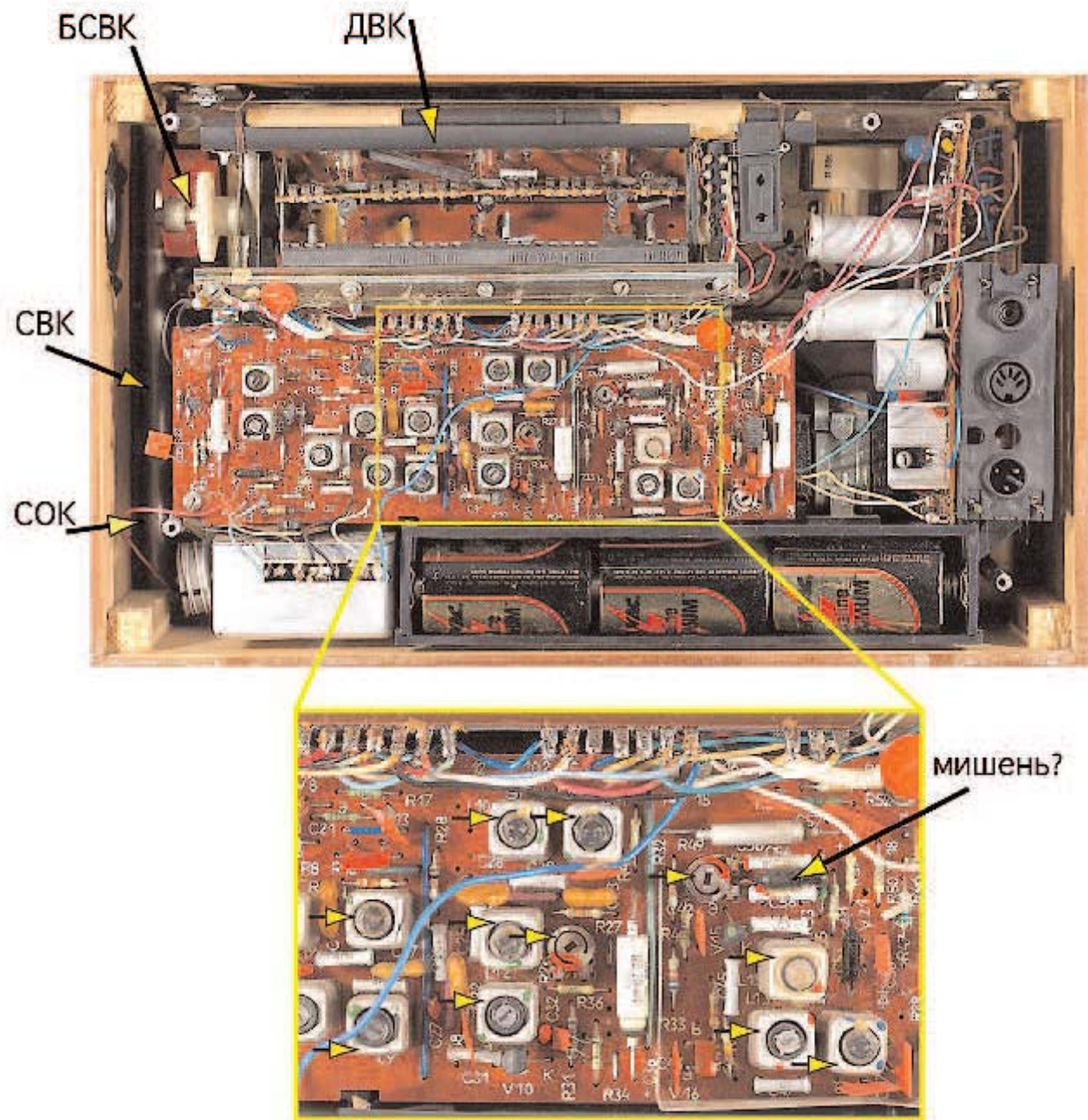

**The W3C Health Care and Life Sciences Interest
Group:
State of the Interest Group**

**M. Scott Marshall
co-chair HCLS IG
Leiden University Medical Center
&
University of Amsterdam**

Biology in a nutshell: Bigger isn't better

- DNA Dogma
 - Transcription = DNA -> mRNA -> Protein
- Molecular pathways allow biologists to 'connect' one process to another.
- Huntington's mutation mapped in 1993 yet there is still no understanding of the mechanism that causes the neurodegeneration.
- Semantic models are necessary to create a 'systems view' of biology.

Can a Biologist Fix a Radio?



What is *knowledge* ?

“data”, “information”, “facts”, “knowledge”

**Knowledge is a statement
that can be tested for truth.**

(by a machine)

Otherwise, computing can't add much

Knowledge Capture

- ***How will we acquire the knowledge?***
 - *Literature*
 - *Other forms of discourse*
 - *Data analysis*
- ***How will we represent and store it?***
 - *In Semantic Web formats such as RDF, OWL, RIF*

What will we do with knowledge?

- ***How will we use it?***
 - *Query it*
 - *Reason across it*
 - *Integrate it with other data*
- ***Link it up***



Linked Data Principles

1. Use URIs as names for things.
2. Use HTTP URIs so that people can look up those names.
3. When someone looks up a URI, provide useful RDF information.
4. Include RDF statements that link to other URIs so that they can discover related things.

- Tim Berners-Lee 2007
- <http://www.w3.org/DesignIssues/LinkedData.html>

Background of the HCLS IG

- Originally chartered in 2005
 - Chairs: Eric Neumann and Tonya Hongsermeier
- Re-chartered in 2008
 - Chairs: Scott Marshall and Susie Stephens
 - Team contact: Eric Prud'hommeaux
- Broad industry participation
 - Over 100 members
 - Mailing list of over 600
- Background Information
 - <http://www.w3.org/2001/sw/hcls/>
 - <http://esw.w3.org/topic/HCLSIG>

Mission of HCLS IG

- The mission of HCLS is to develop, advocate for, and support the use of Semantic Web technologies for
 - Biological science
 - **Translational medicine**
 - Health care

- These domains stand to gain tremendous benefit by adoption of Semantic Web technologies, as they depend on the interoperability of information from many domains and processes for efficient decision support

Group Activities

- Document **use cases** to aid individuals in understanding the business and technical benefits of using Semantic Web technologies
- Document **guidelines** to accelerate the adoption of the technology
- Implement a selection of the use cases as proof-of-concept **demonstrations**
- Develop high-level **vocabularies**
- **Disseminate information** about the group's work at government, industry, and academic events

What are we about?

- Creating applications that solve *real* problems with *real* data and documenting what we did.
- Deliverables:
 - Software
 - Methodologies
 - Vocabularies
 - Documentation
 - Journals, workshops, conferences
 - W3C notes

Current Task Forces

- **BioRDF – integrated neuroscience knowledge base**
 - Kei Cheung (Yale University)
- **Clinical Observations Interoperability – patient recruitment in trials**
 - Vipul Kashyap (Cigna Healthcare)
- **Linking Open Drug Data – aggregation of Web-based drug data**
 - Anja Jentzsch (Free University Berlin)
- **Pharma Ontology – high level patient-centric ontology**
 - Christi Denney (Eli Lilly)
- **Scientific Discourse – building communities through networking**
 - Tim Clark (Harvard University)
- **Terminology – Semantic Web representation of existing resources**
 - John Madden (Duke University)

BioRDF Task Force

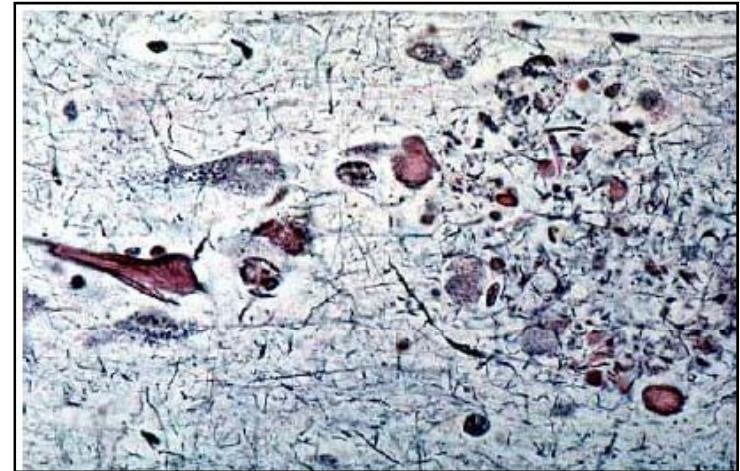
- **Kei Cheung (Yale University)**
- **Helena Deus (University of Texas)**
- **Rob Frost (Vector C)**
- **Kingsley Idehen (OpenLink Software)**
- **Scott Marshall (University of Amsterdam)**
- **Adrian Paschke (Freie Universitat Berlin)**
- **Eric Prud'hommeaux (W3C)**
- **Satya Sahoo (Wright State University)**
- **Matthias Samwald (DERI and Konrad Lorenz Institute)**
- **Jun Zhao (Oxford University)**

BioRDF: Answering Questions

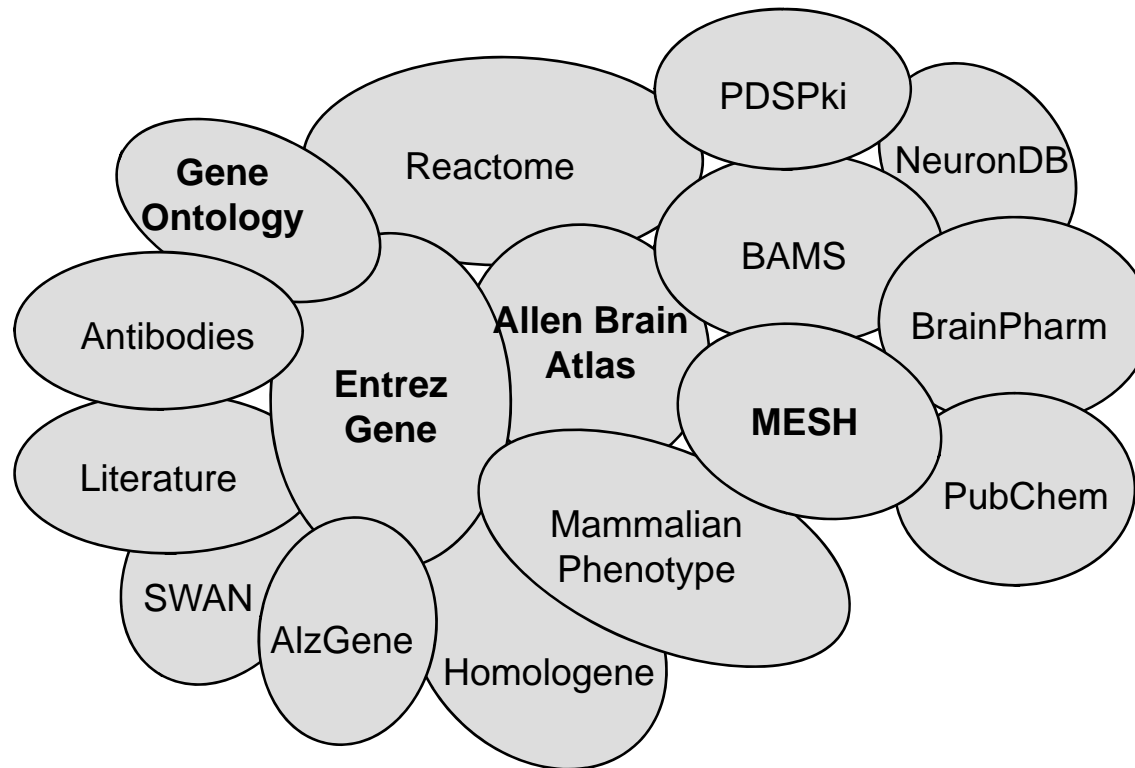
- **Goals:** Get answers to questions posed to a body of collective knowledge in an effective way
- **Knowledge used:** Publicly available databases, and text mining
- **Strategy:** Integrate knowledge using careful modeling, exploiting Semantic Web standards and technologies

BioRDF: Looking for Targets for Alzheimer's

- **Signal transduction pathways are considered to be rich in “druggable” targets**
- **CA1 Pyramidal Neurons are known to be particularly damaged in Alzheimer's disease**
- **Casting a wide net, can we find candidate genes known to be involved in signal transduction and active in Pyramidal Neurons?**



BioRDF: Integrating Heterogeneous Data



BioRDF: SPARQL Query

```
prefix go: <http://purl.org/obo/owl/GO#>
prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#>
prefix owl: <http://www.w3.org/2002/07/owl#>
prefix mesh: <http://purl.org/commons/record/mesh/>
prefix sc: <http://purl.org/science/owl/sciencecommons/>
prefix ro: <http://www.obofoundry.org/ro/ro.owl#>

select ?genename ?processname
where
{
  graph <http://purl.org/commons/hcls/pubmesh>
  {
    ?paper ?p mesh:D017966
    ?article sc:identified_by PMID ?paper.
    ?gene sc:describes_gene_or_gene_product_mentioned_by ?article.
  }
  graph <http://purl.org/commons/hcls/goa>
  {
    ?protein rdfs:subClassOf ?res.
    ?res owl:onProperty ro:has_function.
    ?res owl:someValuesFrom ?res2.
    ?res2 owl:onProperty ro:realized_as.
    ?res2 owl:someValuesFrom ?process.
  }
  graph <http://purl.org/commons/hcls/20070416/classrelations>
  {
    {?process <http://purl.org/obo/owl/obo#part_of> go:GO_0007166}
    union
    {?process rdfs:subClassOf go:GO_0007166 }
    ?protein rdfs:subClassOf ?parent.
    ?parent owl:equivalentClass ?res3.
    ?res3 owl:hasValue ?gene.
  }
  graph <http://purl.org/commons/hcls/gene>
  {
    ?gene rdfs:label ?genename }
  graph <http://purl.org/commons/hcls/20070416>
  {
    ?process rdfs:label ?processname }
}
```

Mesh: Pyramidal Neurons



Pubmed: Journal Articles



Entrez Gene: Genes



GO: Signal Transduction

Inference required

BioRDF: Results: Genes, Processes

- DRD1, 1812
- ADRB2, 154
- ADRB2, 154
- DRD1IP, 50632
- DRD1, 1812
- DRD2, 1813
- GRM7, 2917
- GNG3, 2785
- GNG12, 55970
- DRD2, 1813
- ADRB2, 154
- CALM3, 808
- HTR2A, 3356
- DRD1, 1812
- SSTR5, 6755
- MTNR1A, 4543
- CNR2, 1269
- HTR6, 3362
- GRIK2, 2898
- GRIN1, 2902
- GRIN2A, 2903
- GRIN2B, 2904
- ADAM10, 102
- GRM7, 2917
- LRP1, 4035
- ADAM10, 102
- ASCL1, 429
- HTR2A, 3356
- ADRB2, 154
- PTPRG, 5793
- EPHA4, 2043
- NRTN, 4902
- CTNND1, 1500

adenylate cyclase activation
adenylate cyclase activation
arrestin mediated desensitization of G-protein coupled receptor protein signaling pathway
dopamine receptor signaling pathway
dopamine receptor, adenylylase activating pathway
dopamine receptor, adenylylase inhibiting pathway
G-protein coupled receptor protein signaling pathway
G-protein coupled receptor protein signaling pathway
G-protein coupled receptor protein signaling pathway
G-protein coupled receptor protein signaling pathway
G-protein coupled receptor protein signaling pathway
G-protein coupled receptor protein signaling pathway
G-protein signaling, coupled to cyclic nucleotide second messenger
G-protein signaling, coupled to cyclic nucleotide second messenger
G-protein signaling, coupled to cyclic nucleotide second messenger
G-protein signaling, coupled to cyclic nucleotide second messenger
G-protein signaling, coupled to cyclic nucleotide second messenger
glutamate signaling pathway
glutamate signaling pathway
glutamate signaling pathway
glutamate signaling pathway
integrin-mediated signaling pathway
negative regulation of adenylylase activity
negative regulation of Wnt receptor signaling pathway
Notch receptor processing
Notch signaling pathway
serotonin receptor signaling pathway
transmembrane receptor protein tyrosine kinase activation (dimerization)
transmembrane receptor protein tyrosine kinase signaling pathway
transmembrane receptor protein tyrosine kinase signaling pathway
transmembrane receptor protein tyrosine kinase signaling pathway
Wnt receptor signaling pathway

Many of the genes
are related to AD
through gamma
secretase
(presenilin) activity

Current activities

- **HCLS KB's**
 - DERI Galway and Freie Universitat Berlin
- **Query federation and aTag**
- **Publication**
 - Cheung KH, Frost HR, Marshall MS, Prud'hommeaux E, Samwald M, Zhao J, Paschke A. (2009). A Journey to Semantic Web Query Federation in Life Sciences. *BMC Bioinformatics*, 10(Suppl 10):S10.

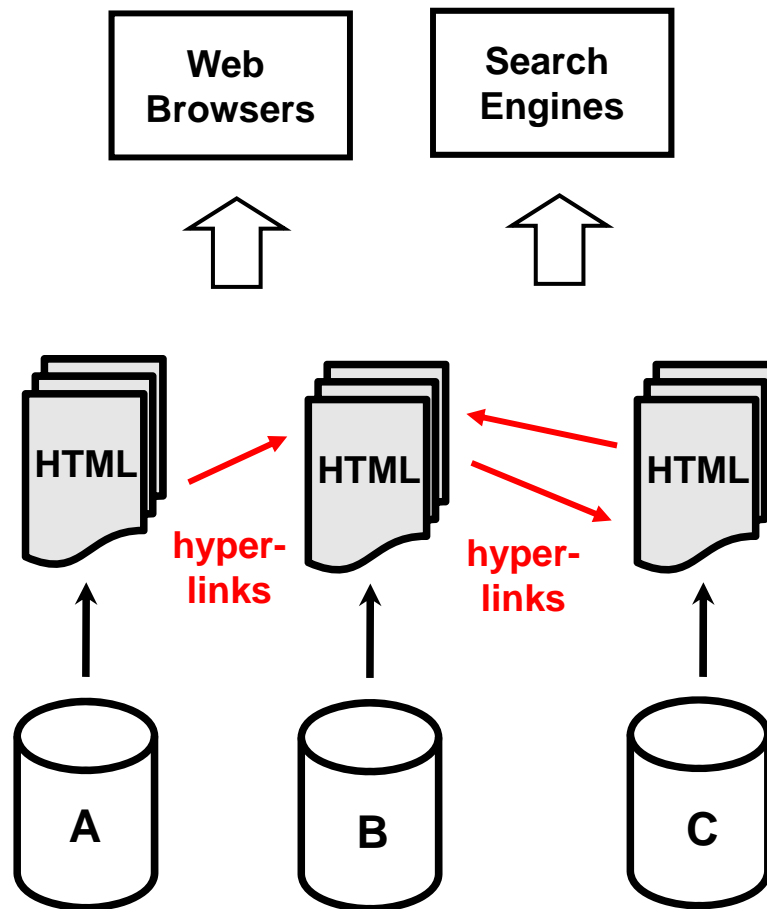
Near future activities

- **Expansion of query federation**
 - **Incorporation of new data types including neuroscience microarray data, image data and TCM data**
 - **Inter-community collaboration with NIF (NeuroLex) and MGED (EBI Expression Atlas)**

Linking Open Drug Data

- HCLSIG task started October 1st, 2008
- Primary Objectives
 - Survey publicly available data sets about drugs
 - Explore interesting questions from pharma, physicians and patients that could be answered with Linked Data
 - Publish and interlink these data sets on the Web
- Participants: Bosse Andersson, Chris Bizer, Kei Cheung, Don Doherty, Oktie Hassanzadeh, Anja Jentsch, Scott Marshall, Eric Prud'hommeaux, Matthias Samwald, Susie Stephens, Jun Zhao

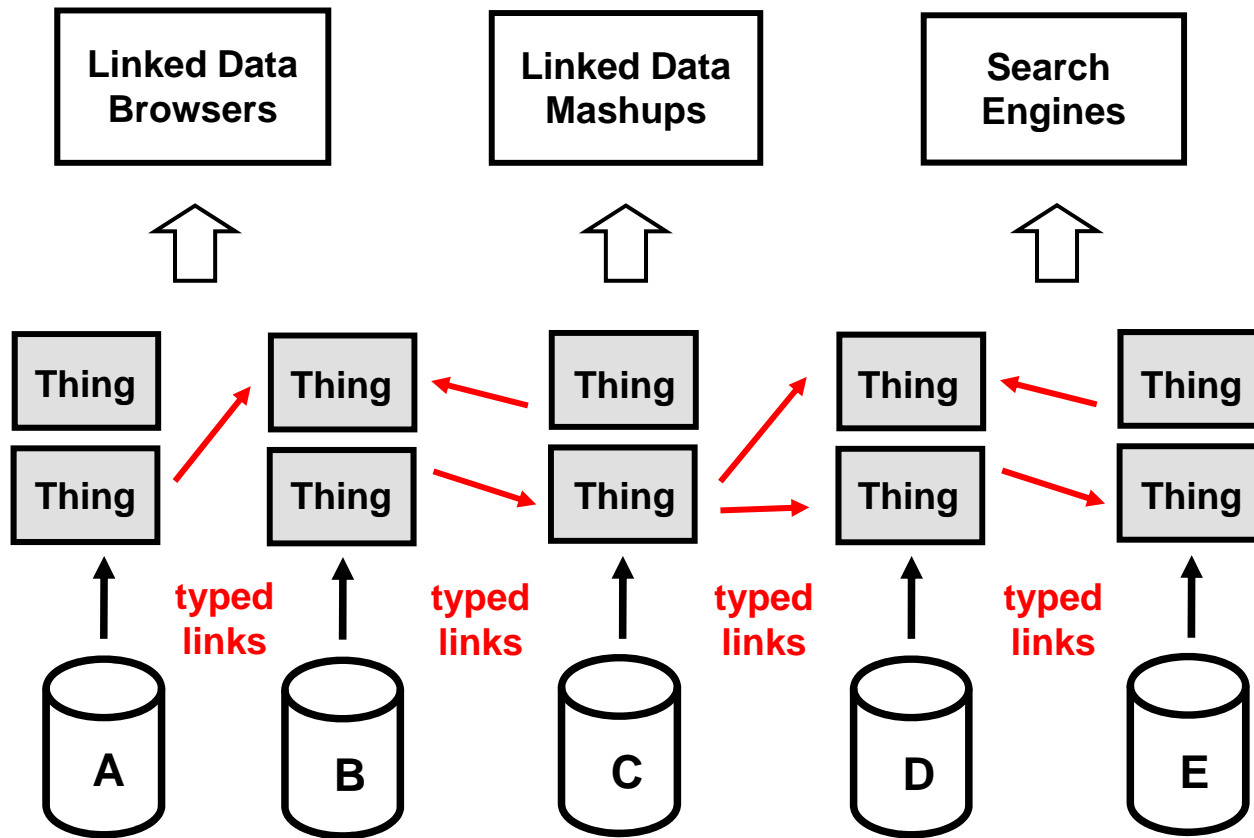
The Classic Web



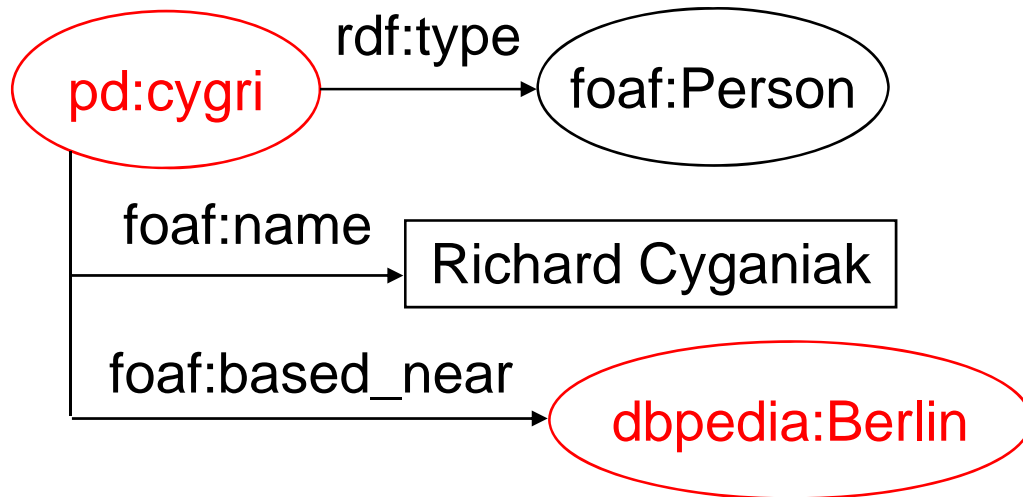
- Single information space
- Built on URIs
 - globally unique IDs
 - retrieval mechanism
- Built on Hyperlinks
 - are the glue that holds everything together

Linked Data

Use Semantic Web technologies to publish structured data on the Web and set links between data from one data source and data from another data sources



Data Objects Identified with HTTP URIs

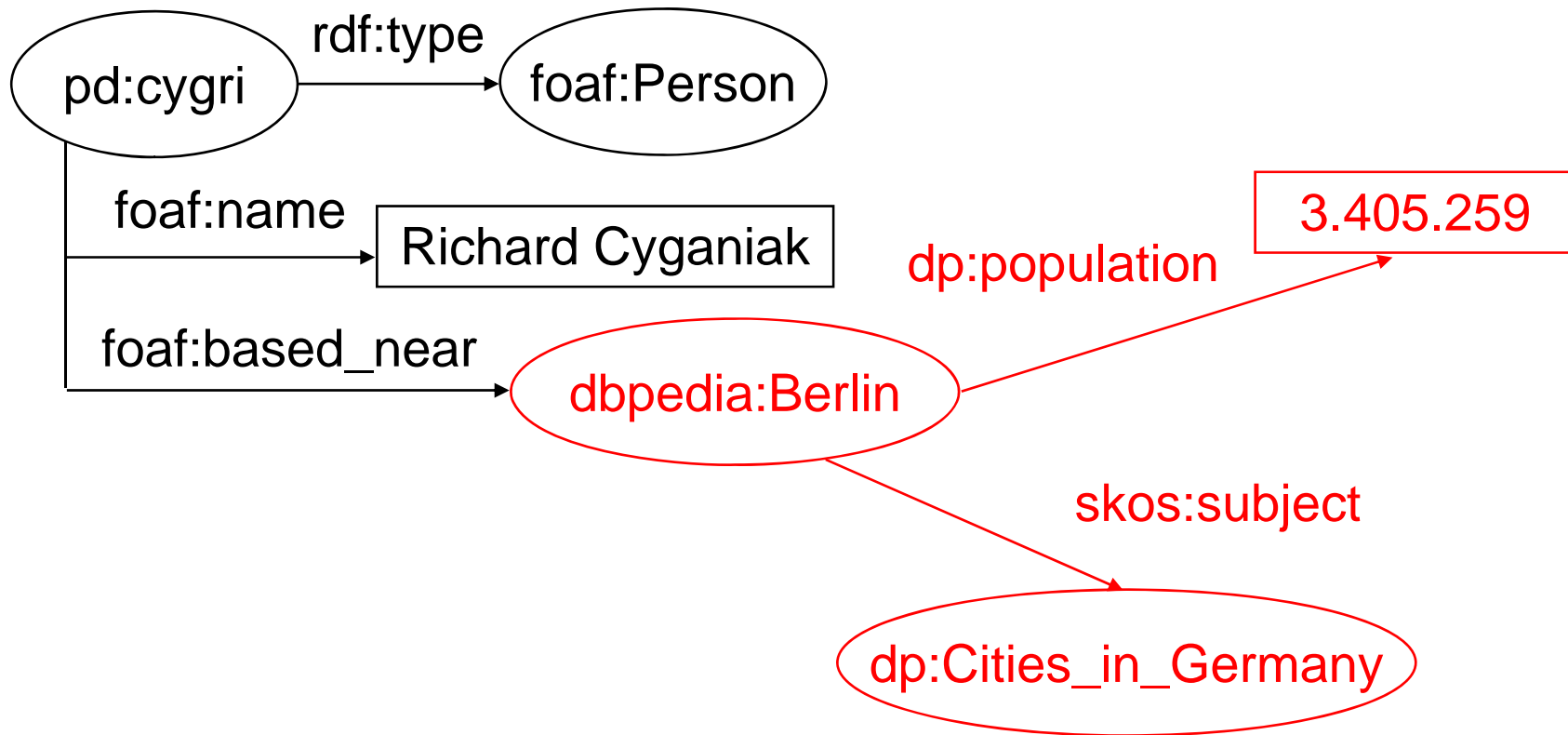


pd:cygri = <http://richard.cyganiak.de/foaf.rdf#cygri>

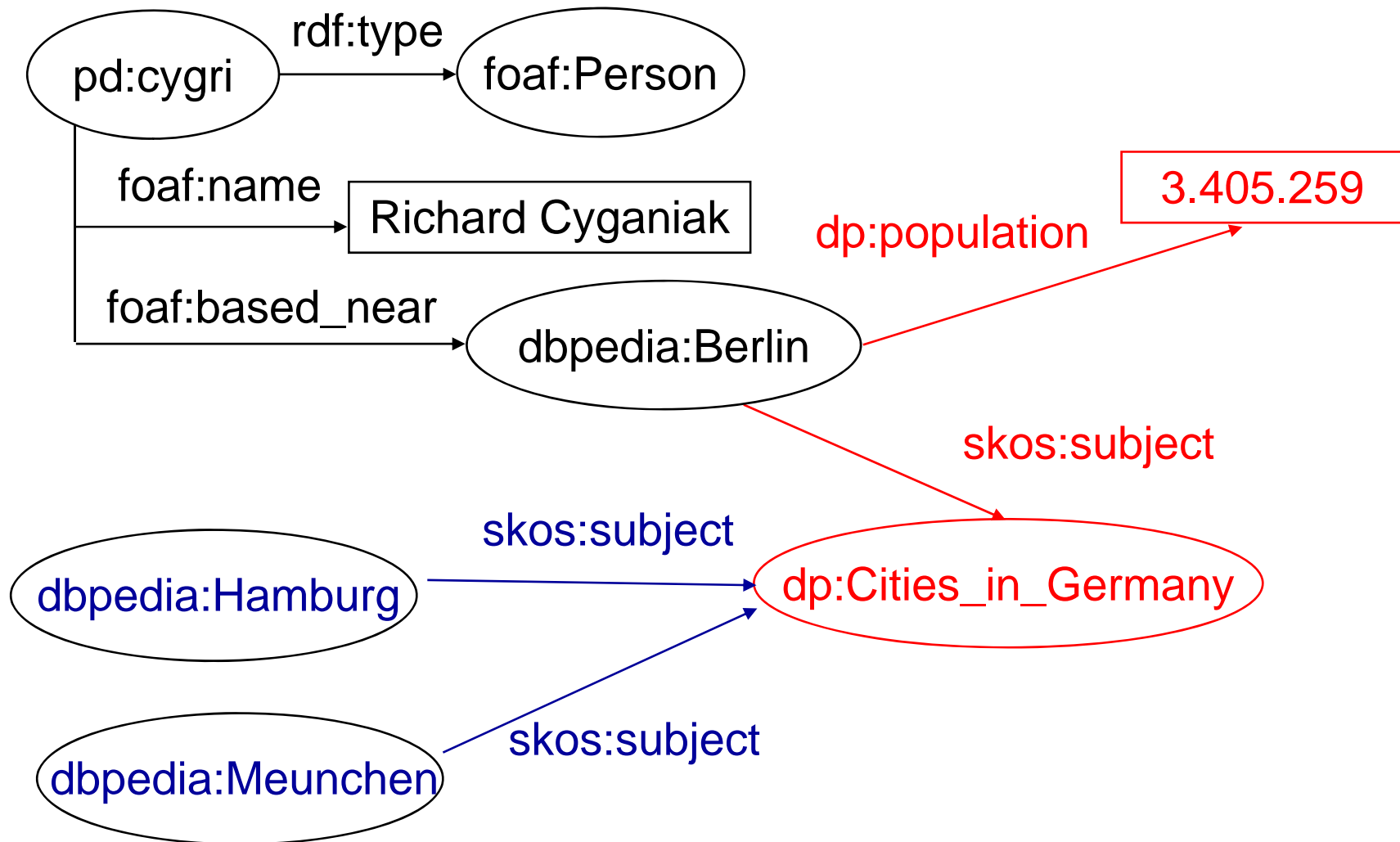
dbpedia:Berlin = <http://dbpedia.org/resource/Berlin>

Forms an RDF link between two data sources

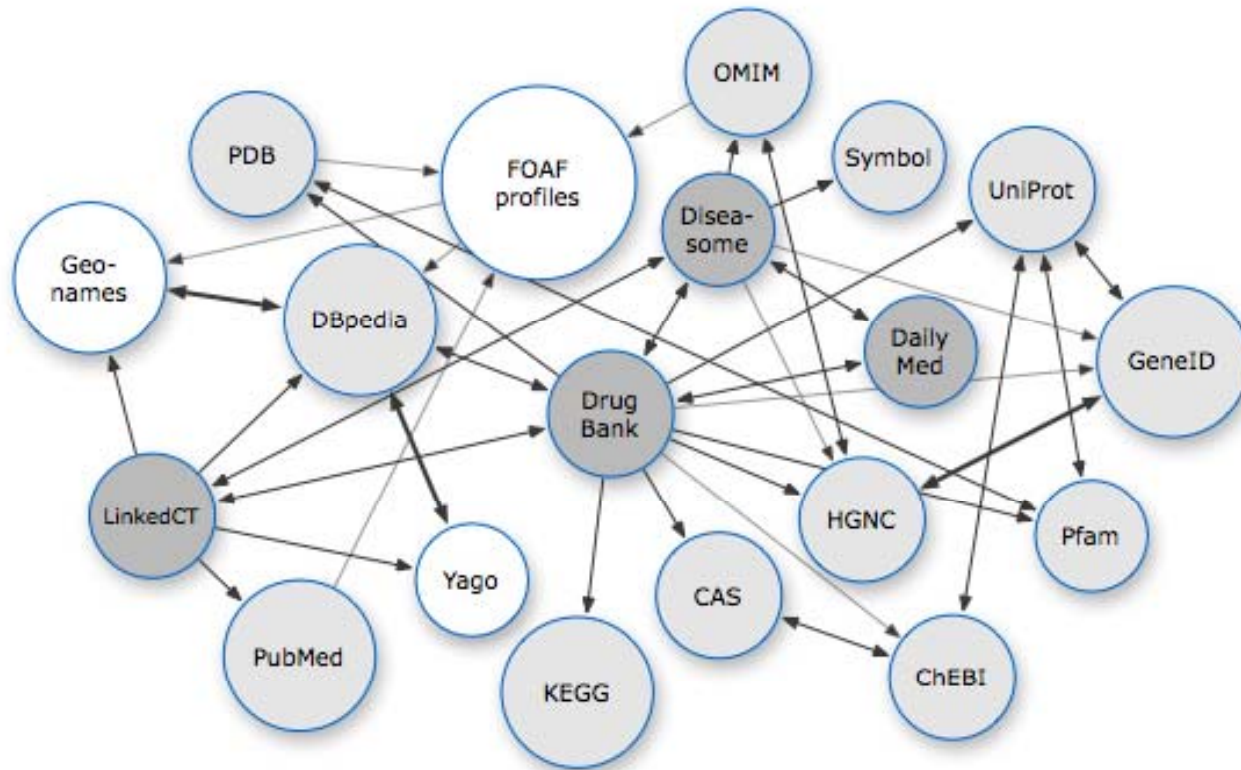
Dereferencing URIs over the Web



Dereferencing URIs over the Web



LODD Data Sets



COI Task Force

- **Task Lead: Vipul Kashap**
- **Participants: Eric Prud'hommeaux, Helen Chen, Jyotishman Pathak, Rachel Richesson, Holger Stenzhorn**

COI: Bridging Bench to Bedside

- How can existing Electronic Health Records (EHR) formats be reused for patient recruitment?
- Quasi standard formats for clinical data:
 - HL7/RIM/DCM – healthcare delivery systems
 - CDISC/SDTM – clinical trial systems
- How can we map across these formats?
 - Can we ask questions in one format when the data is represented in another format?

COI: Use Case

Pharmaceutical companies pay a lot to test drugs

Pharmaceutical companies express protocol in CDISC

-- precipitous gap --

Hospitals exchange information in HL7/RIM

Hospitals have relational databases

Inclusion Criteria

- Type 2 diabetes on diet and exercise therapy or
- monotherapy with metformin, insulin
- secretagogue, or alpha-glucosidase inhibitors, or
- a low-dose combination of these at 50%
- maximal dose. Dosing is stable for 8 weeks prior
- to randomization.
- ...
- ?patient takes metformin .

Exclusion Criteria

Use of warfarin (Coumadin), clopidogrel (Plavix) or other anticoagulants.

...

?patient doesNotTake anticoagulant .

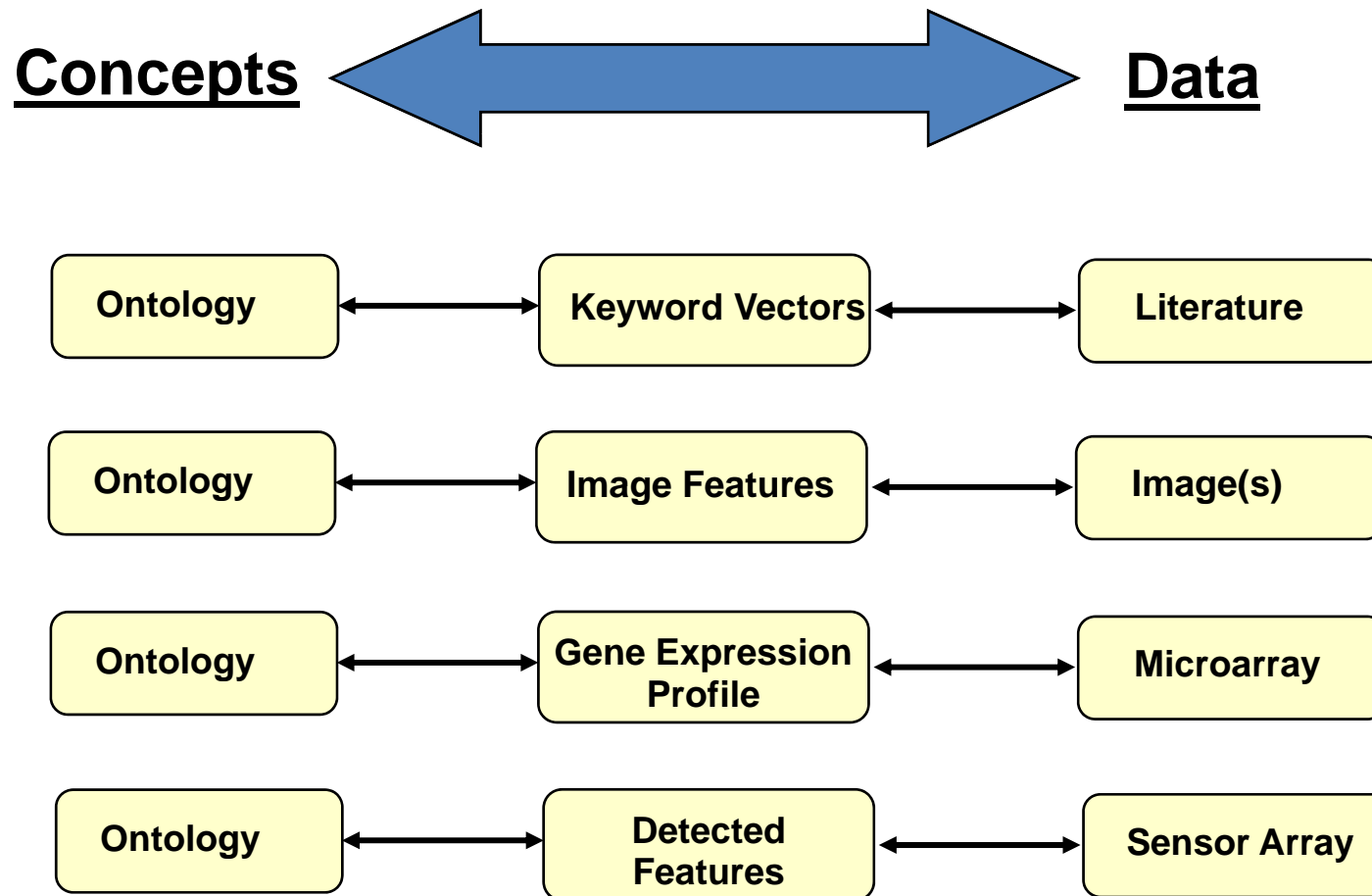
Criteria in SPARQL

```
?medication1 sdtm:subject ?patient ;  
  spl:activeIngredient ?ingredient1 .  
?ingredient1 spl:classCode 6809 . #metformin  
  
OPTIONAL {  
  ?medication2 sdtm:subject ?patient ;  
    spl:activeIngredient ?ingredient2 .  
  ?ingredient2 spl:classCode 11289 . #anticoagulant  
} FILTER (!BOUND(?medication2))
```

Terminology Task Force

- **Task Lead: John Madden**
- **Participants: Chimezie Ogbuji, M. Scott Marshall, Helen Chen, Holger Stenzhorn, Mary Kennedy, Xiashu Wang, Rob Frost, Jonathan Borden, Guoqian Jiang**

Features: the “bridge” to meaning



Terminology: Overview

- **Goal is to identify use cases and methods for extracting Semantic Web representations from existing, standard medical record terminologies, e.g. UMLS**
- **Methods should be reproducible and, to the extent possible, not lossy**
- **Identify and document issues along the way related to identification schemes, expressiveness of the relevant languages**
- **Initial effort will start with SNOMED-CT and UMLS Semantic Networks and focus on a particular sub-domain (e.g. pharmacological classification)**

SKOS & the 80/20 principle: map “down”

- Minimal assumptions about expressiveness of source terminology
- No assumed formal semantics (no model theory)
- Treat it as a knowledge “map”
- Extract 80% of the utility without risk of falsifying intent

W3C Working Draft



SKOS Simple Knowledge Organization System Reference

W3C Working Draft 29 August 2008

This version:

<http://www.w3.org/TR/2008/WD-skos-reference-20080829/>

Latest version:

<http://www.w3.org/TR/skos-reference>

Previous version:

<http://www.w3.org/TR/2008/WD-skos-reference-20080609/>

Editors:

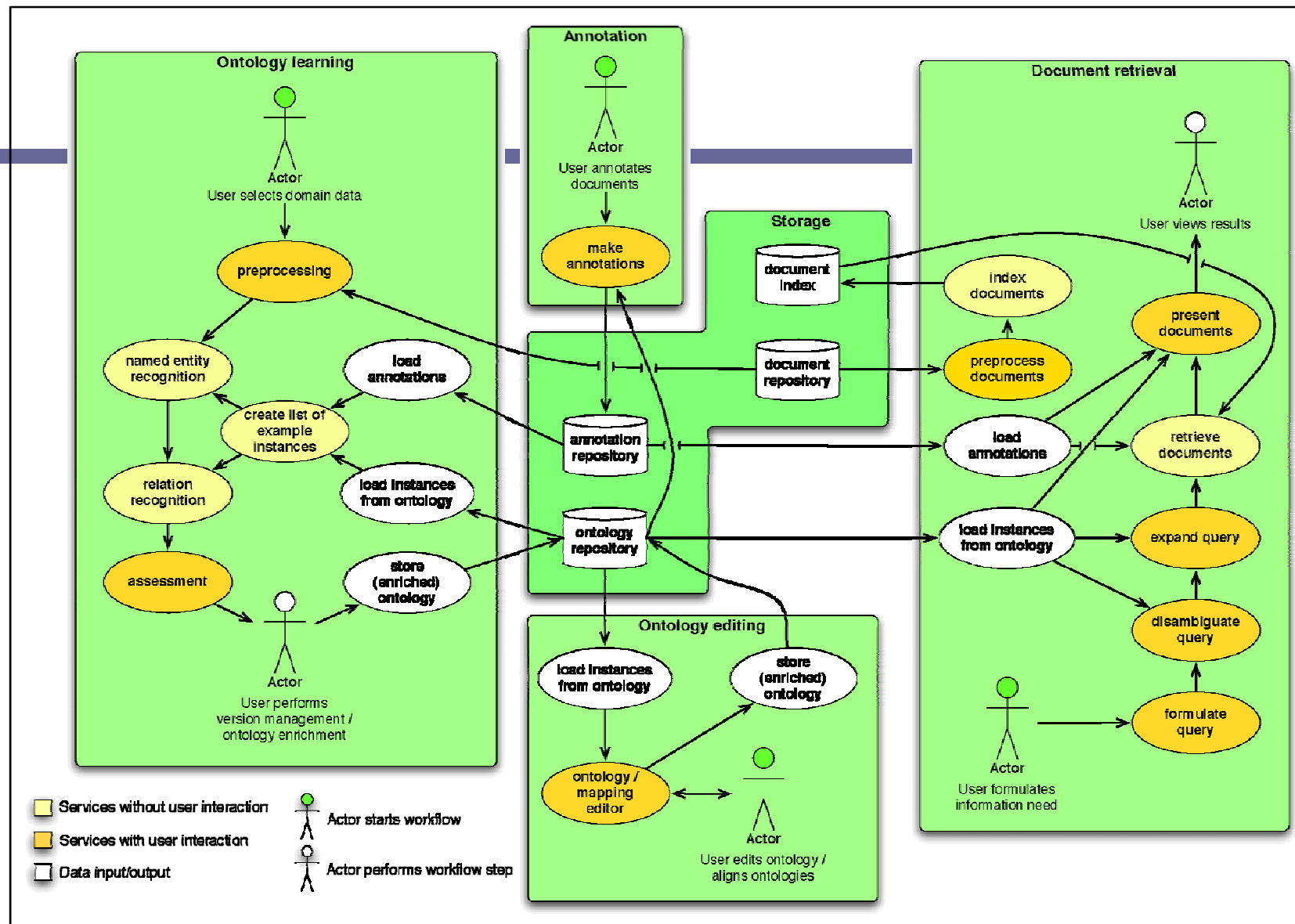
[Alistair Miles](#), STFC Rutherford Appleton Laboratory / University of Oxford
[Sean Bechhofer](#), University of Manchester

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Abstract

This document defines the Simple Knowledge Organization System (SKOS), a comm the Web.

Many knowledge organization systems, such as thesauri, taxonomies, classification s used in similar applications. SKOS captures much of this similarity and makes it explic



The **AIDA** toolbox
for knowledge extraction and knowledge management
in a Virtual Laboratory for e-Science

SNOMED CT/SKOS under AIDA: retrieve

The screenshot displays the AIDA web interface, which is used for retrieving SNOMED CT/SKOS concepts. The interface is divided into several sections:

- AIDA Search:** This section contains a search bar with the text "Med_line" and a dropdown menu set to "content". Below the search bar, there are several search results, each with a title, a brief description, and a "cached" link. The results include:
 - The *tigA* gene is a transcriptional fusion of glycolytic genes encoding triose-phosphate isomerase and glyceraldehyde-3-phosphate dehydrogenase in oomycota.** of the order Pythiales in the phylum Oomycota. This is the first demonstration of glycolytic gene fusion... (Oomycota), as well as more distantly related genera, Saprolegnia and Achlya, in the order Saprolegniales. 9352934 [cached](#)
 - Interspecific hybridization in plant-associated fungi and oomycetes: a review.** Fungi (kingdom Mycota) and oomycetes (kingdom Stramenopila, phylum Oomycota) are crucially... herbivores. Other fungi and many oomycetes are plant pathogens that devastate natural and agricultural. 14629368 [cached](#)
 - Genetic code and phylogenetic origin of oomycetous mitochondria.** mitochondria of Phytophthora parasitica (phylum Oomycota, kingdom Protocista). Comparison... that mitochondria of Oomycota share a direct common ancestor with mitochondria of plants and that mitochondria. 1598508 [cached](#)
 - Phytophthora ramorum: a pathogen with a remarkably wide host range causing sudden oak death on oaks and ramorum blight on woody ornamentals.** : Phytophthora ramorum (Weres, De Cock & Man in't Veld): kingdom Stramenopila; phylum Oomycota, class Peronosporomycetidae; order Pythiales; family Pythiaceae; genus Phytophthora. Host range: The host range. 19019002 [cached](#)
 - Occupational sensitization to Plasmopara viticola.** Molds of the class Oomycetes are of allergologic importance in special cases. However, probes... for the second time a sensitization to Oomycetes. 7509819 [cached](#)
 - A Galpha subunit controls zoospore motility and virulence in the potato late blight pathogen Phytophthora infestans.** infestans belongs to the class oomycetes, a group of organisms in which signal transduction. 14763970 [cached](#)
 - Microbody-like organelles as taxonomic markers among Oomycetes.** criterion by which the phylogeny of these fungi can be evaluated. In zoospores of Oomycetes contain a variety of microbody-like organelles with highly structured. 3910137 [cached](#)
 - Infections in E-beta thalassemia.** insidiosum, is fungus-like, in the kingdom Stramenopila, and in the class Oomycetes. The mortality rate. 11132234 [cached](#)
 - A transmembrane phospholipase D in Phytophthora: a novel PLD subfamily.** PLD subfamily that we identified in Phytophthora, a genus belonging to the class oomycetes. 15826868 [cached](#)
 - The properties and localization of Saprolegnia monoica chitin synthase differ from those of other fungi.** The presence of non-fibrillar alpha-chitin in cellulosic fungi (class Oomycetes) poses intriguing questions as to its role, subcellular localization and evolutionary significance. Previous studies. 9245828 [cached](#)
- AIDA Thesaurus Query Builder:** This section shows a hierarchical tree structure for a query. The root is "My Query", which branches into "Phylum Oomycota", "Class Oomycetes", "Order Saprolegniales", and "Genus Branchiomyces". Under "Genus Branchiomyces", there is a sub-entry for "Branchiomyces demigrans".
- AIDA Thesaurus browser:** This section has a search bar with the text "Enter a concept to search for" and buttons for "Refresh" and "Clear". Below the search bar is a tree structure of concepts. The tree starts with "Linkage concept", which branches into "Observable entity" and "Organism". "Organism" further branches into "Animal", "Infectious agent", "Kingdom Animalia", "Kingdom Chromista", "Phylum Oomycota", "Kingdom Plantae", "Life-cycle form", "Microorganism", "Pathogenic organism", "Renotrophic organism", "Trophic life form", "Unknown living organism", and "Pharmaceutical / biologic product".
- Sesame server:** This section has two tabs: "Server Details" and "Repositories". The "Repositories" tab is active, showing a dropdown menu labeled "Repositories:" with "SCT" selected.

Browser tabs: AIDA Search, Virtuoso SPARQL Query Form

Address bar: <http://ws.adaptivedisclosure.org/search/>

AIDA Search

Search: MedLine content Type in a query...

Growth hormone response to the GABA-B agonist baclofen in 3-week abstinent alcoholics.
-B receptor activity in 3-week abstinent alcoholics using the growth hormone (GH) response to baclofen, a **GABA-B receptor** agonist. The study aimed to investigate the relationship between **GABA-B**
 18047908 [cached](#)

Effects of stress and tranylcypromine on amphetamine-induced locomotor activity and GABA(B) receptor function in rat brain.
 indicate that TCP treatment, but not stress, increases **GABA(B) receptor activity** in the cerebral cortex... **GABA(B) receptor activity**, which may contribute to the antidepressant response to TCP.
 12495786 [cached](#)

Heterodimerization of GABA(B) receptor heterodimers is required for their function in the rat brain.
 GABA(B) receptor heterodimers are formed by the association of **GABA(B) receptor** subunits.
 12217645 [cached](#)

Recognition molecule for GABA(B) receptor activity in the rat brain.
 release in perisomatic inhibitory synapses by suppression of postsynaptic currents in the rat hippocampal CA1 pyramidal cell.
 14572452 [cached](#)

Differential GABA(B) receptor activity in the rat brain.
 to differences in the extent of presynaptic and postsynaptic inhibition.
 15615867 [cached](#)

Amitriptyline prevents GABA(B) receptor activity in the rat brain.
 model of neuropathic pain to maintain spinal cord GABA(B) receptor activity.
 16293232 [cached](#)

Ethanol potentiation of GABA(B) receptor activity in the rat brain.
 GABA(B) receptor agonist baclofen.
 15564584 [cached](#)

Suppression of hindlimb single-cell responses to GABA(B) receptor activity in the rat brain.
 (A) and GABA(B) receptor activity made from normal rats from the same strain.
 10848556 [cached](#)

Disinhibition of spinal GABA(A) or GABA(B) receptor activity in the rat brain.
 on NMDA and metabotropic glutamate receptor activity.
 16412481 [cached](#)

Group I mGlu receptors potentiate synaptosomal (3H)glutamate release independently of exogenously applied arachidonic acid.

AIDA Repository Query Window

GO Run query (ctrl+enter)

```

prefix go: <http://purl.org/obo/owl/GO#>
prefix owl: <http://www.w3.org/2002/07/owl#>
prefix obo: <http://www.geneontology.org/formats/oboInOwl#>
prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#>
select distinct ?name ?class ?definition
where
{
  <http://purl.org/obo/owl/GO#GO_0004965> rdfs:subClassOf ?class.
  ?class rdfs:label ?name.
  ?class obo:hasDefinition ?def.
  ?def rdfs:label ?definition
}

```

Results

name	class	definition
molecular_function	http://purl.org/obo/owl/GO#GO_0003...	Elemental activities, such as catalysi...
signal transducer activity	http://purl.org/obo/owl/GO#GO_0004...	Mediates the transfer of a signal fro...
receptor activity	http://purl.org/obo/owl/GO#GO_0004...	Combining with an extracellular or int...
receptor activity	http://purl.org/obo/owl/GO#GO_0004...	Combining with an extracellular or int...
transmembrane receptor activity	http://purl.org/obo/owl/GO#GO_0004...	Combining with an extracellular or int...
G-protein coupled receptor activity	http://purl.org/obo/owl/GO#GO_0004...	A receptor that binds an extracellular...
glutamate receptor activity	http://purl.org/obo/owl/GO#GO_0008...	Combining with glutamate to initiate a ...

OK, execution time: 19.968 seconds Use proxy

AIDA Concept Query Builder

My Query

- GABA-B receptor activity - <http://purl.org/obo/owl/GO#GO_0004965>

AIDA Thesaurus Browser

SCT-RDFS | senselab | MeSH | GO

GABA-B receptor activity

- GO
- GABA-B receptor activity - <http://purl.org/obo/owl/GO#GO_0004965>

AIDA Thesaurus Server

Server information

Server:

Username:

Password:

Repositories

Repository:

Page 1 of 2 Results 1 - 10 of 13 for ("GABA-B receptor activity")

Access to triples in Taverna via AIDA plugin

Taverna Workbench 2.1-SNAPSHOT 20090506

File Edit Workflows Advanced Help

Design AID Plugin Results

AIDA Browse & Search AIDA Query

AIDA Concept Query Builder

AIDA Thesaurus Browser

BioAID_sandbox

tno BioAID BioAID_sandbox

Concept

- BMI1 homologous polycomb_NO_UNIPROT_ID_FOR_B...
- Bmi1 oncogene_NO_UNIPROT_ID_FOR_Bmi1 oncogene...
- BMI1 proteins_NO_UNIPROT_ID_FOR_BMI1 proteins...
- BMI1_OneOf_P25916_Q5U0M5
- Bmi1_OneOf_P25916_Q5U0M5
- Brm1 ATPase_NO_UNIPROT_ID_FOR_Brm1 ATPase...
- Brm1_NO_UNIPROT_ID_FOR_Brm1
- c-fos gene promoter_NO_UNIPROT_ID_FOR_c-fos ge...
- c-fos gene_NO_UNIPROT_ID_FOR_c-fos gene
- c-fos mRNA_NO_UNIPROT_ID_FOR_c-fos mRNA
- c-fos promoter_NO_UNIPROT_ID_FOR_c-fos promoter
- c-fos_OneOf_P01101_P12841_Q5G6W2_Q5G6W3_Q...
- C-Myb proteins_NO_UNIPROT_ID_FOR_C-Myb proteins
- C-Myb_OneOf_P06876_P10242
- c-Myc promoter_NO_UNIPROT_ID_FOR_c-Myc promoter
- c-myc proto-oncogene_NO_UNIPROT_ID_FOR_c-myc ...
- c-Myc reduces_NO_UNIPROT_ID_FOR_c-Myc reduces

AIDA Search Concept

Search: My Experiment content bioaid

Title	Excerpt
BioAID_ProteinToDiseases_(75).txt	BioAID_ProteinToDiseases_(75) This workflow was based on
BioAID_DiseaseDiscovery_(72).txt	BioAID_DiseaseDiscovery_(72) This workflow finds disease relevant to the query
BioAID_Discover_proteins_from_text_plus_synonyms...	BioAID_Discover_proteins_from_text_plus_synonyms_(81) This workflow discovers proteins from
BioAID_DiseaseDiscovery_count_(91).txt	BioAID_DiseaseDiscovery_count_(91) This adds counting to BioAID_DiseaseDiscovery ,
BioAID_protein_discovery_(74).txt	BioAID_protein_discovery_(74) This workflow finds proteins relevant to the query
DiscoverProteinLink_(31).txt	://rdf.adaptivedisclosure.org/~marco/BioAID/Public/Workflows/BioAID/SwansonProteins.xml
BioAID_DiseaseDiscovery_byHumanUniprot_(110).txt	BioAID_DiseaseDiscovery_byHumanUniprot_(110) This workflow finds disease relevant to of Genetics_director Hideaki Sugawara (see

Page 0 of 0

Query string : *bioaid*

AIDA Thesaurus Server

Original document

Accomplishments

Demonstrations:

- http://hcls.deri.org/hcls_demo.html
- Demonstrator of querying across heterogeneous EHR systems
 - <http://hcls.deri.org/coi/demo/>
- <http://www.w3.org/2009/08/7tmdemo>
- <http://ws.adaptivedisclosure.org/search>
- HCLS KB hosted at 2 institutes
- Linked Open Data contributions

Interest Group Notes:

- HCLS KB
- Integration of SWAN and SIOC ontologies for Scientific Discourse
 - SWAN
 - SIOC
 - SWAN-SIOC

Technologies: <http://sourceforge.net/projects/swobjects/>

Accomplishments II

- **Conference Presentations:**
 - Bio-IT World, WWW, ISMB, AMIA, etc.
- **(Co)Organized Workshops:**
 - C-SHALS, SWASD, SWAT4LS 2009, IEEE Workshop
- **Publications:**
 - Proceedings of LOD Workshop at WWW 2009: Enabling Tailored Therapeutics with Linked Data
 - Proceedings of the ICBO: Pharma Ontology: Creating a Patient-Centric Ontology for Translational Medicine
 - AMIA Spring Symposium: Clinical Observations Interoperability: A Semantic Web Approach
 - BMC Bioinformatics. A Journey to Semantic Web Query Federation in Life Sciences
 - Briefings in Bioinformatics. Life sciences on the Semantic Web: The Neurocommons and Beyond

We've come a long way

- Triplestores have gone from millions to billions
- Linked Open Data cloud
- <http://lod.openlinksw.com/>
- On demand Knowledge Bases: Amazon's EC2
- Terminologies: SNOMED-CT, MeSH, UMLS, ..
- Neurocommons, Flyweb, Biogateway, Bio2RDF, Linked Life Data, ..
- <https://wiki.nbic.nl/index.php/BioWiseInformationManagement2009>

Penetration of ontology in biomedicine

- OBO Foundry - <http://www.obofoundry.org>
- BioPortal - <http://bioportal.bioontology.org>
- National Centers for Biomedical Computing
<http://www.ncbcs.org/>
- Shared Names <http://sharednames.org>
- Concept Web Alliance
<http://conceptweblog.wordpress.com/conferences/>
- Semantic Web Interest Group PRISM Forum
<http://www.prismforum.org/>
- Work packages in ELIXIR <http://www.elixir-europe.org/>

HCLS operations: How does it scale?

How many tasks can we handle? Global reach?

Limiting factors:

Time

- Time for HCLS work for participants
- Time slots for teleconferencing
 - Including participants in Asia is a challenge
- Organizational and communication overhead

Money

- Become a member
- Apply for a grant for HCLS work

Translating across domains

- **Translational medicine – use cases that cross domains**
- **Link across domains and research:**
 - **What are the links?**
 - **gene – transcription factor – protein**
 - **pathway – molecular interaction – chemical compound**
 - **drug – drug side effect – chemical compound**

But also:

- **Link discourse to raw data**

Mememes

- **Joining forces – NCBO, CWA, NIF, EBI, ..**
- **Synergy through Services**
 - SPARQL endpoints
- **Data Stewardship**

Synergy through Services

- **AIDA – remote collaboration simplified**
[image]
- **ISATools** [image]
- **NIF** [image]
- **HCLS with NCBO**
- ...

A SPARQL endpoint on every 'table'

- **Expose knowledge as OWL and RDF for all important data**
- **Example: SPARQL endpoint for**
 - **Uniprot (RDF)**
 - **SWAN (SWAN/SIOC RDF)**
 - **myExperiment (SWAN/SIOC RDF)**
- **Enables us to link workflows stored in myExperiment that are related by a common protein family to discussion forum postings (evidence)**

Pooling resources - collaborative environments

- **Wiki is becoming something more than community edited web pages**
- **Semantic Wiki has the potential to become both:**
 - 1. An interface to knowledge bases**
 - **Templates that generate a view for a particular record – See Wiki Professional**
 - 2. A source of information to be added to knowledge bases – SWAN/SIOC endpoints**
 - **On such a Semantic Wiki, each resource can be cited as a form of support for an assertion**

Use case scenario – Semantic Wiki

- 1. User has posted about Drug A side effect**
- 2. Side effect similarity with Drug B theory is boosted by 1**
- 3. Additional pathway for Drug A theory is boosted by 2**

What do we need?

- **New attitudes towards data – Data Stewardship**
- **Identifiers – people (authors, patients), diseases, drugs, compounds - preferably SharedNames**
- **Scalable triplestores**
- **Lightweight and ‘incomplete’ reasoning**
- **Coordination and cooperation across groups**

