Evaluation of Ontology Merging Tools in Bioinformatics

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What is an ontology?

- From GO website:
- Ontologies are 'specifications of a relational vocabulary'. In other words they are sets of defined terms like the sort that you would find in a dictionary, but the terms are networked. The terms in a given vocabulary are likely to be restricted to those used in a particular field, and in the case of GO, the terms are all biological.

- Why are ontologies important? Ontologies provide a vocabulary for representing and communicating knowledge about a topic, and a set of relationships that hold among the terms of the vocabulary. They can be structurally very complex, or relatively simple. Most importantly, ontologies capture domain knowledge in a way that can easily be dealt with by a computer .

Functions of bio-ontologies

- What are they used for? Enable knowledge sharing and reuse

- Importance of ontology merging? Need for humans and computers to find functionally equivalent terms among different vocabularies. To provide consistent descriptions of gene products, cellular signaling, biological processes, cellular components and molecular functions, in a species-independent manner, in different databases.

- This supports biological applications such as comparative genome analysis, browsing genes from different participating databases, knowledge extraction from texts (text mining), extracting biological insight from enormous sets of data (from genomic sequencing and microarray experiments), genome annotation

Test Ontologies

- Ontologies merged in study:
  - Gene Ontology (GO)-The Gene Ontology project provides a controlled vocabulary to describe gene and gene product attributes in any organism. The GO collaborators are developing three ontologies (describe biological processes, cellular components and molecular functions)
  - Signal Ontology (SO)--Ontology for the cell signaling system, includes both all the nomenclatures of signaling molecules as well as signaling reactions and all the relationships among the terms in the nomenclatures

Tools tested for merging ontologies

- Evaluated in study:
  - Protégé 2000 with PROMPT (plug-in, algorithm for merging and aligning ontologies)
    - Stanford Medical Informatics, free software
    - Goal: creating, editing, browsing ontologies, compatible with other systems for knowledge representation and extraction
    - How it works: continuously generates lists of suggested operations (and explains why made suggestion), determines conflicts, and proposing conflict-resolution strategies to guide user throughout the entire merging process
  - Chimaera
    - Knowledge Systems Laboratory at Stanford, free software
    - Goal: browsing, editing, diagnosing ontologies
    - How it works: name resolution lists--generates lists of terms that are good candidates for merging or for taxonomic relationships, and taxonomy resolution lists--suggests taxonomy areas for reorganization; user makes decisions from lists
  - Main difference: Chimaera-*Where* vs. Protégé-*What*

Protégé 2000 with PROMPT

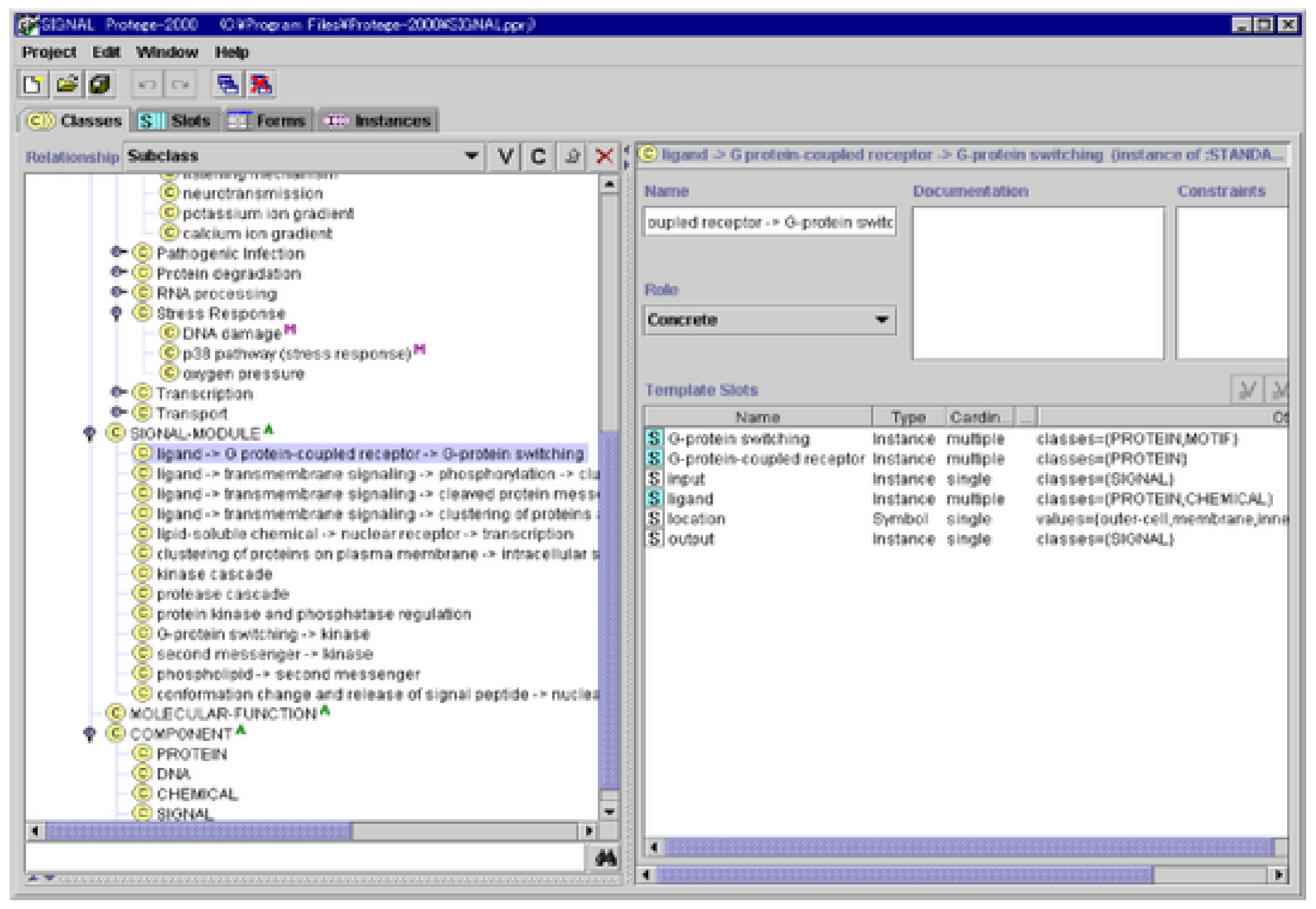
- List of Suggestions; After merging
- Identifies possible conflicts that could occur as result of merging and proposes possible solutions, based on similarities in concept and attribute names
- Concepts in original ontology that are not merged need to be copied into new ontology

Chimaera

- Merging ontologies
- Generates list of concepts and attributes that are candidates for merging-based on similarities in names, definitions, acronyms, name extensions, etc

SO implemented in Protégé 2000

Figure 1: A part of the class hierarchy of SIGNAL-ONTOLOGY. The main elements of the knowledge model are frames representing: classes, slots, forms, and instances. http://hc.ims.u-tokyo.ac.jp/JSBi/journal/GIW00/GIW00P101/index.html



Name: oupled receptor -> G-protein switc

Role: Concrete

Name	Type	Cardin...	Ot
G-protein switching	Instance	multiple	classes={PROTEIN,MOTIF}
G-protein-coupled receptor	Instance	multiple	classes={PROTEIN}
input	Instance	single	classes={SIGNAL}
ligand	Instance	multiple	classes={PROTEIN,CHEMICAL}
location	Symbol	single	values={outer-cell,membrane,inne
output	Instance	single	classes={SIGNAL}

Methods of tool evaluation

Research question:

- Which tool offers better support for merging ontologies?

Methods: Two ‘cases’ chosen from GO and SO:

  - *Behavior (60 (GO), 10 (SO))*
  - *Immune defense (70 (GO), 15 (SO))*

Methods:

2 types of evaluations

- Predefined criteria evaluated:
  - Partly based on literature studies
  - Investigated tools using GO and SO:
    - Looked at representation language tool uses, kind of ontologies that can be merged, assistance given to user, tool availability (stability over time)
  - Measured: Precison (relevance), Recall (total number of relevant suggestions system proposes), Time taken to merge ontologies

  - Critique:
    - Description was vague (i.e, kind of ontologies? Meaning domain specific, or structure specific?
    - Did not say which variables were evaluated from literature study

Methods of tool evaluation

- Evaluation of user interface
  - Experiment with 8 test users (4 computer scientists, 4 biologists with with no experience working with ontologies.)
  - **REAL approach (Relevance, Efficiency, Attitude, Learnability)**
  - **Users given information on concept of ontologies, did tutorial**
  - **Given tasks to perform; asked to think aloud**
  - **Filled in REAL questionnaire**
  - **(total time: 3-5 hours per person)**
- Critique: Did not provide enough background on REAL method or give examples of studies in which it was used to support statement that it “usually gives good results”

Results: precision and recall

Table 1. Quality of suggestions

Precision:

PROMPT had perfect precision for both cases

Chimaera- below 50% for both cases

Recall:

-Chimaera "over" suggested in both cases (out of 5 and 9 total possible cases for merging) provided more suggested terms (higher recall), but had less precision than Prompt.

Results: time

- Table 2: Time in minutes for merging
- Merging faster with Chimaera than PROMPT (so, better for larger ontologies)
- Calculated differently for each tool
- Question: explanation of no additional time for merging in Chimaera (t= 0 mins. in Table 2) with 1 missing suggestion in Table 1? (Time does work with case 2)

Results: User interface

- Evaluated via questionnaire (tables 3 and 4), and user’s observations while testing (think aloud method)
- REAL approach
  - Relevance (Were user’s needs satisfied?)
    - PROMPT was thought to be better.
    - Chimaera-had long response time for operations
  - Efficiency
    - PROMPT- better to use for specific operations, though merging required too much work—awkward to copy concepts not merged
      - Liked color representations of original ontologies
    - Chimaera-hard to see where in hierarchy concept was located
      - harder to find and choose operations

Results: User Interface

- Attitude,
  - PROMPT more fun to use; names of operations more self-explaining
  - Chimaera-boring, unclear
- Learning
  - Equally hard to learn to merge ontologies in both systems, hardest to learn in PROMPT was copying of concepts, etc not merged.
  - Chimaera—provided better help
- Critique: not clear on difference between some questions in questionnaire, did users understand what they were evaluating in each part of REAL?

Conclusions

- Critique of methods:
  - Clearly defined criteria before actual evaluation
  - Varied order in which tools were tested by users, so did not perform task better in second tool

Conclusions:

-No significant difference between biologists test users and users with high school knowledge of biology

- Critique: Any difference between computer scientists and biologists relating to their navigation of each tool?

Would have liked table evaluating interface, where participants grouped by profession (CS and biology)

Conclusions

- Both tools can model current bio-ontologies
- PROMPT: better user interface, easier to work with
- Chimaera: faster for merging ontologies, better functionality, provides better help
- Both tools need to increase the quality of suggestions
- Critique: provided 2 concrete results (easier interface, speed of merging)
    - PROMPT-early phases of ontology creation
    - Chimaera-analysis, maintenance, diagnosis (did results show?)
    - Would be good to see study that looks at which tools work best with merging specific ontologies; also study of merging of more than 2 ontologies; evaluations by more advanced users of ontologies