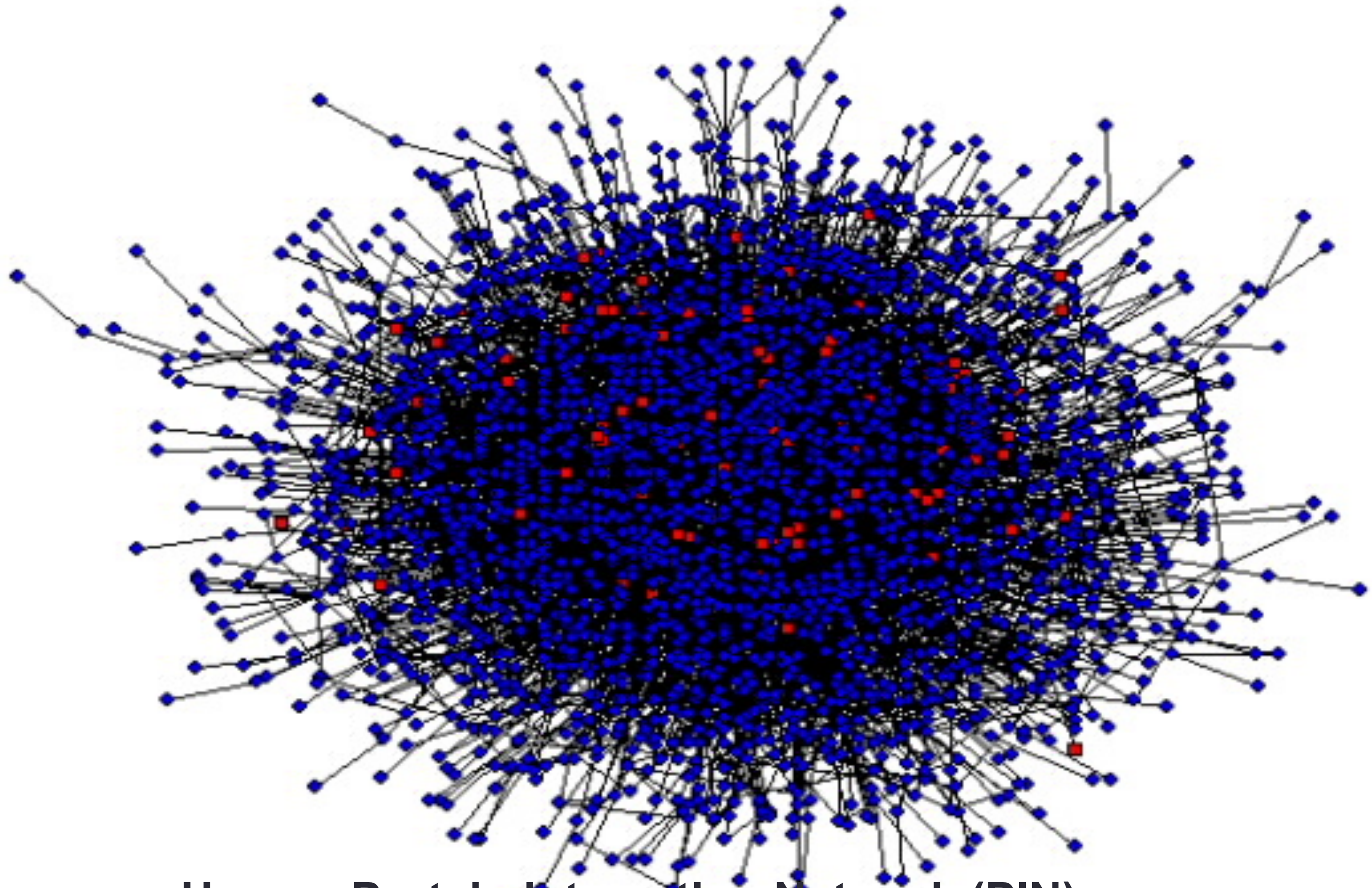
Cells are complex systems whose physiology is governed by an intricate network of Molecular Interactions (MIs) of which a relevant subset are protein–protein interactions (PPIs).

Biological Pathways of the Cell

BioPAX

Molecular Interaction Networks

BioPAX Level 2



Human Protein Interaction Network (PIN)

http://www.estradalab.org/research/

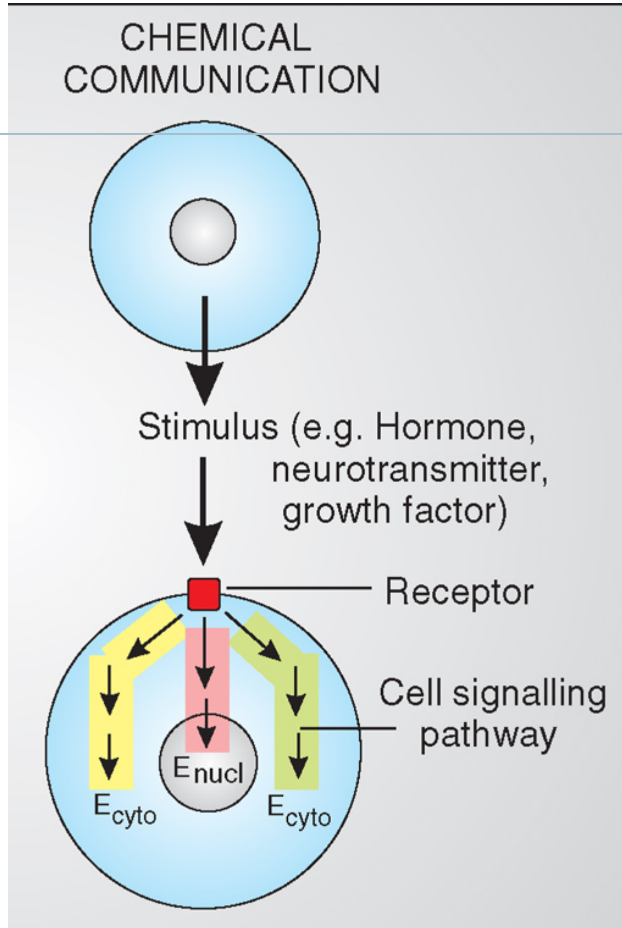
Biological Pathways of the Cell

BioPAX

BioPAX Level 3

Signaling molecules trigger cellular responses.

Molecules bind to the cell surface causing a cascade of activation Reactions

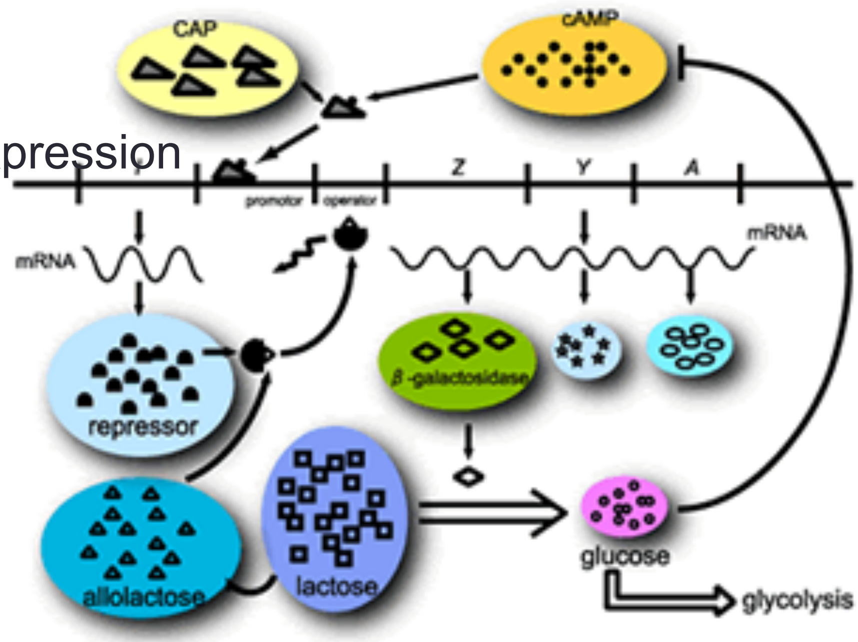


A activates B activates C….

Adapted from Cell Signalling Biology - Michael J. Berridge - www.cellsignallingbiology.org - 2012 and http://www.hartnell.edu/tutorials/biology/signaltransduction.html

Biological Pathways of the Cell

BioPAX



The modulation of any of the stages of gene expression that control:
  which genes are switched on and off
  when, how long, and how much

Gene regulation may occur many stages:
Transcription
Post-transcriptional modification
RNA transport
Translation
mRNA degradation
Post-translational modifications
    among many others (more recently discovered!)

Gene Regulation

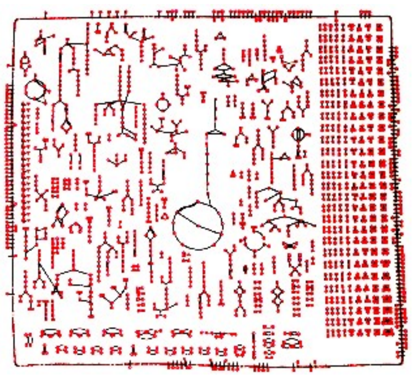
http://en.wikipedia.org/wiki/Regulation_of_gene_expression

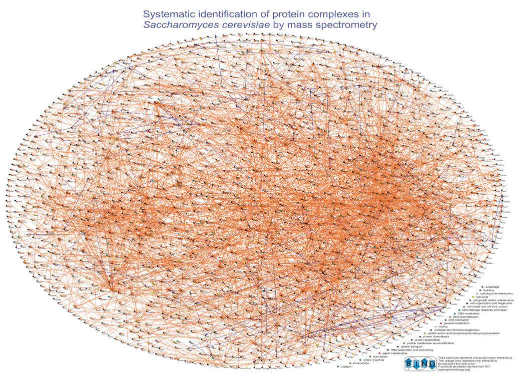
http://www.biology-online.org/dictionary/Gene_regulation

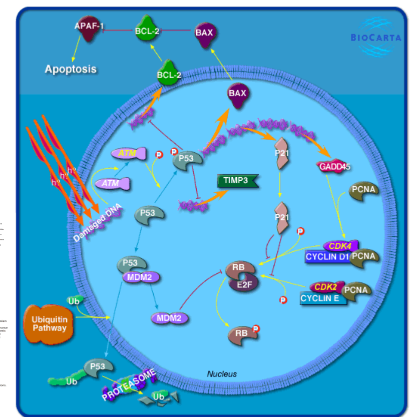
Biological Pathways of the Cell

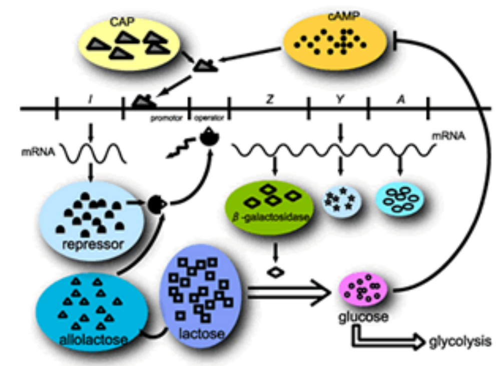
What's a pathway?
Depends on who you ask!

BioPAX









Metabolic Pathways	**Molecular Interaction Networks**	**Signaling Pathways**	**Gene Regulation**
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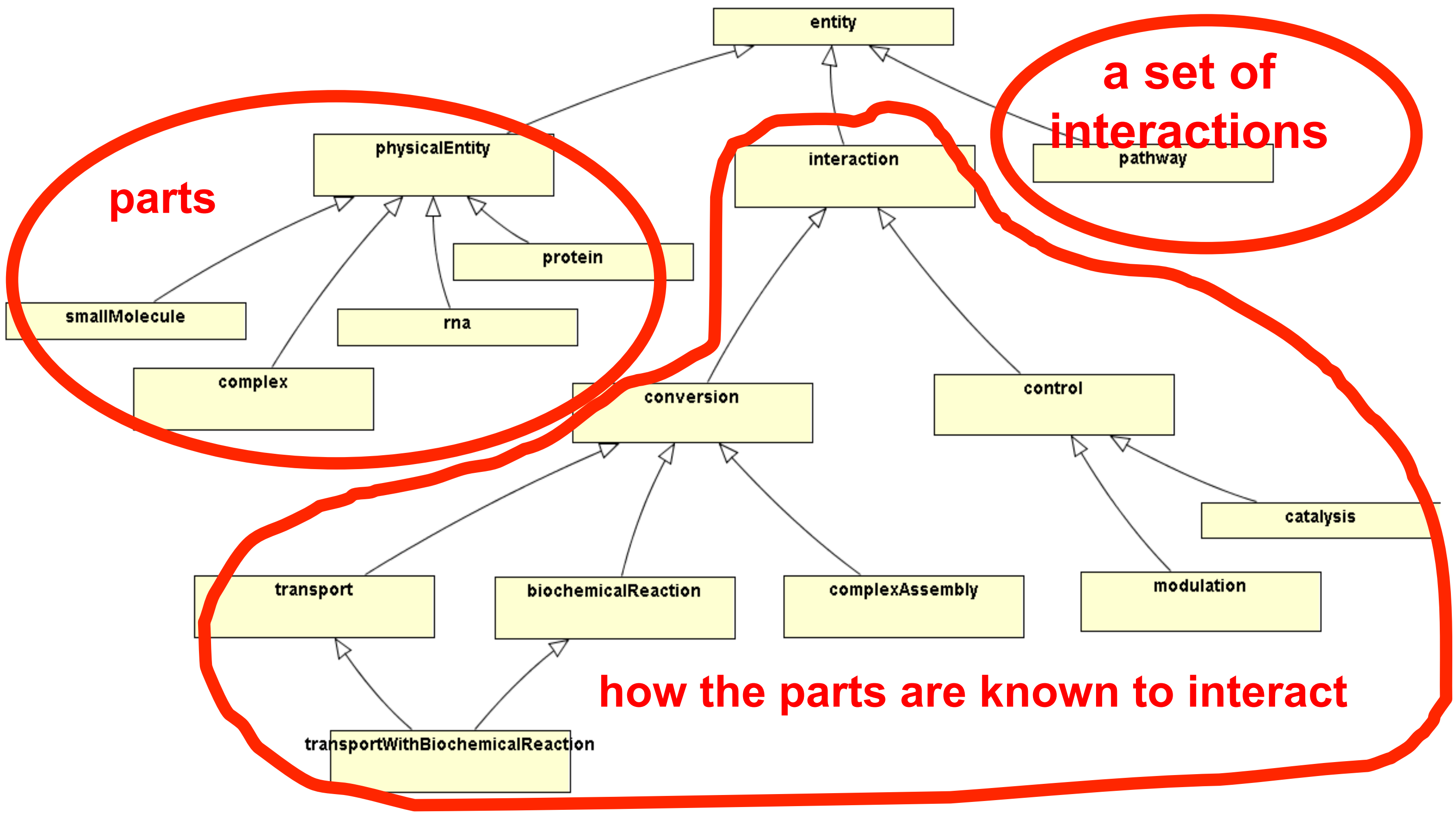
BioPAX Ontology



Level 1 v1.0 (July 7th, 2004)

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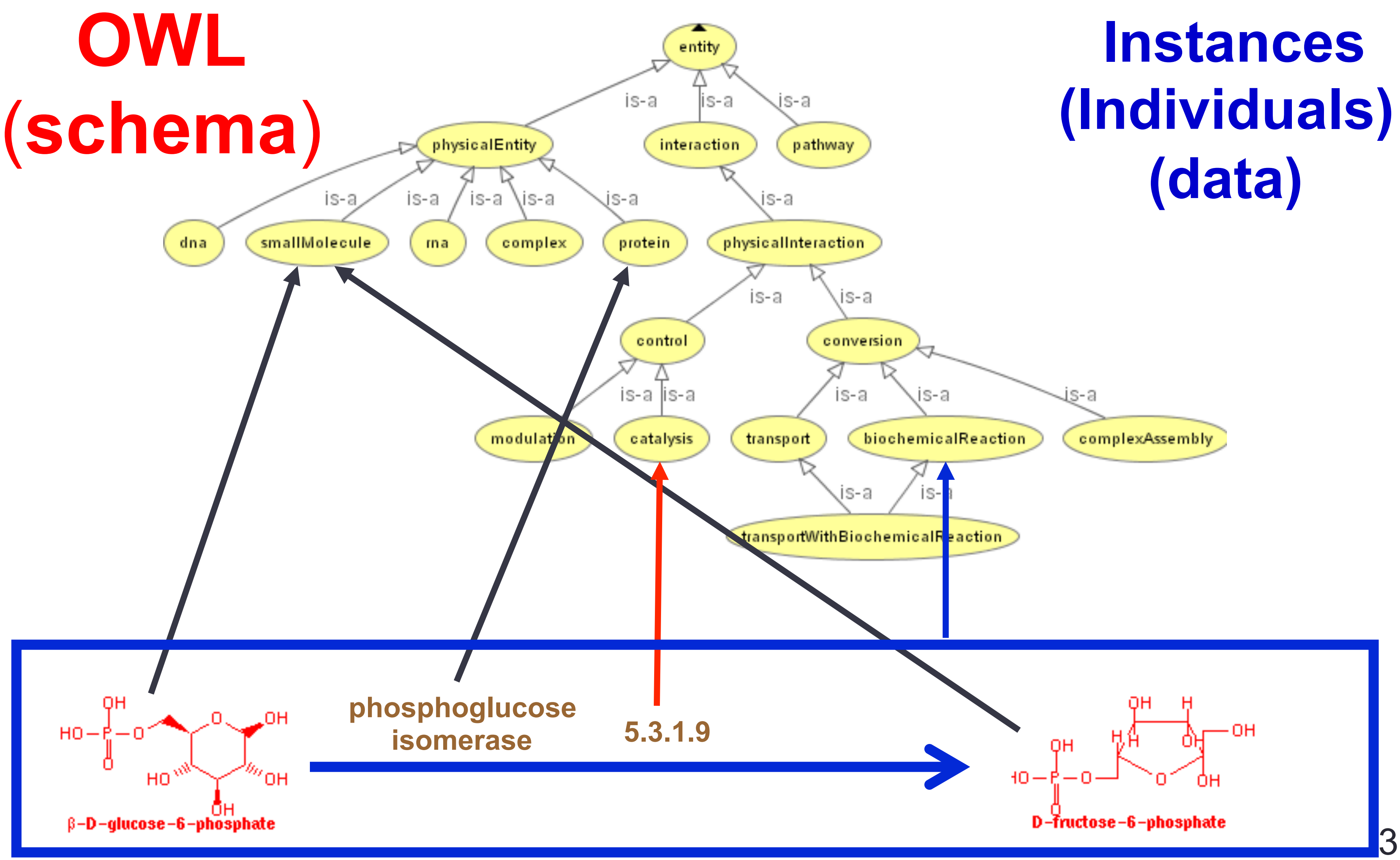
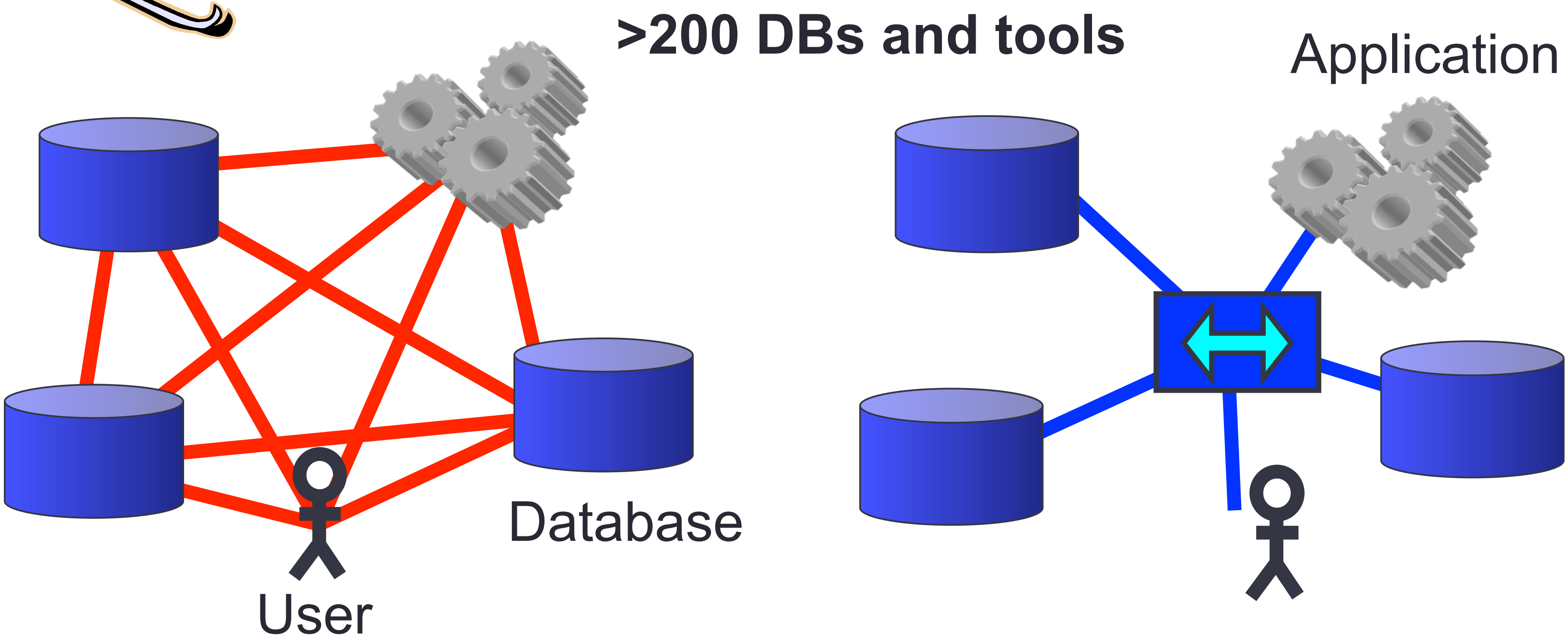
BioPAX Biochemical Reaction



BioPAX - Simplify



Before BioPAX **With BioPAX**

Common "computable semantic" enables scientific discovery

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Examples

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2 Application Ontology Example

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

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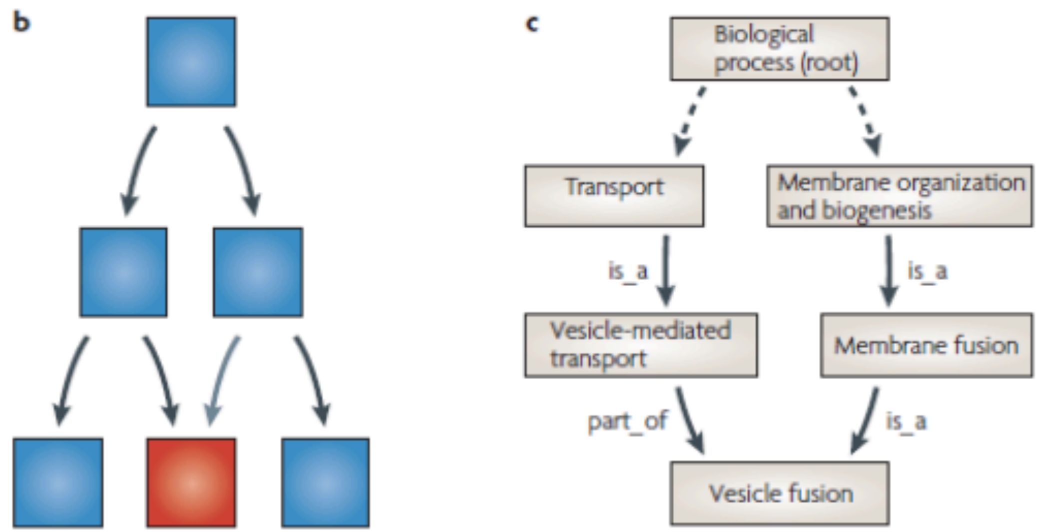
Gene Ontology (GO)

Standard representations:

- Gene and gene product attributes
- Across species and databases

Structured controlled vocabularies organized as 3 independent Ontologies

- Molecular Interactions
- Biological Processes
- Cellular Location



[1] Rhee, S.Y, Wood, V., Dolinski, K. and Draghici, S. 2008. Use and misuse of the gene ontology annotations. Nature Reviews Genetics 9:509-515.
[2] http://people.oregonstate.edu/~knausb/rna_seq/annot.pdf

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Gene Ontology

Two Key Uses:

- Resource: to look up genes with similar functionality or location within the cell to help characterize the function of a sequence or structure
- Use to annotate genomes to enable the analysis of the genome through the annotation terms.

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Gene Ontology
Evidence Codes

Manually-assigned evidence codes fall into

Four categories:

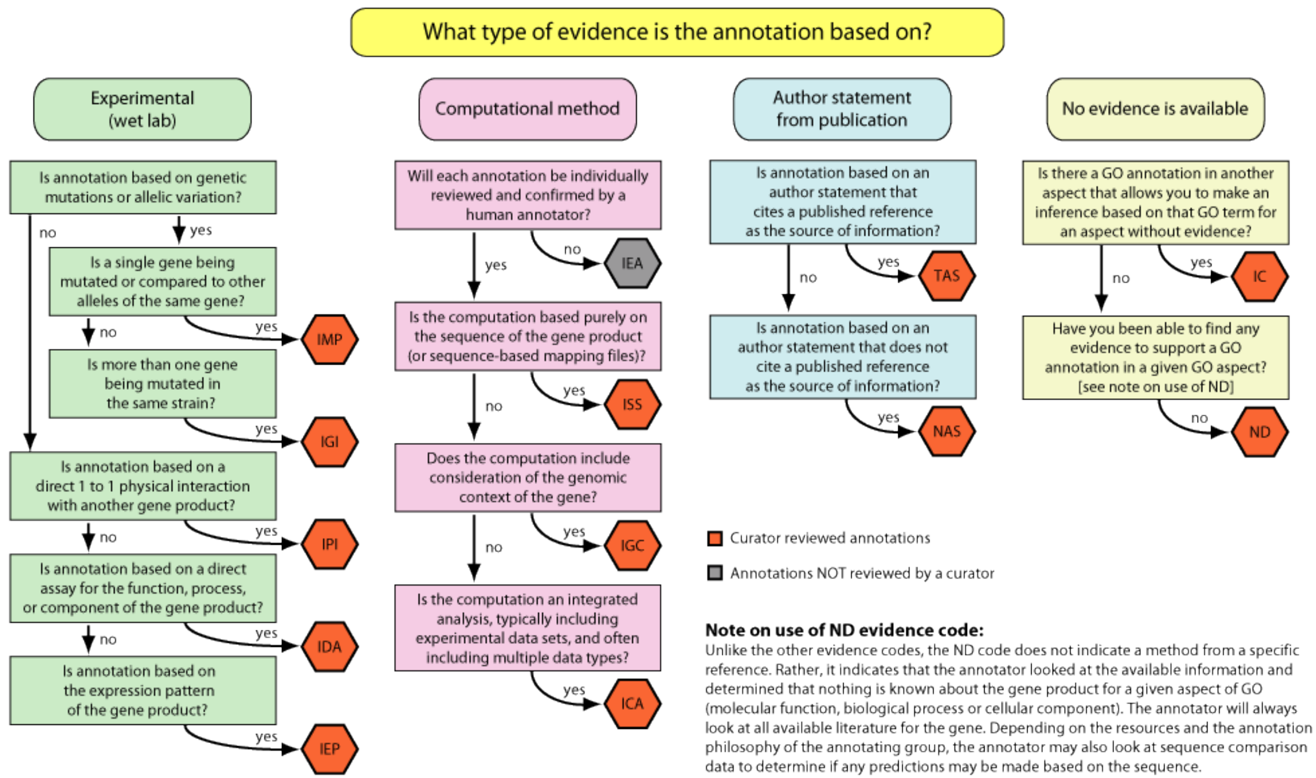
Experimental

Computational analysis

Author statements,

Curatorial statements

GO Evidence Code Decision Tree



Inferred from Electronic Annotation (IEA) is not assigned by a curator.

Adapted from: http://people.oregonstate.edu/~knausb/rna_seq/annot.pdf
Rhee, S.Y, Wood, V., Dolinski, K. and Draghici, S. 2008. Use and misuse of the gene ontology annotations. Nature Reviews Genetics 9:509-515.
See also: http://www.geneontology.org/GO.evidence.shtml

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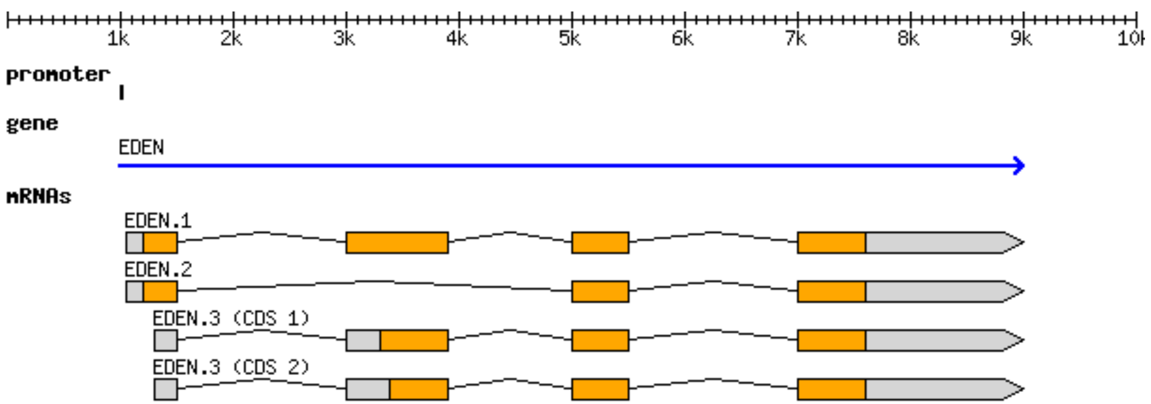
Sequence Ontology

Sequence Ontology (SO) 'terms and relationships used to describe the features and attributes of biological sequence.' (E.g., binding_site, exon, etc.)

- sequence_attribute
  - feature_attribute
  - polymer_attribute
  - sequence_location
  - variant_quality
- sequence_feature
  - junction
  - region
  - sequence_alteration
- sequence_variant
  - functional_variant
  - structural_variant
- Relationship (lots!)



(snuck this one in as another example)

SO http://www.sequenceontology.org/

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Application vs. Reference Ontology

Reference Ontology

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- Used by others

- 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

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Application Ontology
Influenza Ontology

Ontology Support for Influenza Research and Surveillance

Joanne Luciano, PhD,
Lynette Hirschman, PhD, Marc Colosimo, PhD

Approved for Public Release; Distribution Unlimited.

28 April 2008 Case Number 08-0738

MITRE

http://www-test.ebi.ac.uk/industry/Documents/workshop-materials/DiseaseOntologiesAndInformation190608/The%20Influenza%20Infectious%20Disease%20Ontology%20(I-IDO)%20-%20Joanne%20Luciano.pdf

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Application Ontology
Influenza Ontology

Case Study 2: UK

- Outbreak of H5N1 in the UK at a turkey farm Feb 1, 2007
- What is the source of the outbreak?
  - Contact with infected wild birds?
    - But turkeys were in an enclosed “biosecure” unit
    - No H5N1 detected in the region in the 2 previous months
  - Govt. veterinarian suggested turkey meat from Hungary might be source of infection
    - Turkey farm is adjacent to a poultry packing plant that had processed poultry products from Hungary
    - Hungary had reported an H5N1 outbreak 2 weeks earlier
- Sequence data showed that strain infecting the turkeys was 99.96% identical to strain that had infected Hungarian birds
- Conclusion: Infected Hungarian poultry was source of H5N1 infection
  - Open question (relevant to food defense): how did H5N1 spread from processing plant to live turkeys?

MITRE

2

http://www-test.ebi.ac.uk/industry/Documents/workshop-materials/DiseaseOntologiesAndInformation190608/The%20Influenza%20Infectious%20Disease%20Ontology%20(I-IDO)%20-%20Joanne%20Luciano.pdf

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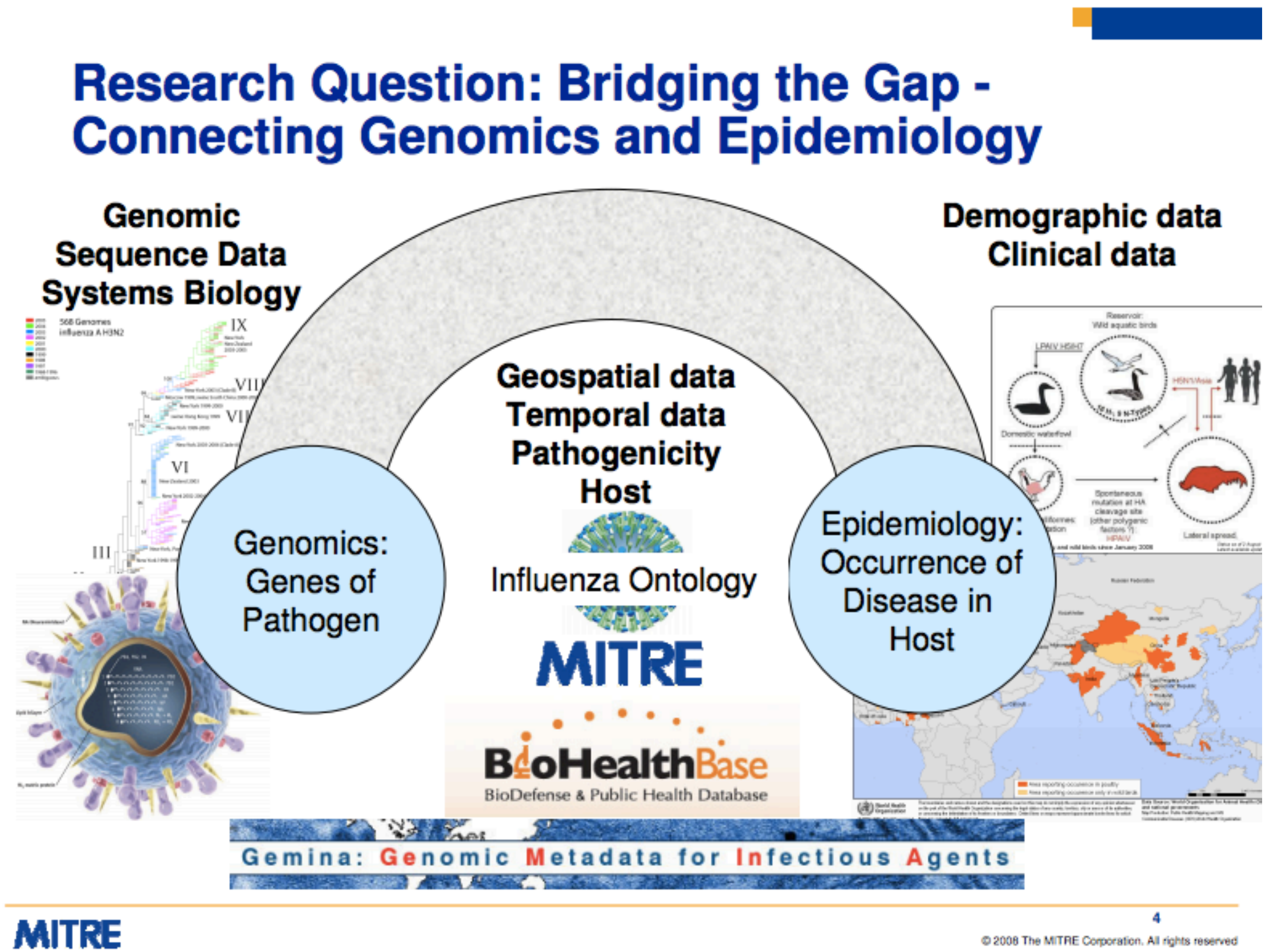
Application Ontology
Influenza Ontology



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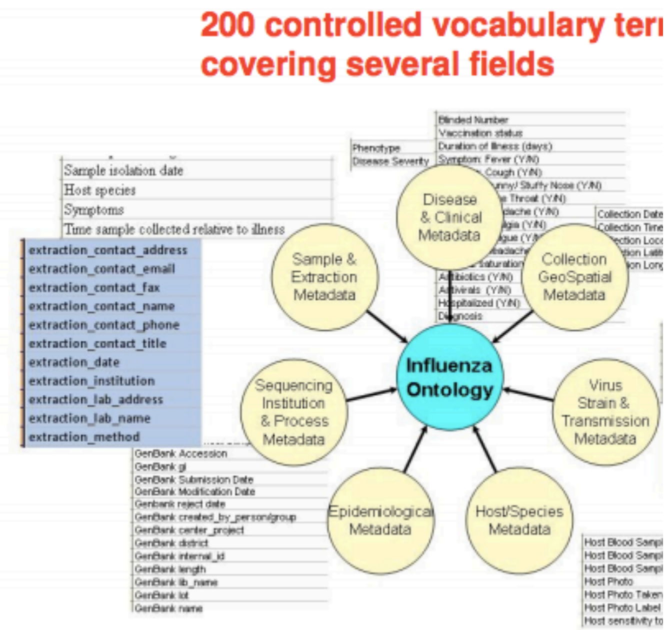

Reuse of existing ontologies & metadata standards

- OBI – Ontology of Biomedical Investigations
- EnvO – Environmental Ontology (habitat of pathogen)
- GAZ – Gazeteer (geographic locations)
- FMA – Foundational Model of Anatomy
- DC – Dublin Core (publication metadata)
- PATO – Phenotype
- SO – Sequence Ontology (sequence features)
- Cell – Cell Ontology (types of cells)

- DO – Disease Ontology
- IDO – Infectious Disease Ontology

http://www-test.ebi.ac.uk/industry/Documents/workshop-materials/DiseaseOntologiesAndInformation190608/The%20Influenza%20Infectious%20Disease%20Ontology%20(I-IDO)%20-%20Joanne%20Luciano.pdf

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Metadata (simplified)

Biochemical Reaction

```
<reaction
   id="pyruvate_dehydrogenase_rxn"/>
 <listOfReactants>
   <speciesRef species="NADP+"/>
   <speciesRef species="CoA"/>
   <speciesRef species="pyruvate"/>
 </listOfReactants>
 <listOfProducts>
   <speciesRef species="NADPH"/>
   <speciesRef species="acetyl-CoA"/>
   <speciesRef species="CO2"/>
 </listOfProducts>
<listOfModifers>
   <modifierSpeciesRef
   species="pyruvate_dehydrogenase_E1"/
   >
  </listOfModifiers>
</reaction>
```

Synonyms

```
<species id="pyruvate" metaid="pyruvate">
<annotation xmlns:bp="http://biopax.org/relea
<bp:smallMolecule rdf:ID="#pyruvate" >
  <bp:SYNONYMS>pyroracemic acid</bp:SYN
  <bp:SYNONYMS>2-oxo-propionic acid</bp:S
  <bp:SYNONYMS>alpha-ketopropionic acid</
  <bp:SYNONYMS>2-oxopropanoate</bp:SYN
  <bp:SYNONYMS>2-oxopropanoic acid</bp:S
  <bp:SYNONYMS>BTS</bp:SYNONYMS>
  <bp:SYNONYMS>pyruvic acid</bp:SYNONYM
</bp:smallMolecule>
</annotation>
</species>
```


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Metadata (Webified)

Instead of textual labels

```
<bp:smallMolecule rdf:ID=“#pyruvate”>
  <bp:Xref>
    <bp:unificationXref rdf:ID=“#unificationXref119">
      <bp:DB>LIGAND</bp:DB>
      <bp:ID>c00022</bp:ID>
    </bp:unificationXref>
  </bp:Xref>
</bp:smallMolecule>
```

Use actual URIs

gene	pathway
http://lsrn.org/KEGG:hsa:7498	http://lsrn.org/KEGG_PATHWAY:hsa00983
http://lsrn.org/KEGG:hsa:7498	http://lsrn.org/KEGG_PATHWAY:hsa00230
http://lsrn.org/KEGG:hsa:7498	http://lsrn.org/KEGG_PATHWAY:hsa04146
http://lsrn.org/KEGG:hsa:7498	http://lsrn.org/KEGG_PATHWAY:hsa01100
http://lsrn.org/KEGG:hsa:7498	http://lsrn.org/KEGG_PATHWAY:hsa00232
http://lsrn.org/GeneID:7498	http://lsrn.org/GO:0006144
http://lsrn.org/GeneID:7498	http://lsrn.org/GO:0009115
http://lsrn.org/GeneID:7498	http://lsrn.org/GO:0055086
http://lsrn.org/GeneID:7498	http://lsrn.org/GO:0006195

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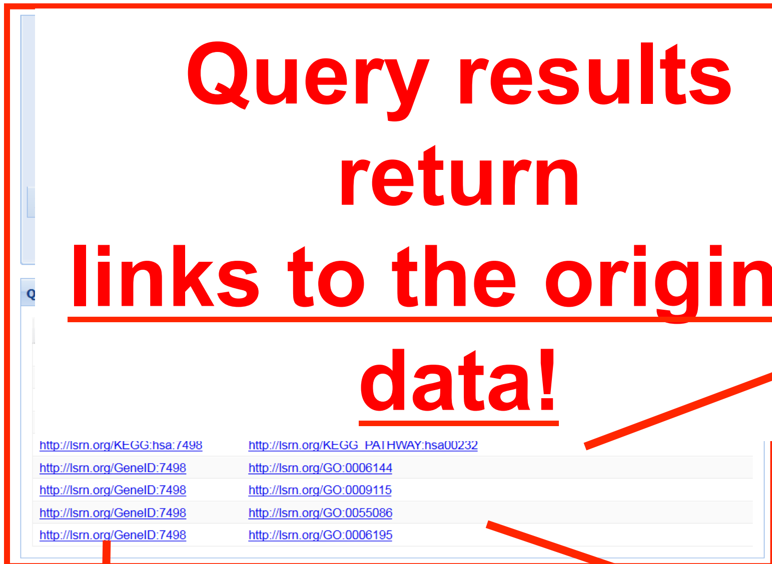
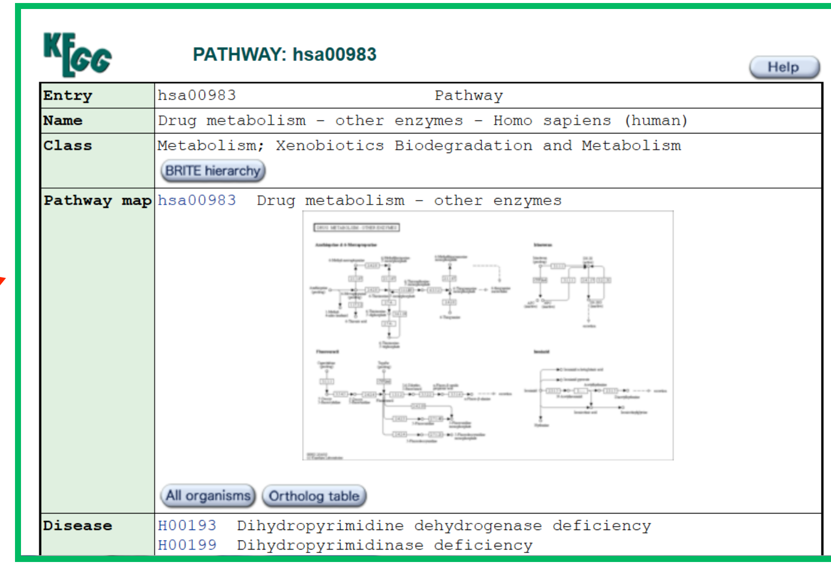
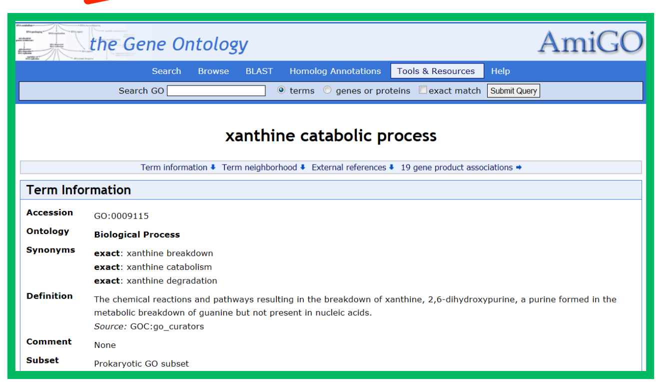
Metadata (Webified)



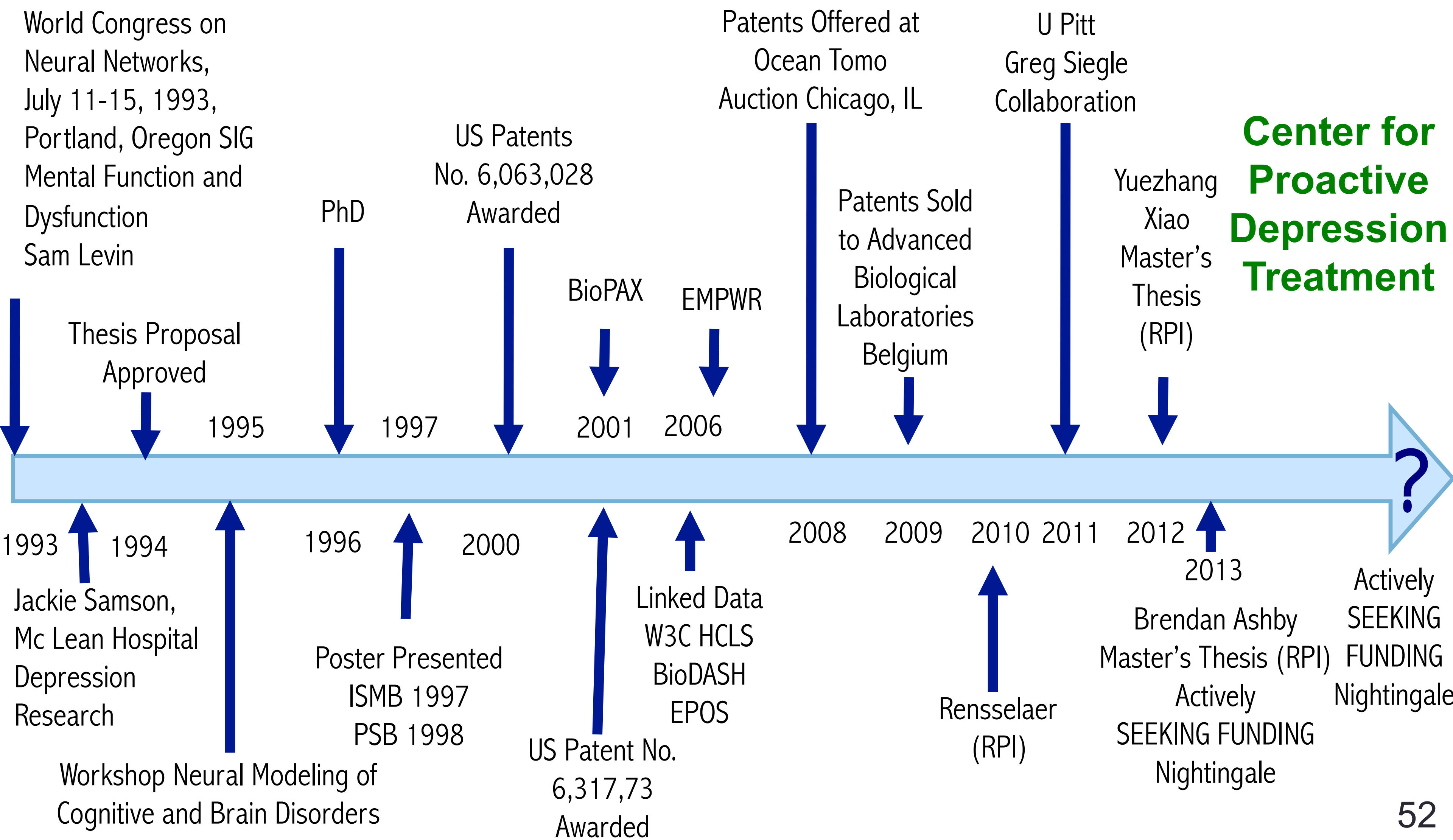




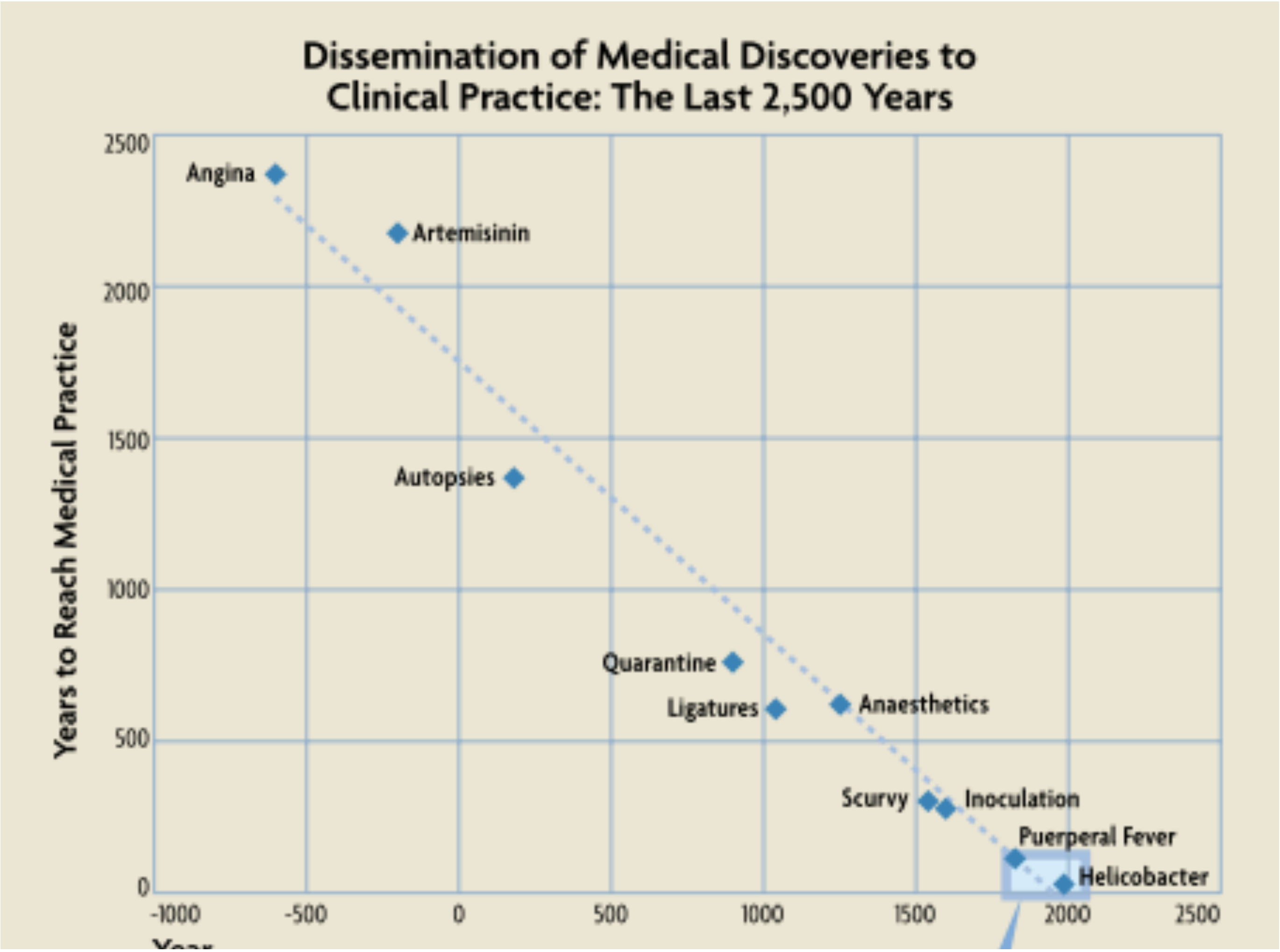
Adapted from Mark Wilkinson webscience20-120829124752-phpapp01

Research to Practice Timeline

(earlier work: 10 years in Software Research & Development and Product Development)



Healthcare Singularity and the age of Semantic Medicine



2,300 years after the first report of angina for the condition to be commonly taught in medical curricula, modern discoveries are being disseminated at an increasingly rapid pace.

http://research.microsoft.com/en-us/collaboration/fourthparadigm/4th_paradigm_book_part2_gillam.pdf

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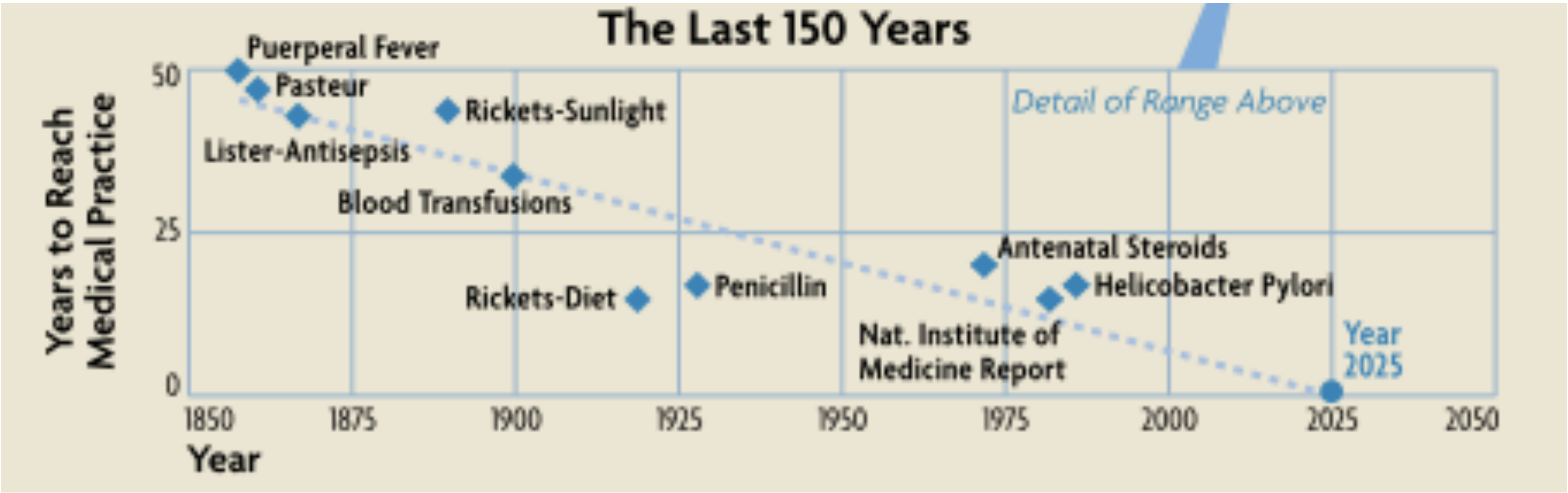
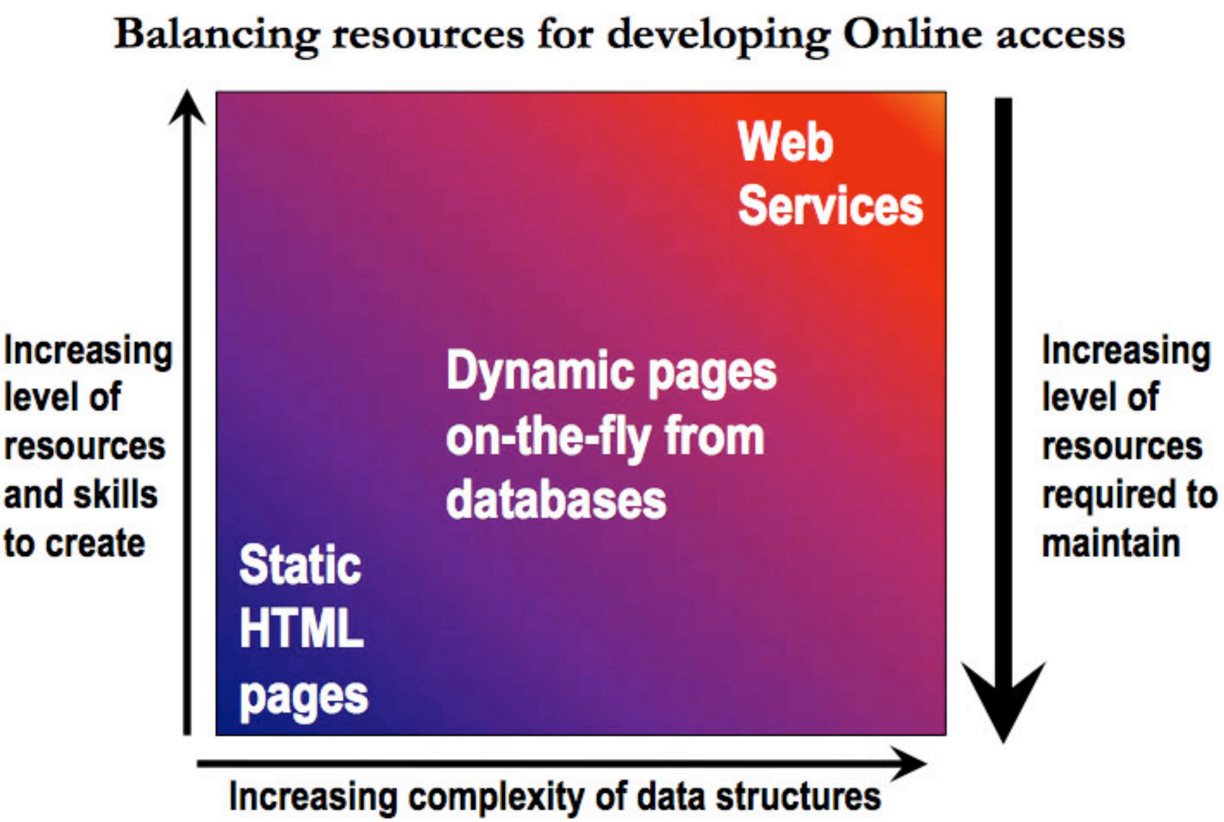
Healthcare Singularity and the age of Semantic Medicine

Focusing on the last 150 years, the trend still appears to be linear, approaching the axis around **2025**.



http://research.microsoft.com/en-us/collaboration/fourthparadigm/4th_paradigm_book_part2_gillam.pdf

Shifts in Data Access



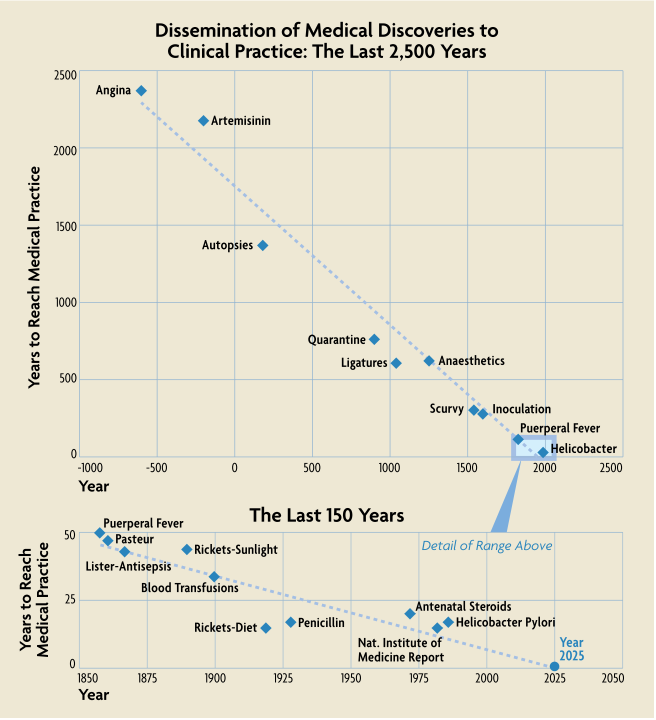
The burden of making data accessible is shifting from the user of the data to the provider of the data

Other people help too!
CSV2RDF4LOD

Context

“Web Driven Medicine”

“While it took 2,300 years after the first report of angina for the condition to be commonly taught in medical curricula, modern discoveries are being disseminated at an increasingly rapid pace. Focusing on the last 150 years, the trend still appears to be linear, approaching the axis around 2025.”

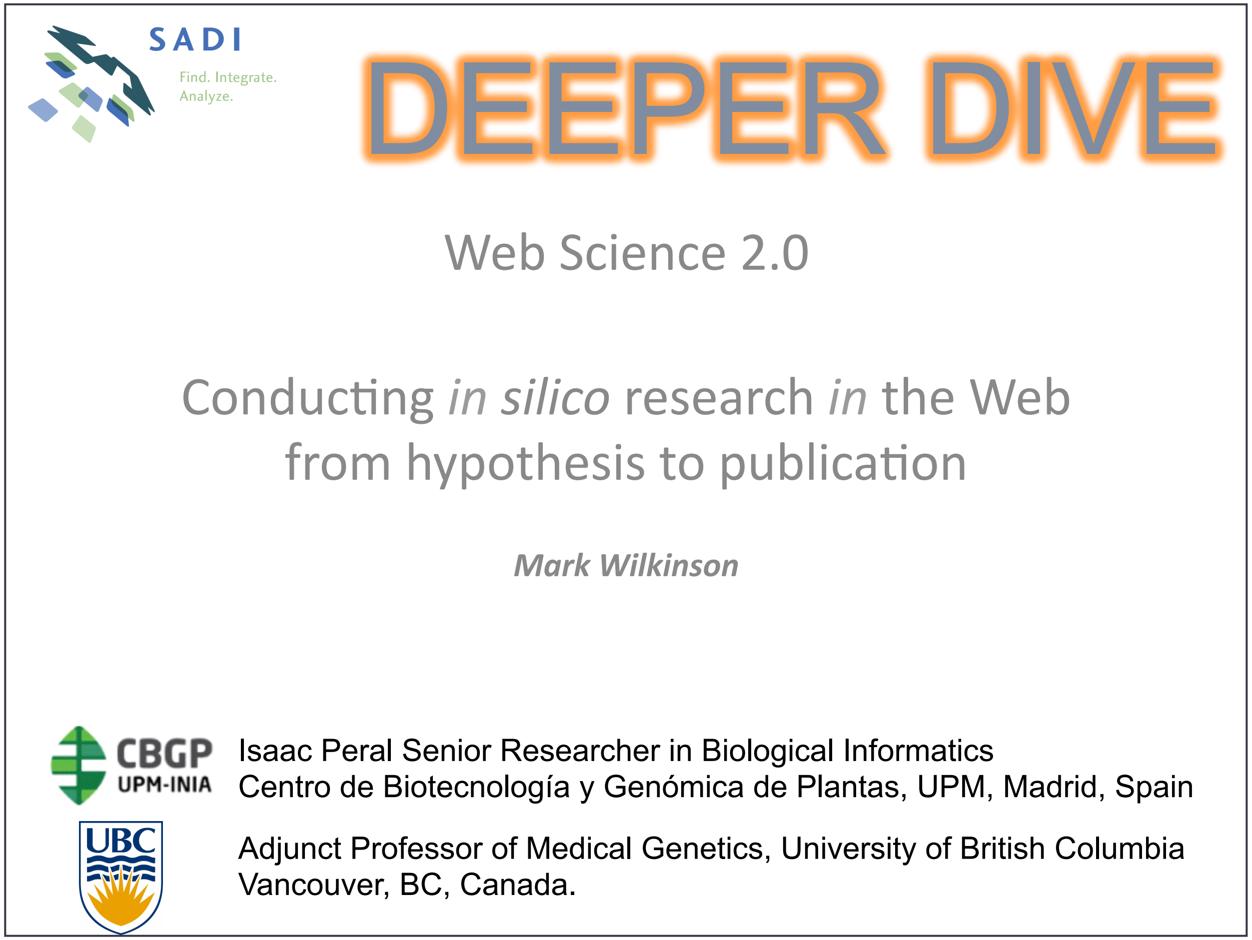


The Healthcare Singularity and the Age of Semantic Medicine, Michael Gillam, et al, *The Fourth Paradigm: Data-Intensive Scientific Discovery* Tony Hey (Editor), 2009

Slide adapted with permission from Joanne Luciano, Presentation at Health Web Science Workshop 2012, Evanston IL, USA June 22, 2012.

Computing in WWW

Conducting
in silico
research
in
the Web



http://www.**slideshare**.net/markmoby/**web-science**-sadi-and-the-singularity

Overview

Introduction (10 minutes)

1. Background
    1. BioMed Domain – Health care and Life Science
    2. Reference and Application
    3. Ontology Granularity and Layout
2. Examples: (35 minutes)
    1. Reference Ontology Examples
        1. BioPAX – Mid level – biological pathways (10)
        2. Gene Ontology ("GO") – Gene annotation (5)
    2. Application Ontology Examples
        1. Influenza Ontology (5)
        2. Best Practices (10)
3. Conclusion (5 minutes)
    1. Process: Start with Use Case, develop prototype, Evaluation
    2. Standards: BioMedical Ontology Best practices (BioPortal, BFO, SIO)
    3. Conferences

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Examples

2 Reference Ontology Examples

- BioPAX – Mid level – biological pathways
- Gene Ontology (“GO”) – Gene annotation

2 Application Ontology Example

- Influenza Ontology
- Translational Medicine Ontology

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Application vs. Reference Ontology

- Reference Ontology
  - Intended as an authoritative source
  - True to the limits of what is known (which does change!)
  - Used by others
- 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

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Translational Medicine Ontology

The Translational Medicine Ontology and Knowledge Base: driving personalized medicine by bridging the gap between bench and bedside

Luciano et al. Journal of Biomedical Semantics 2011, 2(Suppl 2):S1 http://www.jbiomedsem.com/content/2/S2/S1

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Individuals, Not Populations

safety-code.org

A 2D barcode that helps predict a patient's personal response to medical therapy

A **Medicine Safety Code** captures data about a patient's 400 most important pharmacogenomic markers and allergies as a 2D barcode. It can help to predict how medications are metabolized and tolerated by the patient. Medical doctors can use this information to make the delivery of medications safer and more effective.

Quickly retrieve pharmacogenomic markers of patients when needed

No central storage of data is necessary, giving patients full control over their personal health information.

http://safety-code.org/

Photo: http://www.flickr.com/photos/sepblog/4014143391/

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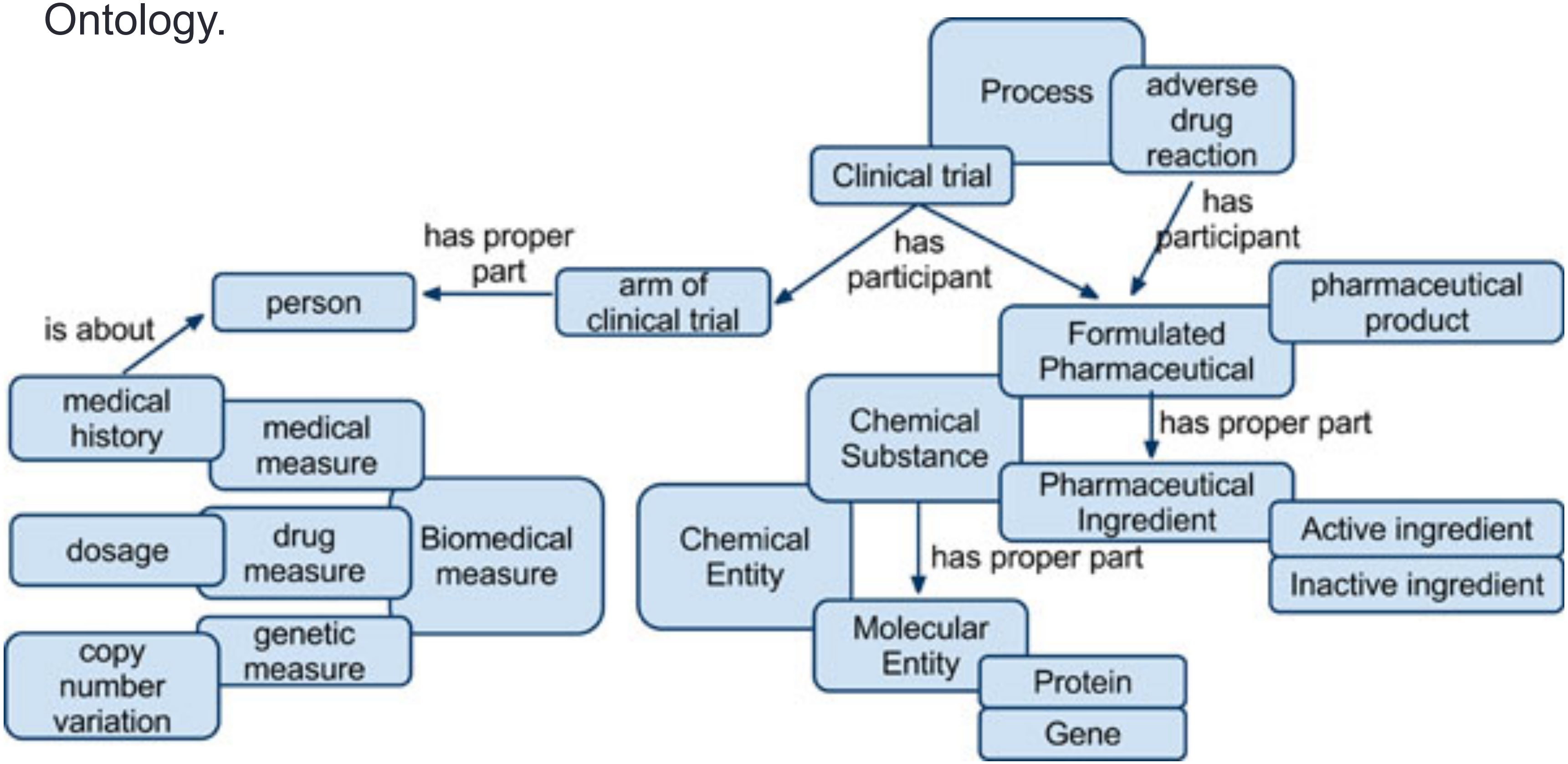
Translational Medicine Ontology

Overview of selected types, subtypes (overlap) and existential restrictions (arrows) in the Translational Medicine Ontology.



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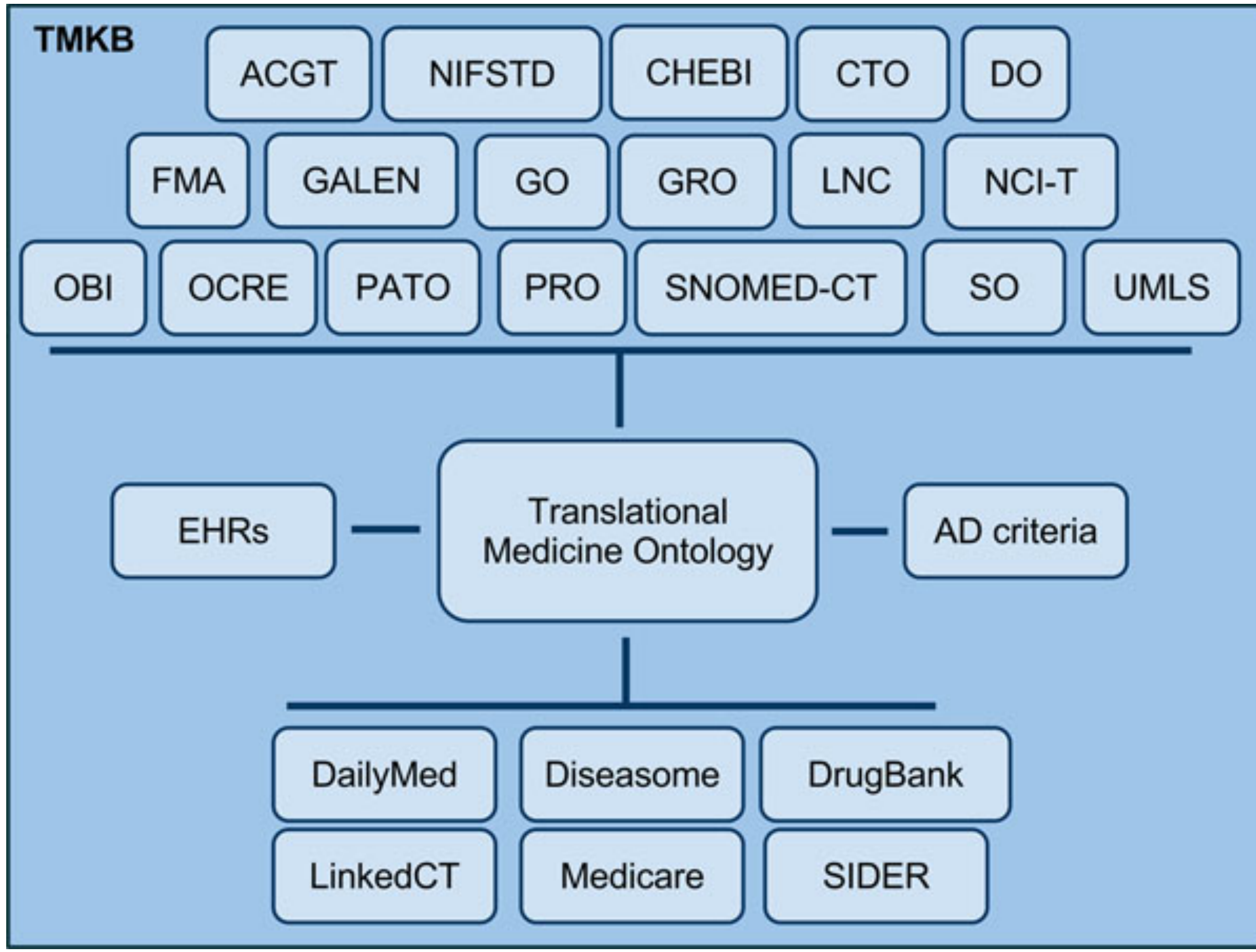
Translational Medicine Knowledge Base

Translational Medicine Ontology with mappings to ontologies and terminologies listed in the NCBO BioPortal.

The TMO provides a global schema for Indivo-based electronic health records (EHRs) and can be used with formalized criteria for Alzheimer's Disease. The TMO maps types from Linking Open Data sources.



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Overview

Introduction (10 minutes)

1. Background
    1. BioMed Domain – Health care and Life Science
    2. Reference and Application
    3. Ontology Granularity and Layout
2. Examples: (40 minutes)
    1. Reference Ontology Examples
        1. UMLS – High level across biomedicine (5)
        2. BioPAX – Mid level – biological pathways (10)
        3. Gene Ontology ("GO") – Gene annotation (5)
    2. Application Ontology Examples
        1. Influenza Ontology (5)
        2. Best Practices (10)
3. Conclusion (5 minutes)
    1. Process: Start with Use Case, develop prototype, Evaluation
    2. Standards: BioMedical Ontology Best practices (BioPortal, BFO, SIO)
    3. Conferences

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Best Practices

Semantic Web Methodology & Technology Development Process

Fox, Peter & McGuinness, Deborah 2008
http://tw.rpi.edu/web/doc/TWC_SemanticWebMethodology

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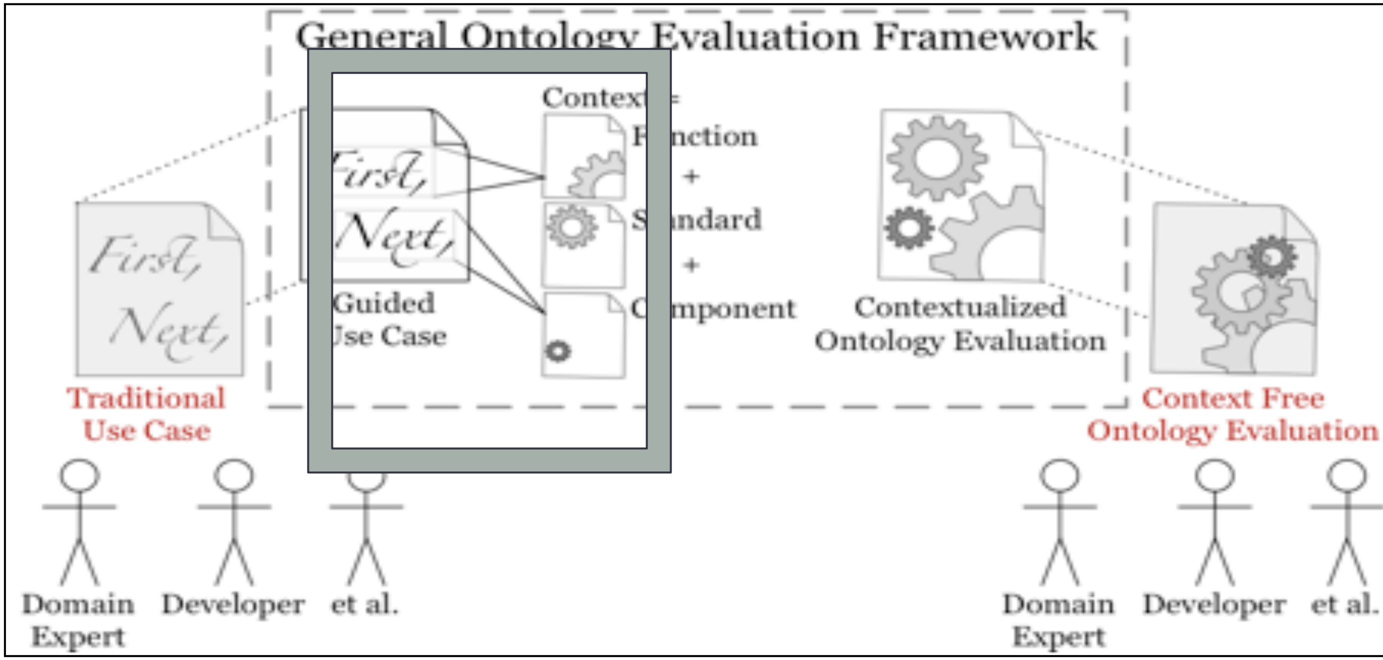
Generalized Ontology Evaluation Framework (GOEF)



Two stages:

1. Recast use case into its components:
   Three Levels of Evaluation
2. Evaluate components using objective metrics

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BioPortal

http://bioportal.bioontology.org/

Provides access to commonly used biomedical ontologies and to tools for working with them. BioPortal allows you to

- **Browse**
  - the library of ontologies
  - mappings between terms in different ontologies
  - a selection of projects that use BioPortal resources
- **Search**
  - biomedical resources for a term
  - for a term across multiple ontologies
- **Receive recommendations**
  - on which ontologies are most relevant for a corpus
- **Annotate text**
  - with terms from ontologies

All information available through the BioPortal Web site is also available through the NCBO Web service REST API. Please see REST API documentation for more information.

http://www.bioontology.org/wiki/index.php/NCBO_REST_services

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Backup Slides

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HL-7 and RIM

HL-7 and RIM: http://www.w3.org/2013/HCLS-tutorials/RIM/#%286%29

- **RDF RIM Tutorial** Eric Prud'hommeaux, <eric@w3.org>
- Basic understanding of the structure of how data written in HL7's RIM can be expressed in RDF.
- It is not a substitute for HL7's documentation, but instead the author's notion of a quick way to familiarize oneself with the concepts and terms used in the RIM and how the graph structure of RDF is a natural way to represent this data.

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Scope
Ontology Uses

- Knowledge Management
  - Annotate data (such as genomes)
  - Access information (search, find, and access)
  - Map across ontologies relate
- Data integration and exchange
  - Model dynamic cellular processes
  - Identify Drug Interactions
- Decision support
  - SafetyCodes
  - Diabetic Care
  - Lab Alerts

(Bodenreider YBMI 2008)
http://themindwobbles.wordpress.com/2009/05/04/olivier-bodenreider-nlm-best-practices-pitfalls-and-positives-cbo-2009/

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