Semantic Web Technologies

Lecture 8: SWT for the Life Sciences 1: Background and data integration

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15 December 2009

Outline

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Ontologies to solve real problems
- 'Historical' overview from GO to OBO Foundry
- Late early adopters

Linking Data
- Data integration strategies
- Linked data using ontologies

Linking technologies
- Preliminary points
- SWT for SWLS

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Early bioinformatics

- Advances in technologies to sequence genomes in the late ’80s-early’90s, as well as more technologies for proteins
- Need to store the data: in databases (’90s)
- Several ‘model organism’ databases with genes (and genomes) of the fruitfly, yeast, mouse, a flowering plant, flatworm, zebrafish
- Compare genes and genomes
  - One observation (of many): About 12% (some 18,000) of the worm genes encode proteins whose biological roles could be inferred from their similarity to their (putative) orthologues in yeast, comprising about 27% of the yeast genes (about 5,700)
  - *What else can we infer from comparing genes and genomes (across species)?*
  - *What about the possibility of automated transfer of biological annotations from the model organisms to less ‘fancy’ organisms based on gene and protein sequence similarity, to use to improve human health or agriculture?*

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Scope and requirements

- Need: a mainly computational system for comparing or transferring annotation among different species
- Methods for sequence comparison existed
- Main requirements:
    - One needs a shared, controlled, vocabulary for annotation of the gene products, the location where they are active, the function they perform
    - To take on board and be compatible with existing terminologies, like gene and protein keyword databases such as UniProt, GenBank, Pfam, ENZYME etc.
    - Database interoperability among, at least, the model organism databases
    - Organize, describe, query and visualize biological knowledge at vastly different stages of completeness
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How to meet such requirements?

- Two main strands in activities:
  - Very early adopters from late 1990s (by sub-cellular bio), i.e., starting *without* Semantic Web Technologies
  - Early adopters from mid 2000s (e.g., eco), starting *with* Semantic Web Technologies
- The Gene Ontology Consortium
  - Initiated by fly, yeast and mouse database curators[1] and others came on board (see `http://ww.geneontology.org` for a full list)
  - In the beginning, there was the flat file format .obo to store the ontologies, definitions of terms and gene associations
  - Several techniques on offer for data(base) integration that could be experimented with

[1] more precisely: FlyBase (http://www.flybase.bio.indiana.edu), Berkeley Drosophila Genome Project (http://fruitfly.bdgp.berkeley.edu), Saccharomyces Genome Database (http://genome-www.stanford.edu), and Mouse Genome Database and Gene Expression Database (http://www.informatics.jax.org).

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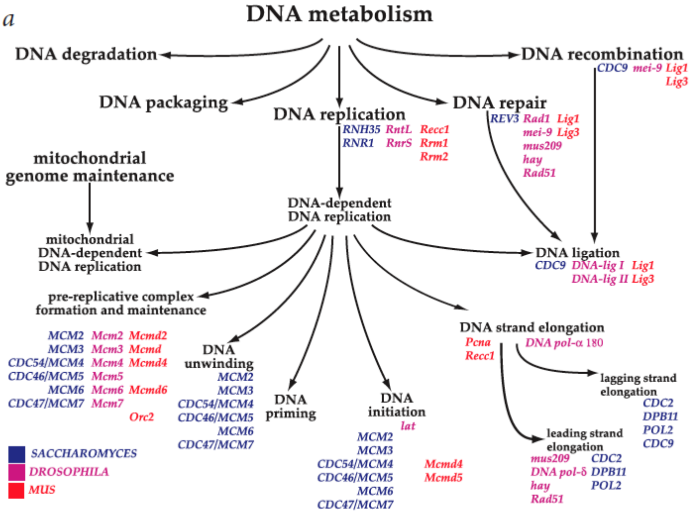
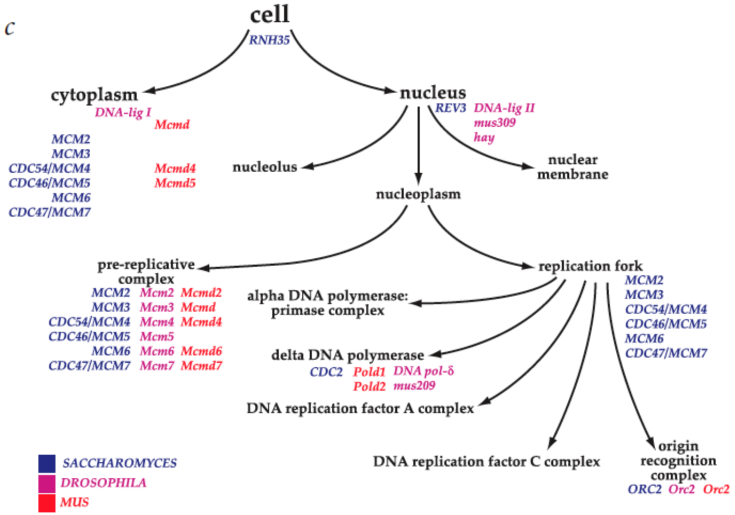


from GOC, 2000

GO contents example (cellular component)



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    - annotate genes (semi-automatically)
    - link the three GO ontologies
    - mine the literature (NLP)
- Content development: more in the GO, extensions to the GO (e.g., rice traits), copy of the principle to other subject domains (e.g., zebrafish anatomy)
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- Problems:
  - one can infer very little knowledge from the obo-based bio-ontologies (mainly where there are errors, but not *new* insights)—but note that that was not it's original aim
  - semantics of the relations overloaded
  - mushrooming of obo-based bio-ontologies by different communities, which makes interoperation of the ontologies difficult
  - greater needs for collaborative ontology development, maintenance, etc.
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  - open,
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  - same syntax,
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- ... to one for the **O**pen **B**iomedical **O**ntologies:
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  - usage of common relations that are unambiguously defined (*in casu*: the Relation Ontology)
  - provide procedures for user feedback and for identifying successive versions
  - has to have a clearly bounded subject-matter ("so that an ontology devoted to cell components, for example, should not include terms like 'database' or 'integer' " ...)

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- Sorting out the ontologies we have; e.g.,
  - harmonizing the four cell type ontologies into one (CL)
  - coordinating the anatomy ontologies of the various model organisms through a Common Aanatomy Reference Ontology
  - modularization of the subject domain by granularity, continuants, and occurents
- Adding ontologies to fill the gaps
- making OBO and OWL ontologies interoperable
- "Our long-term goal is that the data generated through biomedical research should form a single, consistent, cumulatively expanding and algorithmically tractable whole"
- "The result is an expanding family of ontologies designed to be interoperable and logically well formed and to incorporate accurate representations of biological reality"
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  - Biopax, who are now going into two directions: one as ontology-as-scientific-theory and another version as ontology-for-applications (see `http://www.biopax.org`)
  - protein phosphatases (Wolstencroft et al, 2007)
- Linking external data to the ontologies, e.g.:
  - HistOn ontology (in OWL) and an RDF triple store with Sesame (Marshall et al, 2006)
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- Aim also to do *automated reasoning* over it; e.g.:
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  - hypothesis testing
  - intelligent access to the data by using terms in the ontology instead of the gory details of the database
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Shopping for approaches to achieve data integration

I. Physical schema mappings
   - Global As View (GAV)
   - Local As View (LAV)
   - GLAV

II. Conceptual model-based data integration

III. Data federation

IV. Data warehouses

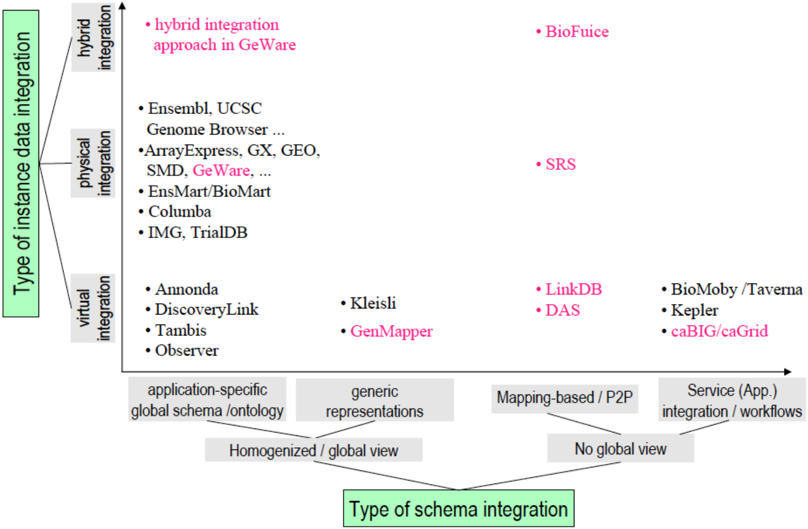
V. Data marts

VI. Services-mediated integration

VII. Peer-to-peer data integration

VIII. Ontology-based data integration
   - I or II (possibly in conjunction with the others) through an ontology
   - Linked data by means of an ontology

Classification of data integration approaches and tools



E. Rahm et al.: Data Integration in Bioinformatics and Life Sciences   EDBT summer school 2007   21

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- Ontology on top of conceptual data models
- Ontology to mediate between services
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Linked data in Bio

- *Data-level integration*
- Annotated instances stored in databases
- Across databases at physically different locations
- On the Web
- Where the ontology tells you which ones are the same, or instantiating the same universal represented in the ontology

Web-links based 'integration'

- Web-Link = URL of a source + ID of the object of interest
- Little integration effort, Scaleable, Navigational analysis: only *one object at a time*
- A mere link is semantics-poor w.r.t. language and subject domain meaning, e.g.:
  - How would one do automated reasoning with it to derive implicit knowledge? (not)
  - "related to" versus, among others, *partOf*, *isA*, *containedIn* etc; i.e., even poorer than the thesaurus' RT, BT, NT
- DBGET + LinkDB
- see also `http://www.genome.jp/dbget/`

Integration and annotations examples

- GenMapper
  - Centralised, with a global view
  - Exploits existing mappings between objects/sources
  - Links between the databases through the annotations of the objects (e.g., genes, proteins)
  - Links to terms of the ontology (GO), i.e., (semi-)*manual* classification
- Distributed Annotation Systems (DAS)
  - Distributed, mapping-based, no global view
  - Central genome server as primary source that contains the reference genome sequence
  - Separately, several annotation servers where the sources are wrapped
  - Recalculation of all annotations when the reference sequence has changed

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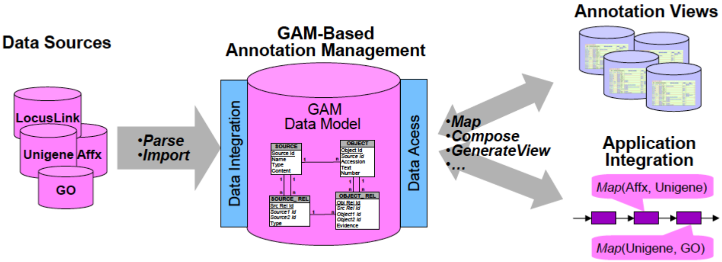

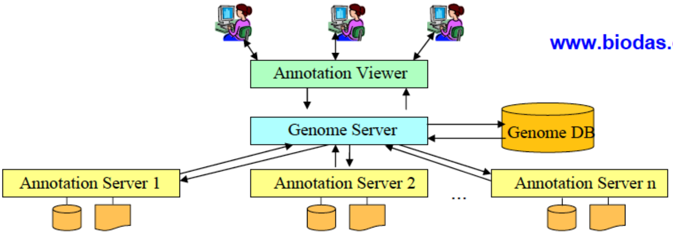

www.biodas.org

from Rahm et al, 2007

Other integration systems (examples)

- BioFuice, based on iFuice:
  - Use instance-level cross-references for instance-level mappings between sources
  - Mappings have a semantic mapping type
  - Domain model ($\pm$ an ontology) indicates available object types and relationships
- Sequence Retrieval System: wrapping sources, making them accessible through one interface
- BioGuide: selecting appropriate sources and tools using chosen preferences and strategy
- IMGT-Choreography based on the IMGT-ONTOLOGY concepts to coordinate services among databases
- Mash-ups, RDF, XML, ...

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- BioGuide: selecting appropriate sources and tools using chosen preferences and strategy
- IMGT-Choreography based on the IMGT-ONTOLOGY concepts to coordinate services among databases
- Mash-ups, RDF, XML, ...

Generalising the current bio-integration implementations

- Many CS theory and technologies 'on offer' that purport to solve each integration problem
- All of them experimented with by the users, who added linked data, annotations, and web-links to the array of options
- For all: still a lot of manual work
- For all: for various reasons fairly simple end-user level queries (which might well be complicated at the back-end)
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Ontologies to solve real problems
- 'Historical' overview from GO to OBO Foundry
- Late early adopters

Linking Data
- Data integration strategies
- Linked data using ontologies

Linking technologies
- Preliminary points
- SWT for SWLS

Outline

Background

- Main players in SWLS are engineers, domain experts, bioinformaticians, bio-ontologists. “Something bio” covers many disciplines: e.g., genomics, metabolomics, ecoinformatics, and, above all: biomed & healthcare. Diverse fields, diverse needs.
- Some current characteristics:
  - Collaboration & interdisciplinary work
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  - Novel-ness of the technologies: data integration techniques of the '90s did not solve the issues, SW tech will?
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Expressive ontology vs scalability & performance

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From bench to bedside — and from CS theory to software application

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- "A significant barrier to translational research is the lack of uniformly structured data across related biomedical domains."
- "Current tools and standards are already adequate to implement components of the bench-to-bedside vision."
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- Querying the data across databases
- Expressive ontology languages to represent biological knowledge
- Manage (query) the data silos ('write-only database')
- Building upon the web of data
- Automation to 'upgrade' 'legacy' material to SemWeb technologies and standards
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