Introduction to the Gene Ontology

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EBI & GO Consortium
EMBRACE Workshop — Applied Gene Ontology
7–9 November 2007

Reasons for GO

- The problem: how to
  - ... communicate
  - ... represent biological knowledge in databases
  - ... query databases — find useful answers to biological questions like
    - What does the gene product do?
    - Where and when does it act?
    - Why does it perform these activities?

The Challenge of Language:

People call the same thing by different names

A Common Vocabulary:

GO uses one term and many characterized synonyms

Tactition

Taction

Tactile sense

perception of touch ; GO:0050975

GO project aims

- Compile structured vocabularies describing aspects of molecular biology
- Describe gene products using vocabulary terms (annotation)
- Provide public resource of data and tools:
    - to query and modify the vocabularies and annotations
    - annotation tools for curators

GO Scope: 3 domains

- Molecular Function — elemental activity or task
  - *nuclease, DNA binding, transcription factor*
- Biological Process — broad objective or goal
  - *mitosis, signal transduction, metabolism*
- Cellular Component — location or complex
  - *nucleus, ribosome, origin recognition complex*

✶ ‘Normal’ functions and processes only:
  - *No pathological processes*
  - *No experimental conditions*

GO term features

```
[Term]
id: GO:0046423
name: allene-oxide cyclase activity
namespace: molecular_function
def: "Catalysis of the reaction: (9Z)-(13S)-12,13-
epoxyoctadeca-9,11,15-trienoate = (15Z)-12-
oxophyto-10,15-dienoate." [EC:5.3.99.6]
synonym: "(9Z)-(13S)-12,13-epoxyoctadeca-9,11,15-
trienoate isomerase (cyclizing)" EXACT [EC:5.3.99.6]
xref: EC:5.3.99.6
xref: MetaCyc:ALLENE-OXIDE-CYCLASE-RXN
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is_a: GO:0016860 ! intramolecular oxidoreductase
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unique ID

name

namespace

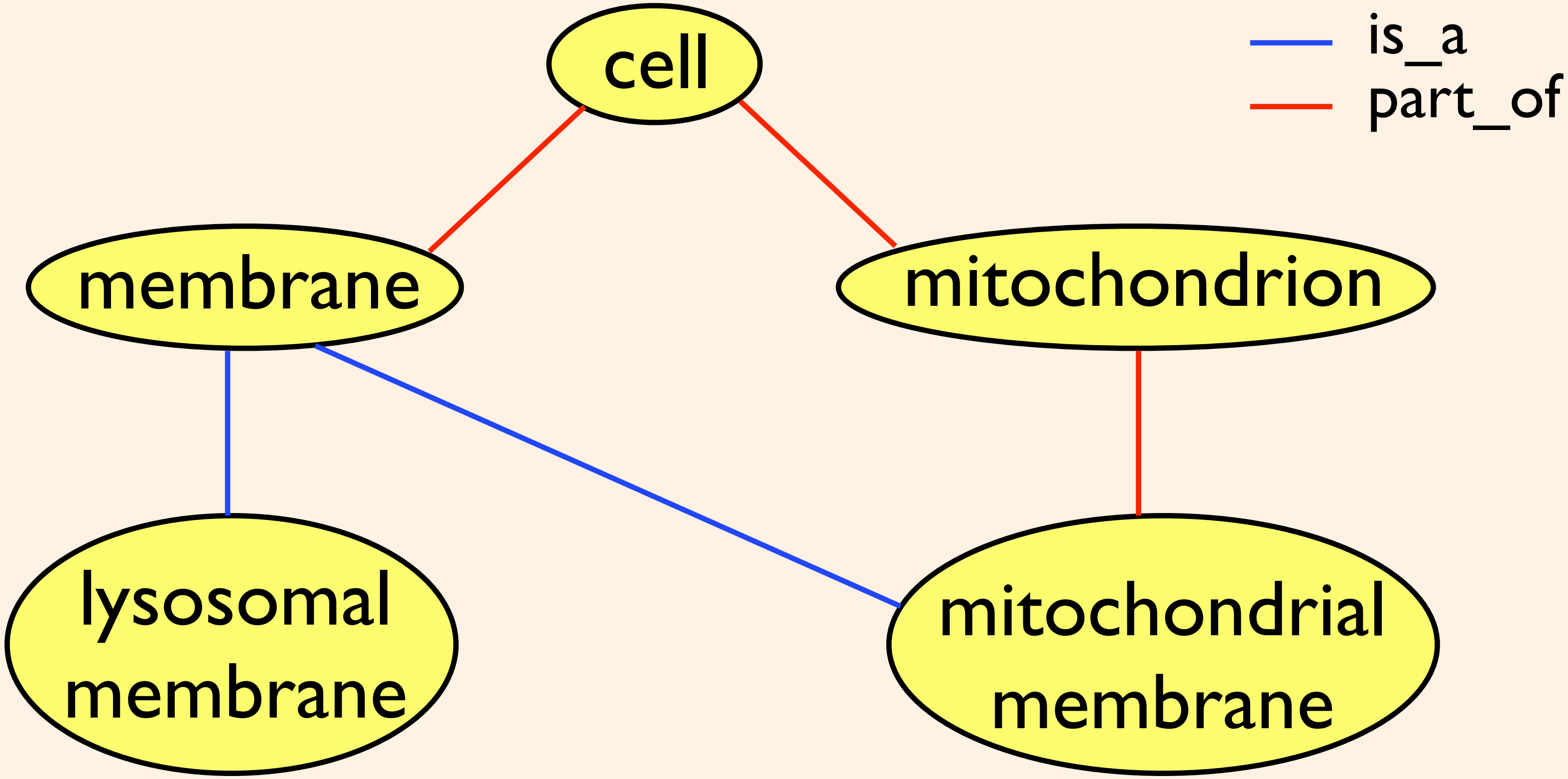
text definition

synonyms

cross-references

parents

GO Structure: DAG



is_a

part_of

cell

membrane

mitochondrion

lysosomal membrane

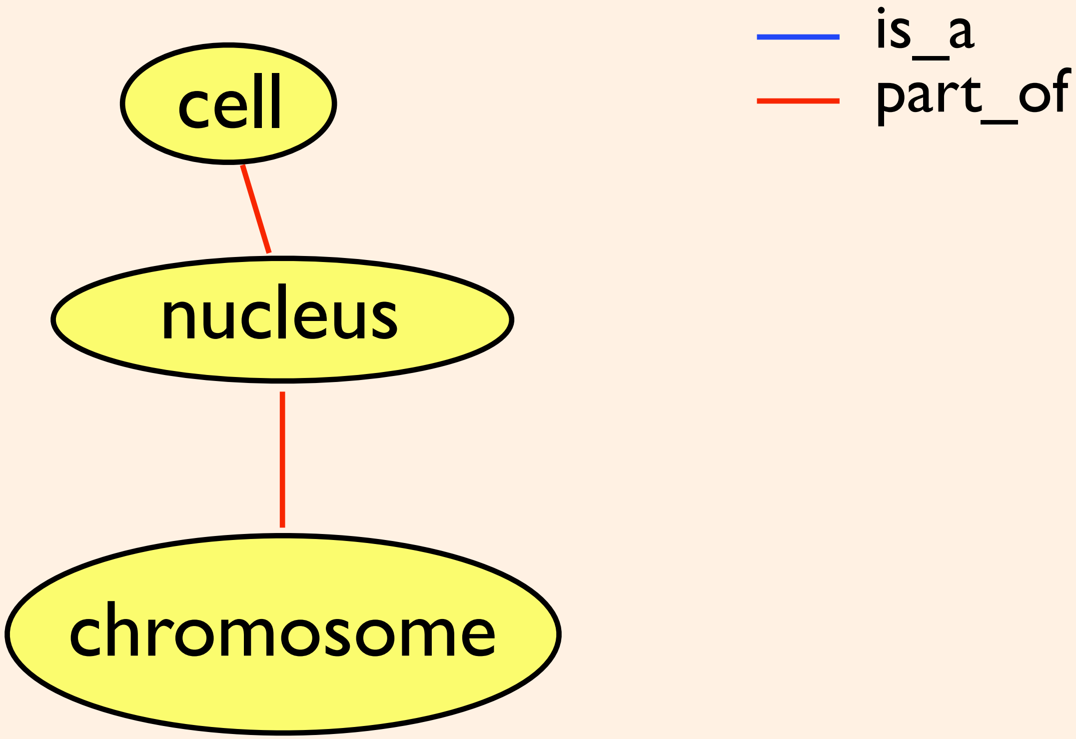
mitochondrial membrane

GO is a directed acyclic graph (DAG):
a term can have one or more parent(s)

GO relationships

- now in use:
  - is_a – subclass; a is a type of b
  - part_of
    - physical part of (component)
    - subprocess of (process)
- to be added:
  - regulates

True Path Rule



is_a

part_of

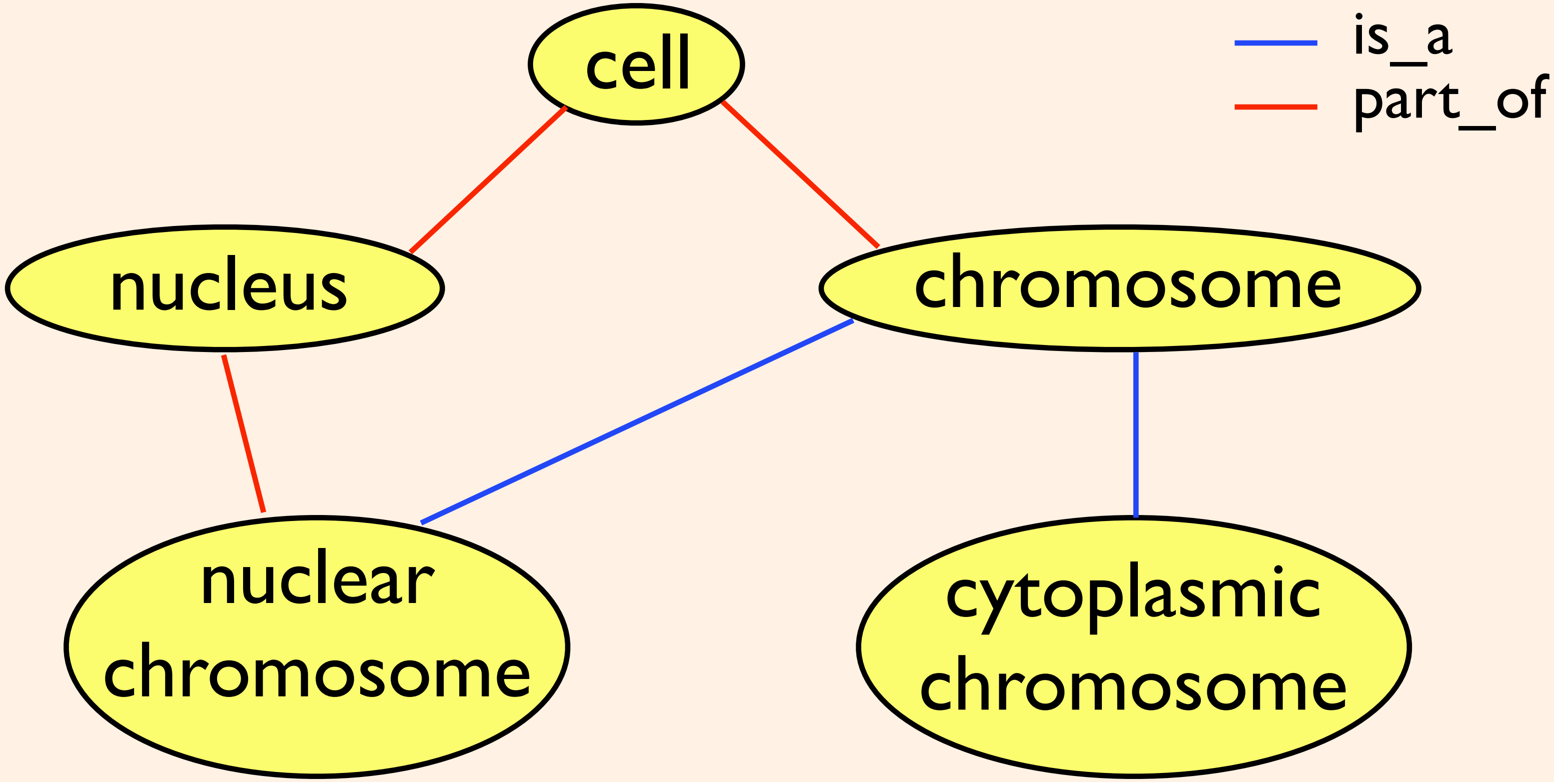
cell

nucleus

chromosome

But what about bacteria??

True Path Rule



Resolved structure

(some terms omitted for clarity)

Ontology Rules

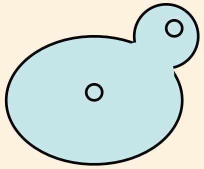
(examples)

- Univocity
  - A term (or relationship) should mean the same thing every time it’s used
- Positivity
  - Complements of classes are not themselves classes; i.e., terms such as ‘non-mammal’ or ‘non-membrane’ do not designate genuine classes
- Objectivity
  - Which classes exist does not depend on our biological knowledge; terms such as 'unknown', 'unclassified' or 'unlocalized' are thus unsuitable

The Challenge of Univocity:

People use the same words to describe different things

= bud initiation



= bud initiation

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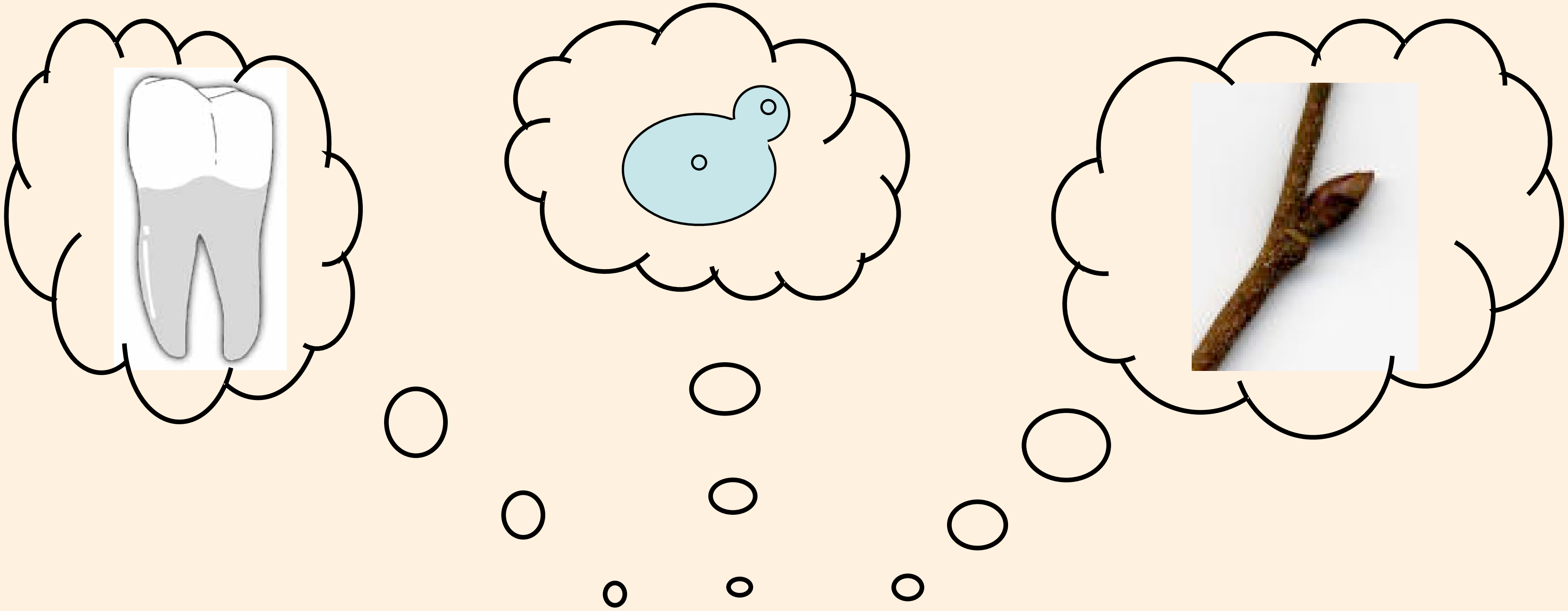
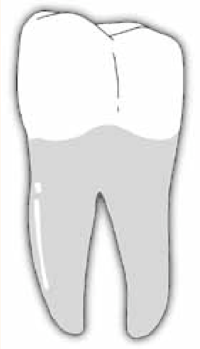


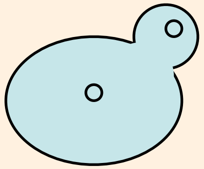
Bud initiation?
How is a computer
to know?

Univocity:

GO adds descriptors to discriminate among organisms



= tooth bud initiation

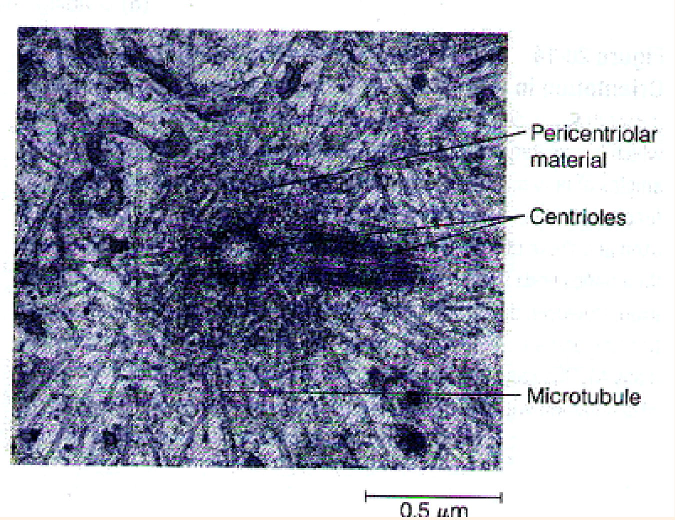


= cellular bud initiation

= leaf bud initiation

note: hypothetical examples

The Challenge of Positivity

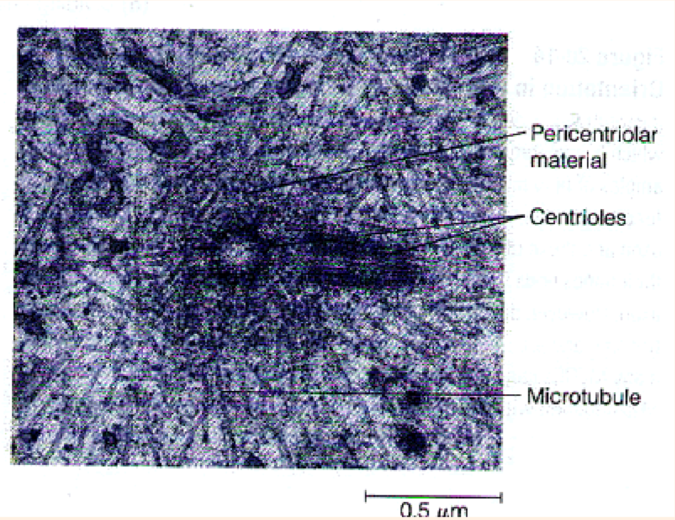


centriole

- Some organelles are membrane-bound.
- A centrosome is not a membrane bound organelle, but it still may be considered an organelle.

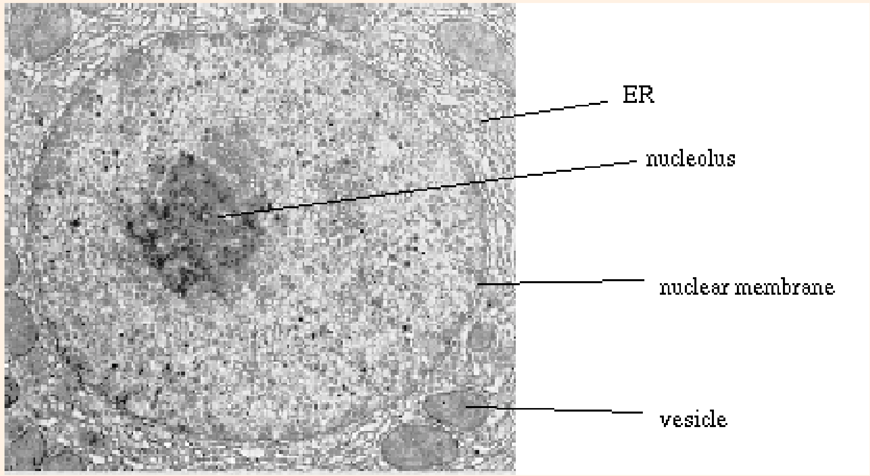
The Challenge of Positivity:

Sometimes absence is a distinction in a biologist’s mind



centriole

non-membrane-bound organelle GO:0043228



nucleus

membrane-bound organelle GO:0043227

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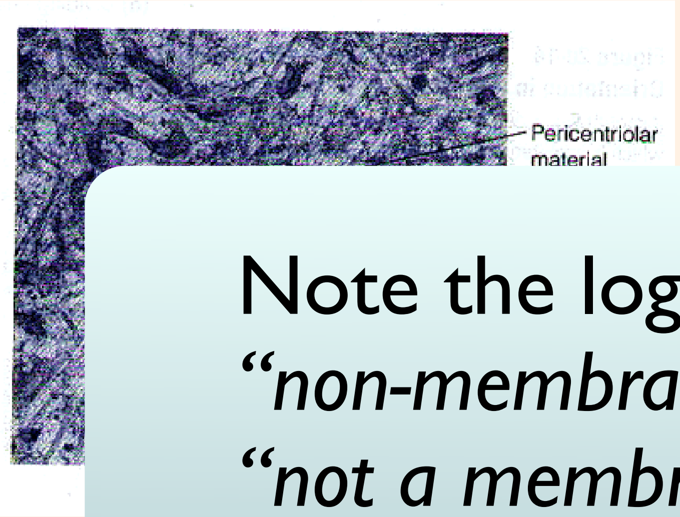
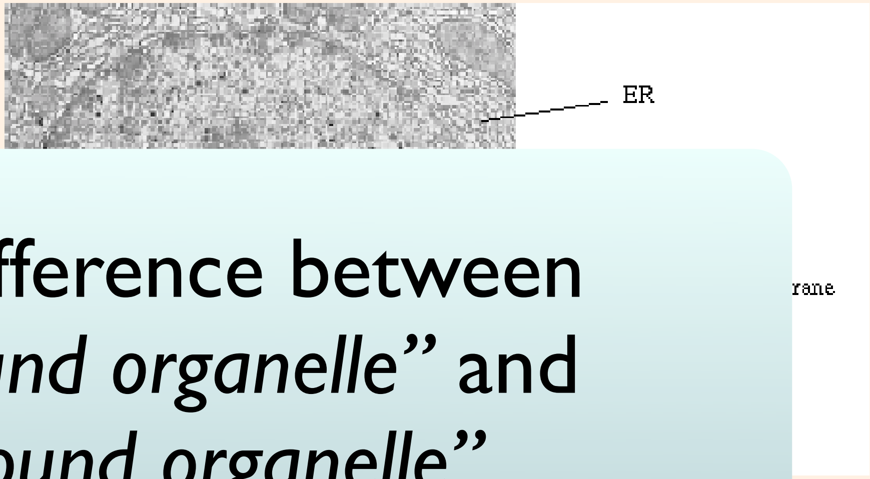




nor
org

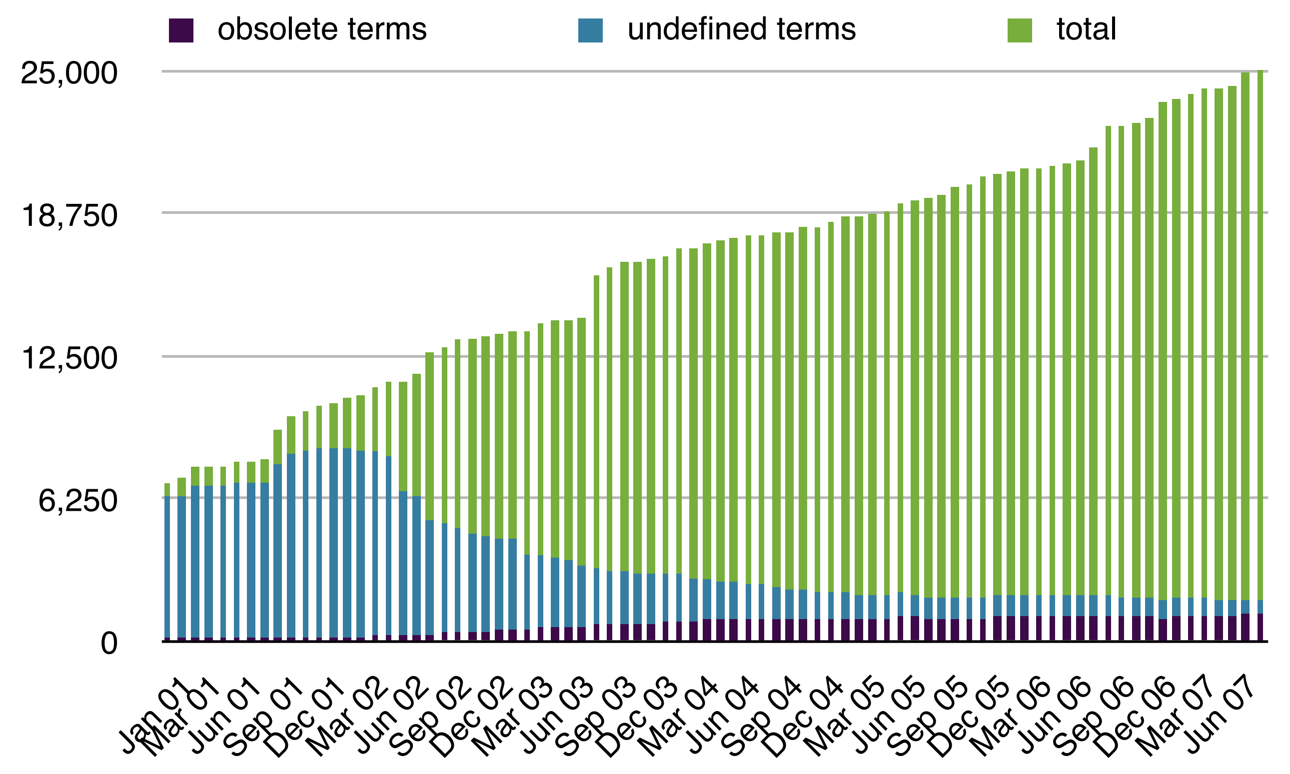
lle

Note the logical difference between "*non-membrane-bound organelle*" and "*not a membrane-bound organelle*"

The latter includes everything that is not a membrane bound organelle!

GO Term History

Jan. 2001–August 2007



Why GO Changes

- Advances in biology
- New groups join, requiring new terms or different relationships between terms
- Update legacy terms
- Improve logical consistency

What kinds of changes?

Small or large scale

Biological or logical/structural motivation

Small-scale Changes

- Individuals (e.g. annotators) request changes
  - Usually one or a few things at a time
  - New terms
  - Path corrections/additions
  - Improved definitions
- Closely coordinated with annotation

Large-scale Changes

- Improve ontology for a biological topic all at once
  - Motivation: improve usefulness for biologists, especially annotators
  - Usually connected with content meeting
  - Expert biologists involved
- Apply a logical/structural improvement to entire ontology at once
  - Motivation: improve consistency; enhance “computability”
  - Software developers involved

Applications of GO

- Gene product annotation
  - model organism databases
  - genome sequence analysis
- Expression data analysis
- Text mining
- More ...

GO Annotation

- What is GO annotation?
  - An annotation is a statement that a gene product …
    - … has a particular molecular function
    - … is involved in a particular biological process
    - … is located within a certain cellular component
  - … as determined by a particular method
  - … as described in a particular reference

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gene product

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GO terms

evidence

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High Conservation of the Set1/Rad6 Axis of Histone 3 Lysine 4 Methylation in Budding and Fission Yeasts*

Received for publication, September 18, 2002, and in revised form, December 17, 2002
Published, JBC Papers in Press, December 17, 2002, DOI 10.1074/jbc.M209562200

Assen Roguev‡§, Daniel Schaft‡§, Anna Shevchenko¶, Rein Aasland‖, Andrej Shevchenko¶, and A. Francis Stewart‡ ‡‡**

From ‡BIOTEC, Technische Universitaet Dresden, c/o Max Planck Institute for Molecular Cell Biology and Genetics and the ¶Max Planck Institute for Molecular Cell Biology and Genetics, Pfotenhauerstrasse 108, D-01307 Dresden, Germany and the ‖Department of Molecular Biology, University of Bergen, Thromoehlenstrasse 55, N-5020 Bergen, Norway

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Trithorax (4)), a well conserved 150-amino acid domain found in all eukaryotes, which mediates histone methyltransferase activity (5, 6). Trx and Ash1 both contain a SET domain. There is little evidence so far that Trx methylates histones; however, binding to histones has been documented (7). The third trxG3 member, Ash2, does not contain a SET domain but has a PHD finger (8) and a SPRY domain (9).

Although budding yeast does not have a Trx homologue, it has a protein, Set1, with a very similar type of SET domain (10). For this reason, we determined the composition of the Set1 complex, Set1C, and found that it has H3 Lys^4 methyltransferase activity *in vitro* (3). Concomitantly, two other groups have identified most members of Set1C (11, 12). The complete Set1C and the *in vitro* specificity for H3 Lys^4 meth-

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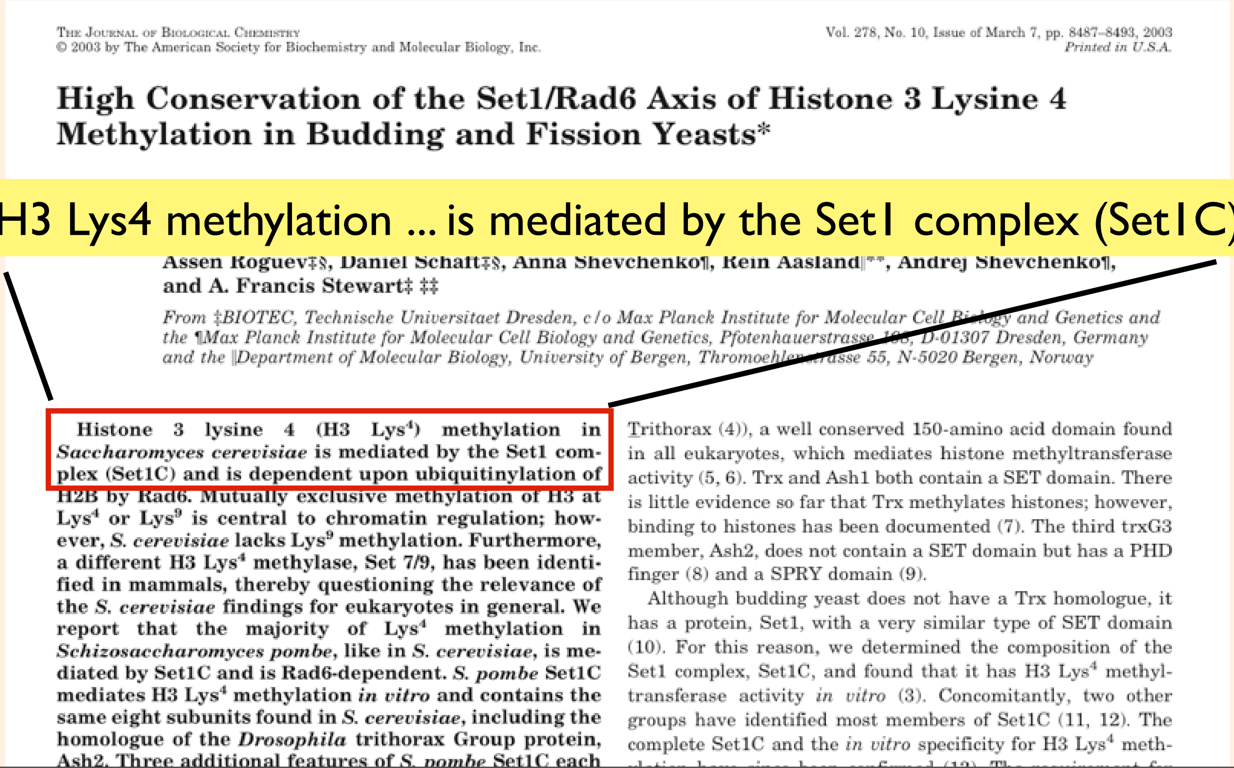

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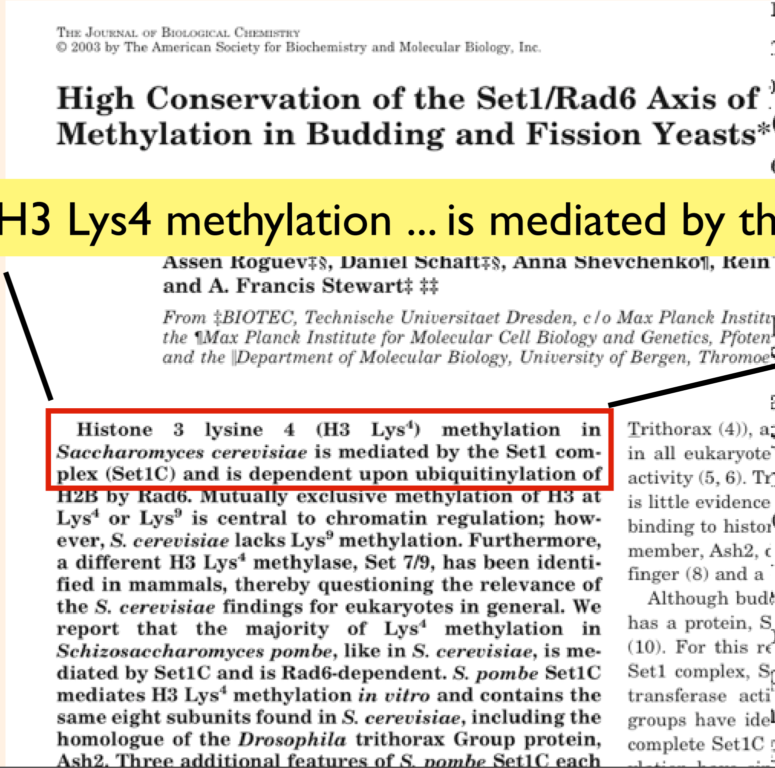

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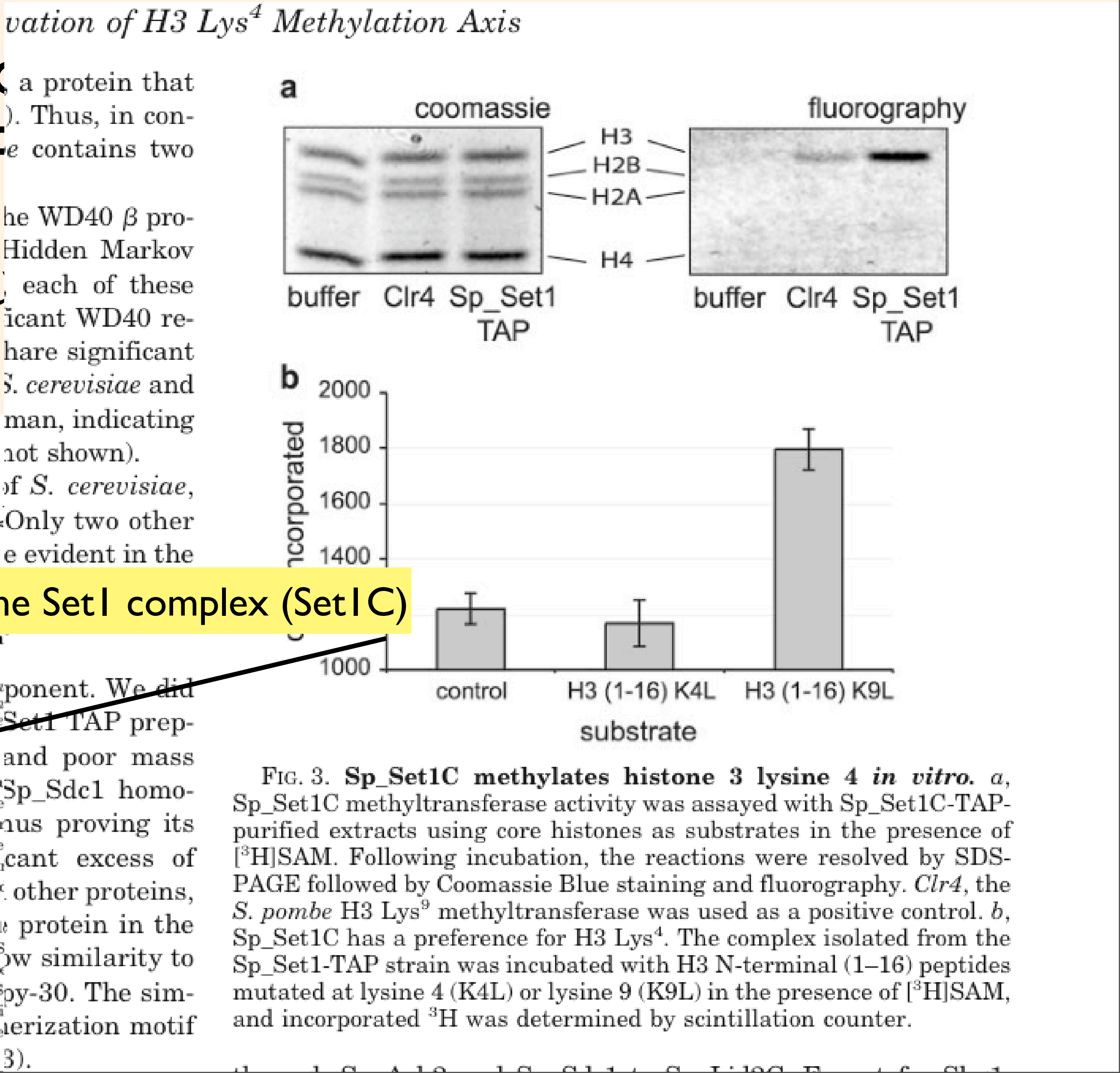

vation of H3 Lys⁴ Methylation Axis

a protein that
). Thus, in con-
e contains two

he WD40 β pro-
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GO & Text Mining

- GO provides sc
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- Text mining ma
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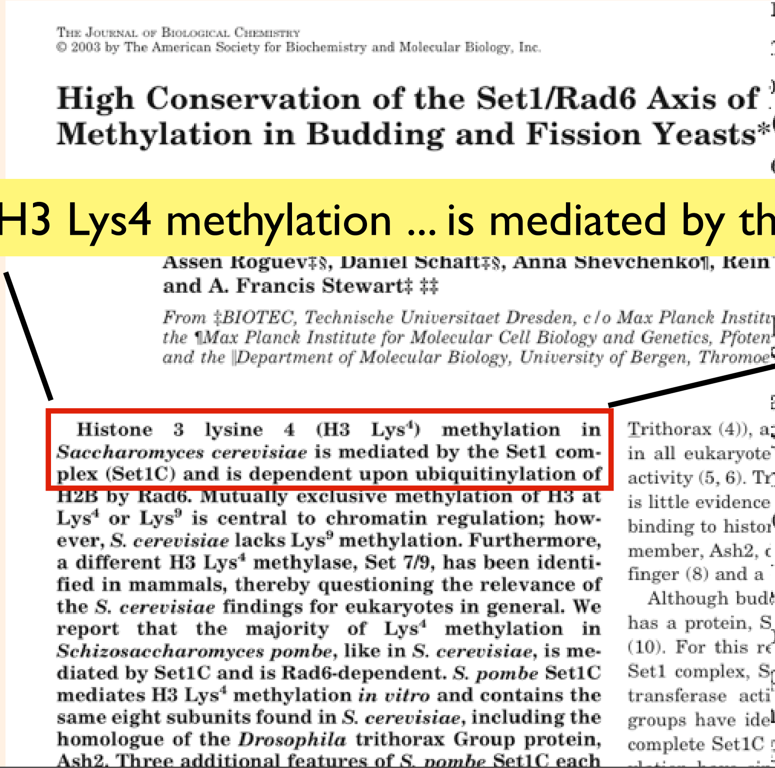

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High Conservation of the Set1/Rad6 Axis of
Methylation in Budding and Fission Yeasts*

Assen Roguev‡§, Daniel Schaft‡§, Anna Shevchenko¶, Rein
and A. Francis Stewart‡ ‡‡

From ‡BIOTEC, Technische Universitaet Dresden, c/o Max Planck Institu
the ¶Max Planck Institute for Molecular Cell Biology and Genetics, Pfoten
and the ‖Department of Molecular Biology, University of Bergen, Thromoe

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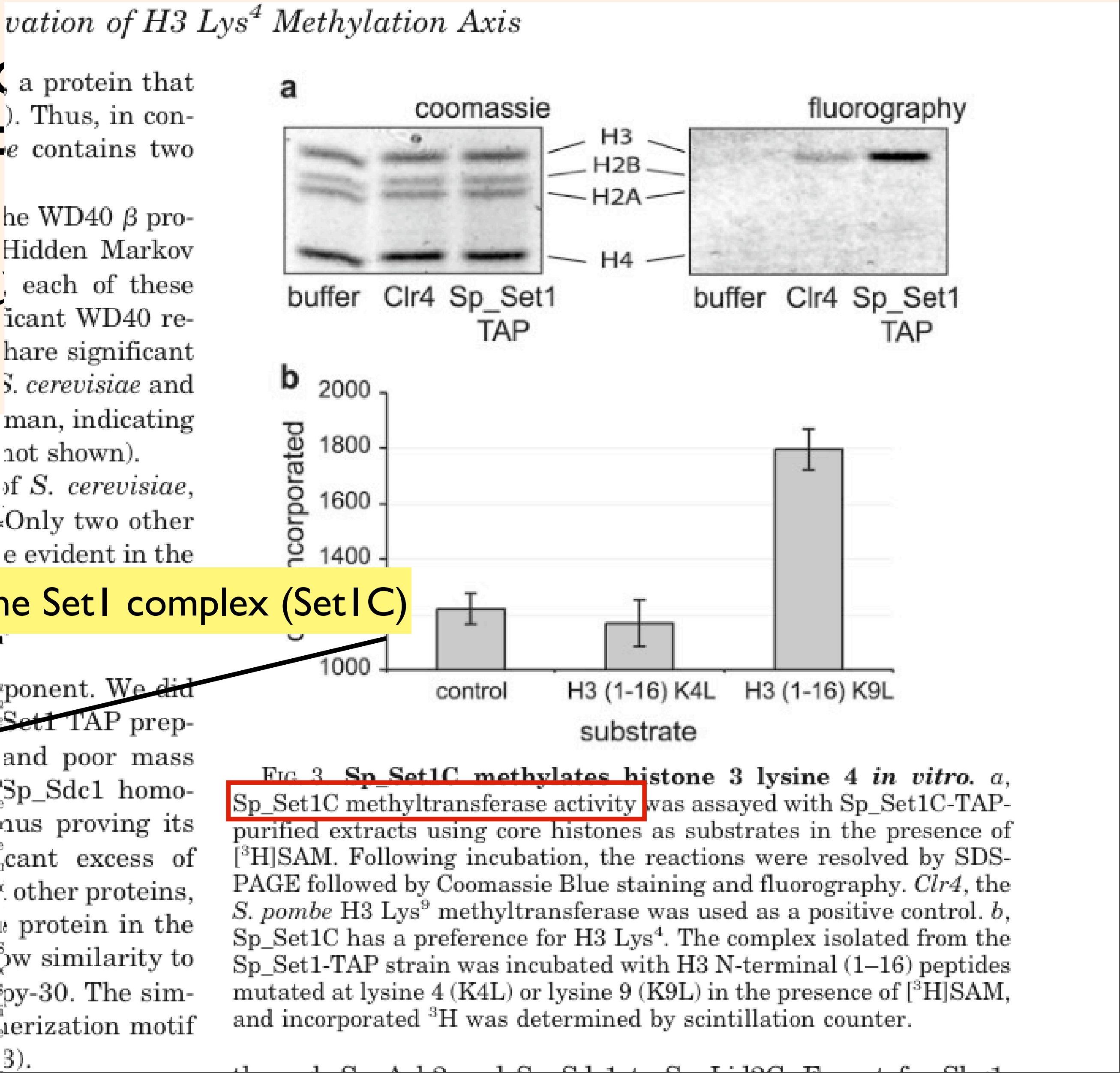

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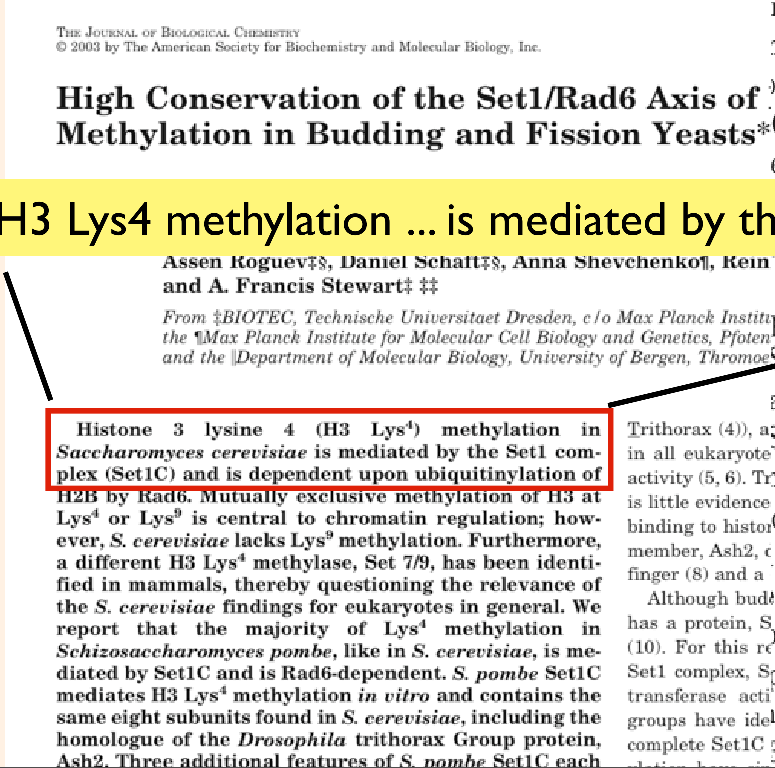

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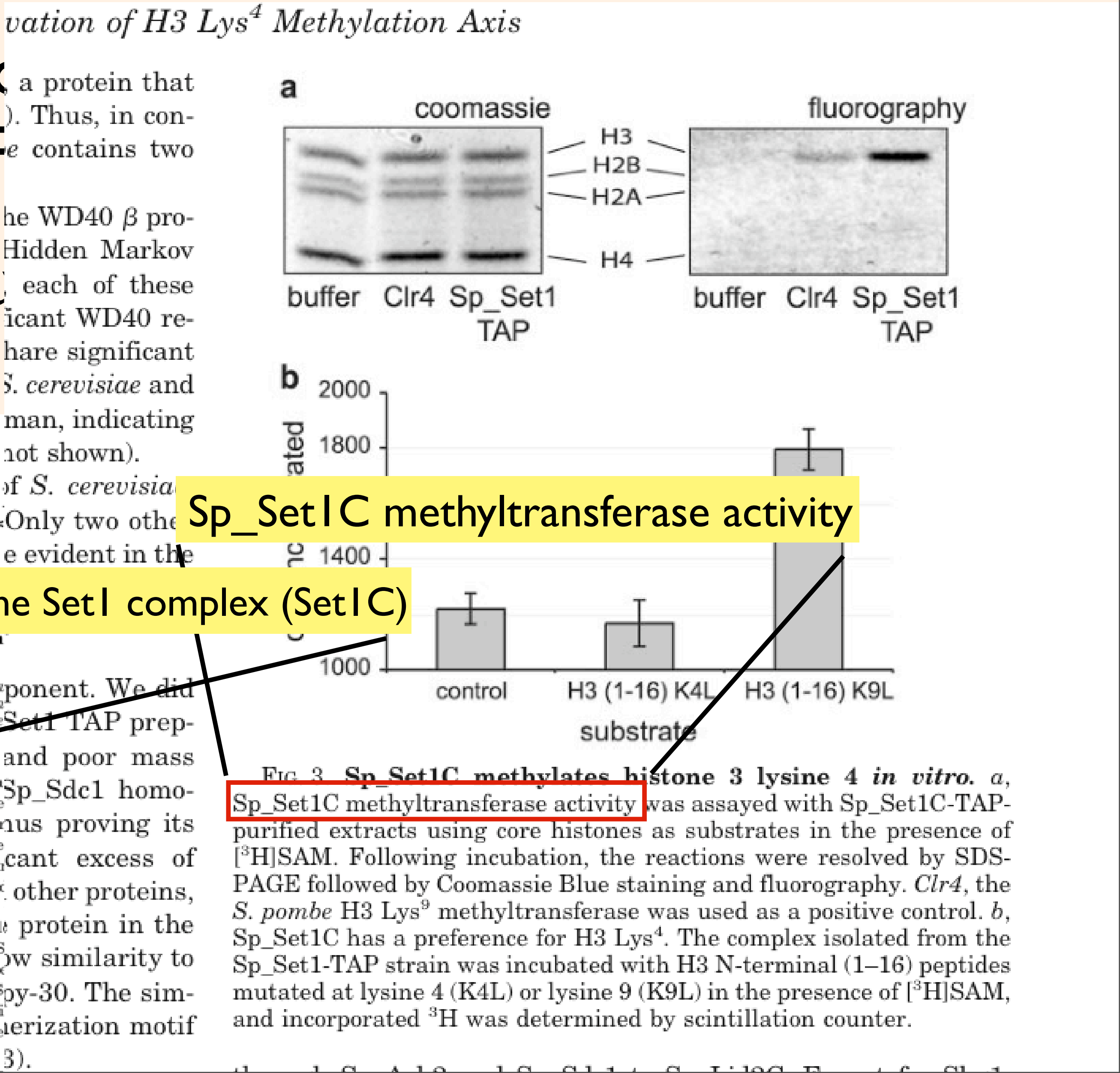

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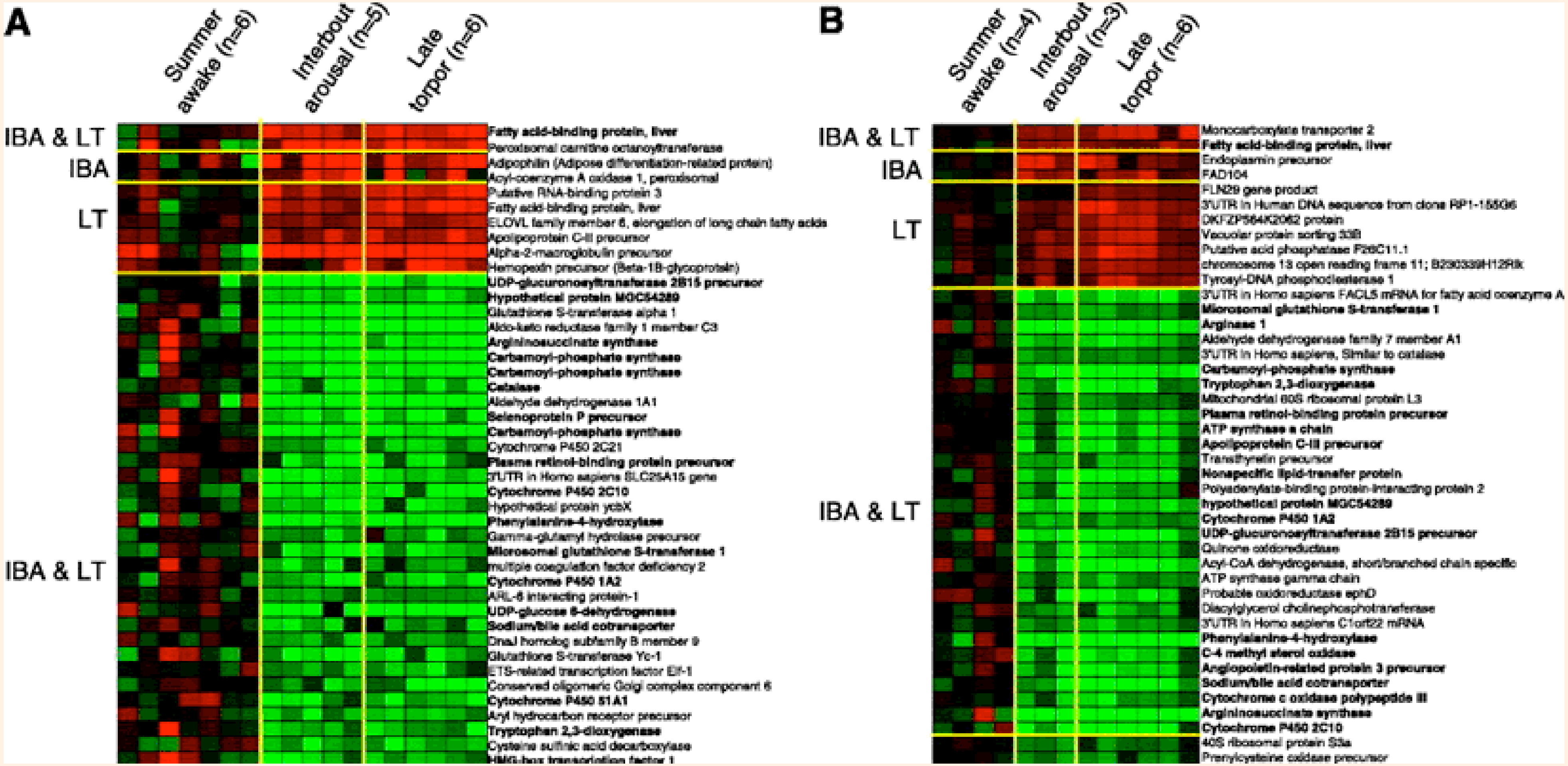
Sp_Set1C methyltransferase activity

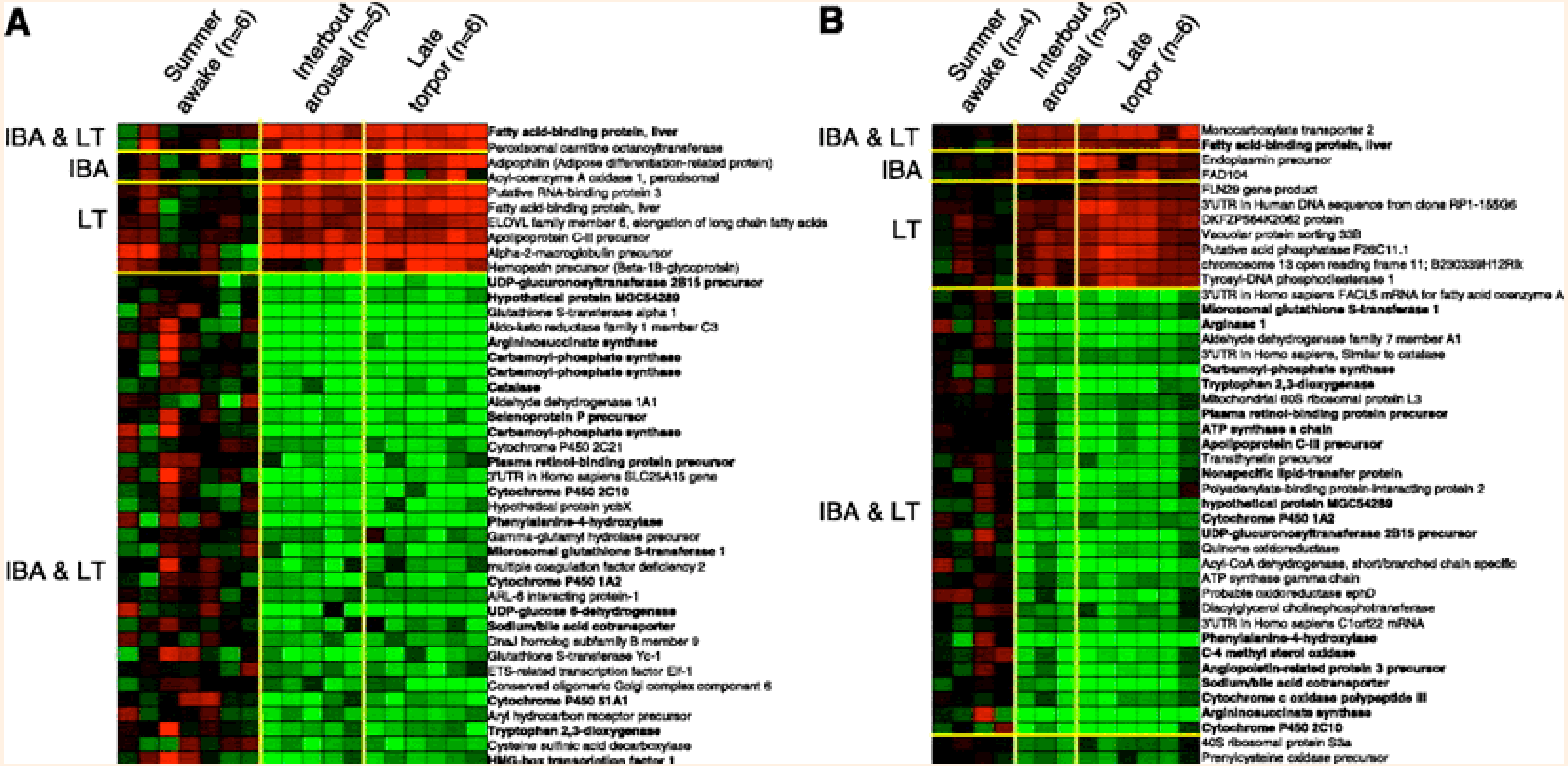
H3 Lys4 methylation ... is mediated by the Set1 complex (Set1C)

GO & gene expression

GO & gene expression

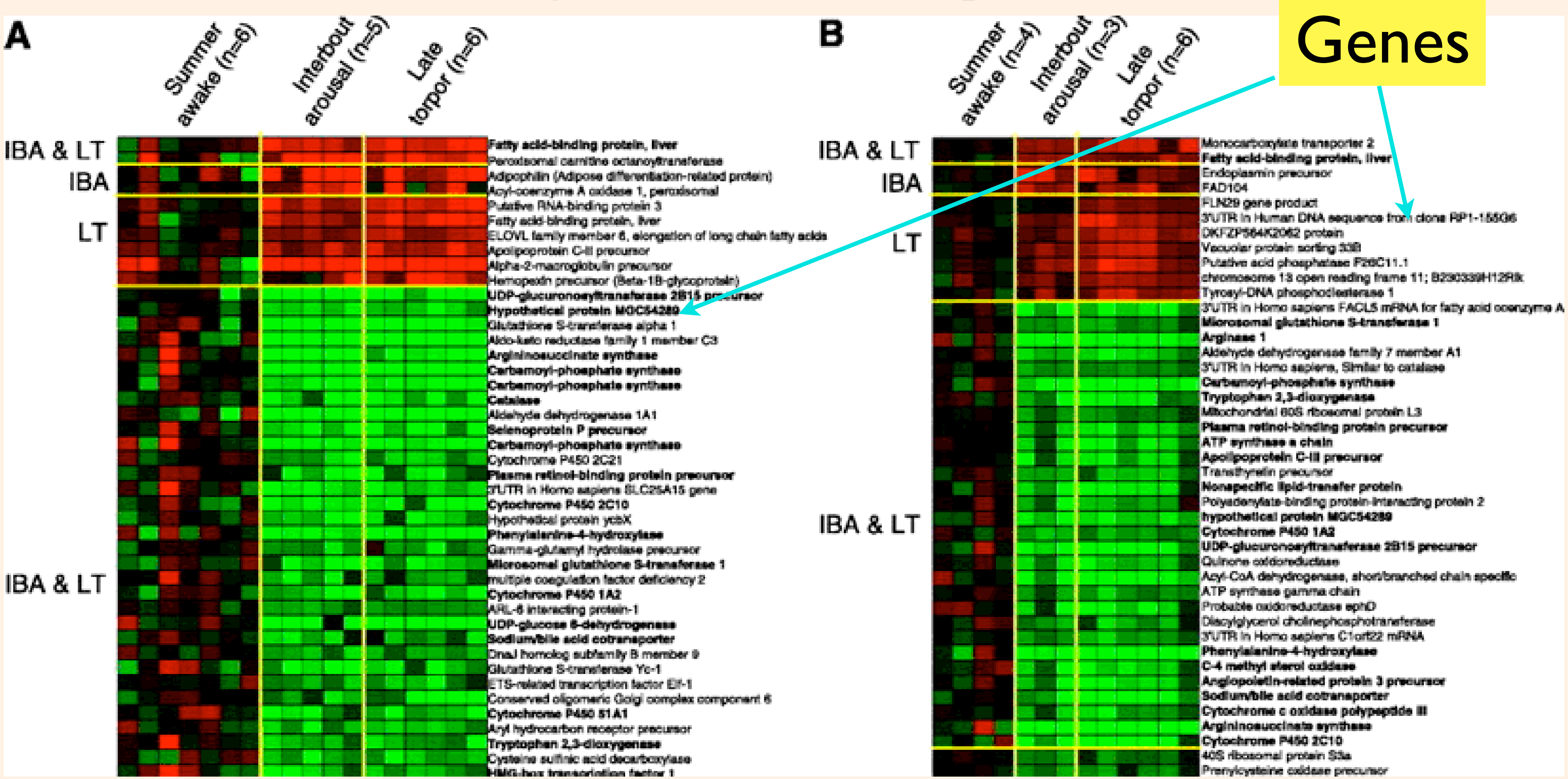


GO & gene expression



D.R. Williams et al. (2005) Physiol Genomics 24(1): 13–22 (Fig. 2)

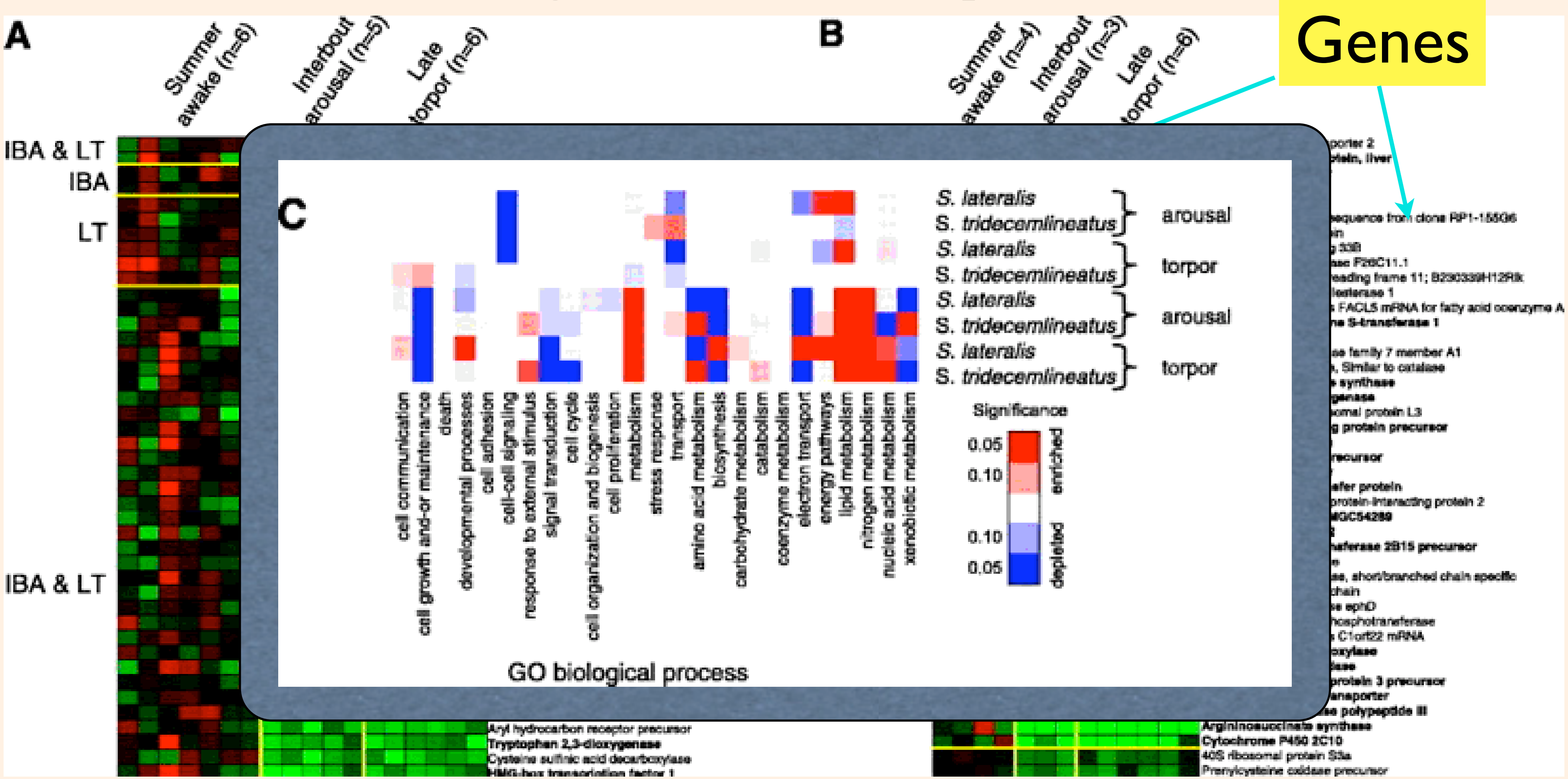
GO & gene expression



Genes

D.R. Williams et al. (2005) Physiol Genomics 24(1): 13–22 (Fig. 2)

GO & gene expression



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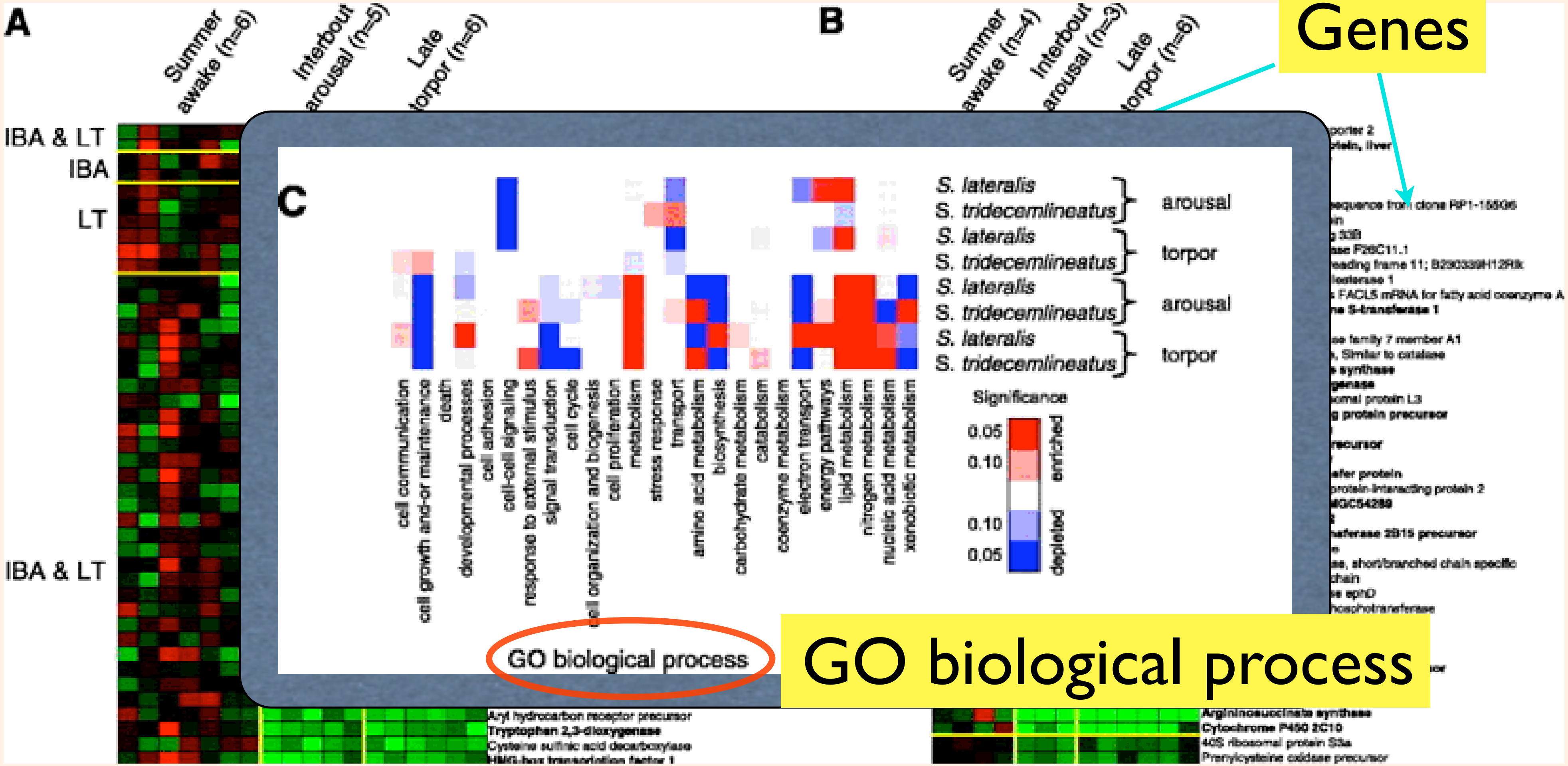


Genes

GO biological process

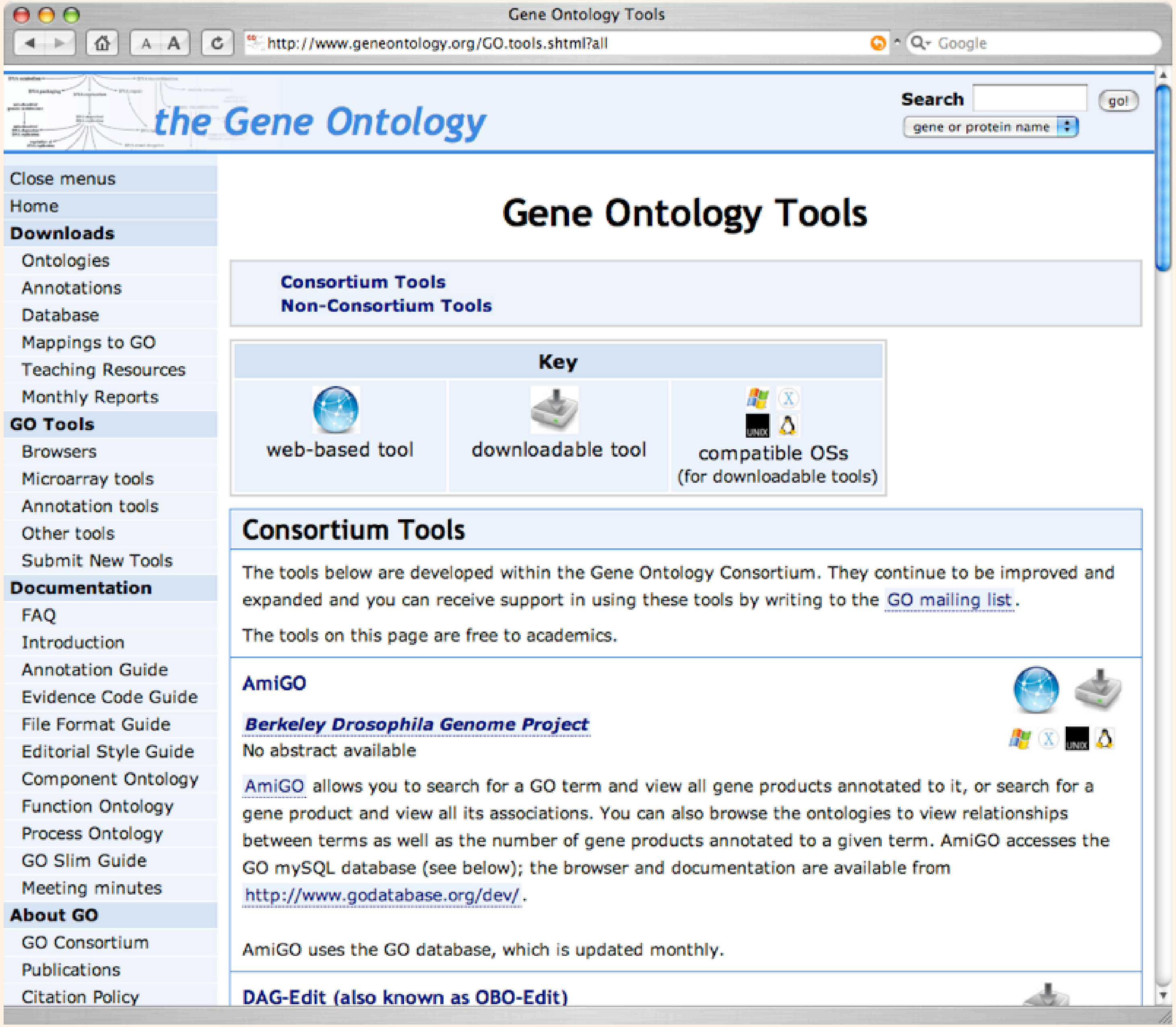
D.R. Williams et al. (2005) Physiol Genomics 24(1): 13–22 (Fig. 2)

GO Tools

- Database (and schema)
- Perl API
- Browsers, e.g. AmiGO
- Editing tool: OBO-Edit
- Tool collection:
  - Microarray analysis tools
  - Annotation tools
  - others ...

http://www.geneontology.org/GO.tools.shtml

GO Tools page



Gene Ontology Tools

Consortium Tools
Non-Consortium Tools

Key		
web-based tool	downloadable tool	compatible OSs (for downloadable tools)

Consortium Tools

The tools below are developed within the Gene Ontology Consortium. They continue to be improved and expanded and you can receive support in using these tools by writing to the GO mailing list.

The tools on this page are free to academics.

AmiGO

Berkeley Drosophila Genome Project

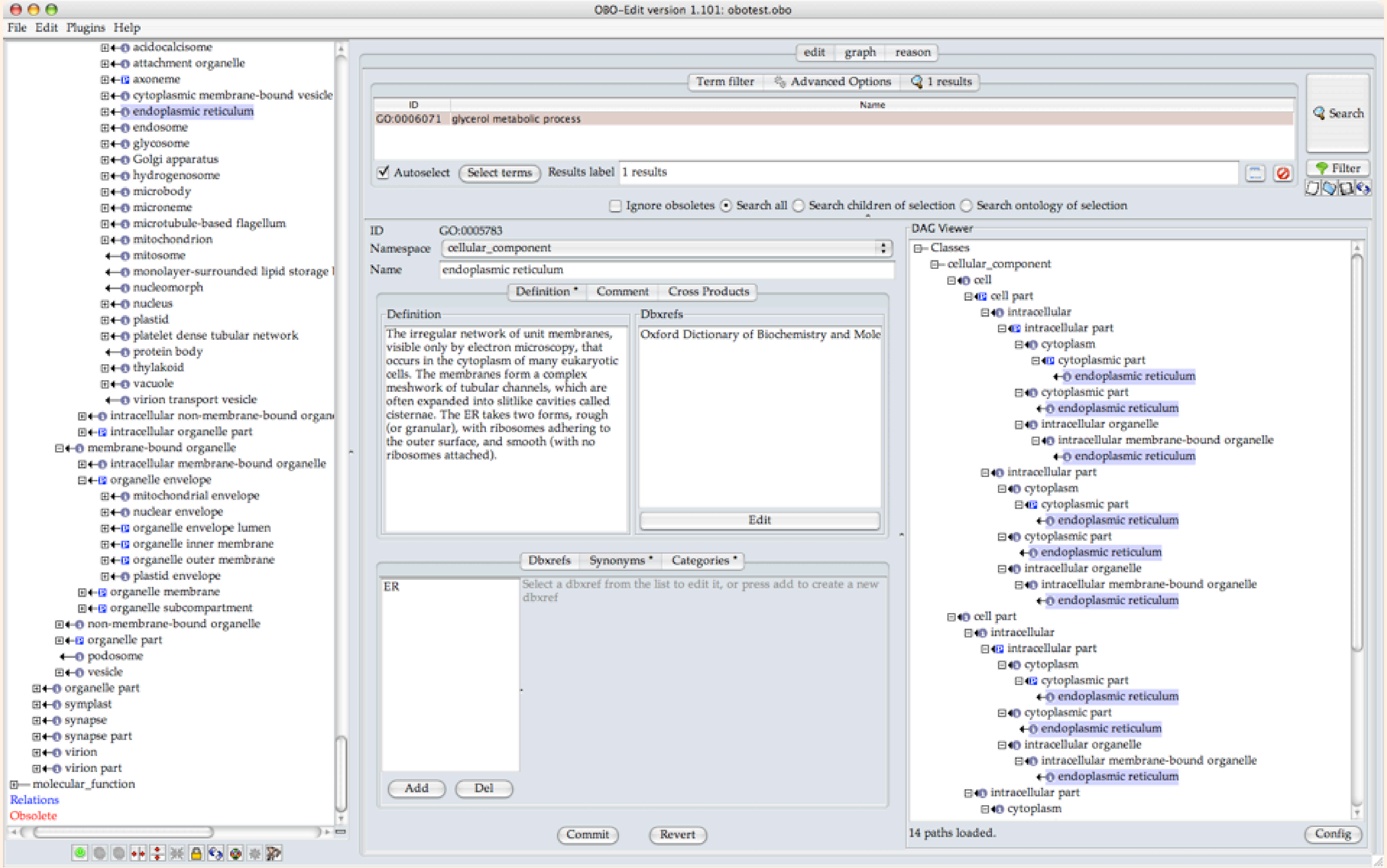
No abstract available

AmiGO allows you to search for a GO term and view all gene products annotated to it, or search for a gene product and view all its associations. You can also browse the ontologies to view relationships between terms as well as the number of gene products annotated to a given term. AmiGO accesses the GO mySQL database (see below); the browser and documentation are available from http://www.godatabase.org/dev/.

AmiGO uses the GO database, which is updated monthly.

DAG-Edit (also known as OBO-Edit)

OBO-Edit



OBO-Edit version 1.101: obotest.obo

ID	Name
GO:0006071	glycerol metabolic process

☑ Autoselect

Results label: 1 results

☐ Ignore obsoletes ● Search all ○ Search children of selection ○ Search ontology of selection

ID: GO:0005783

Namespace: cellular_component

Name: endoplasmic reticulum

Definition: The irregular network of unit membranes, visible only by electron microscopy, that occurs in the cytoplasm of many eukaryotic cells. The membranes form a complex meshwork of tubular channels, which are often expanded into slitlike cavities called cisternae. The ER takes two forms, rough (or granular), with ribosomes adhering to the outer surface, and smooth (with no ribosomes attached).

Dbxrefs: Oxford Dictionary of Biochemistry and Mole

Synonyms: ER

Select a dbxref from the list to edit it, or press add to create a new dbxref

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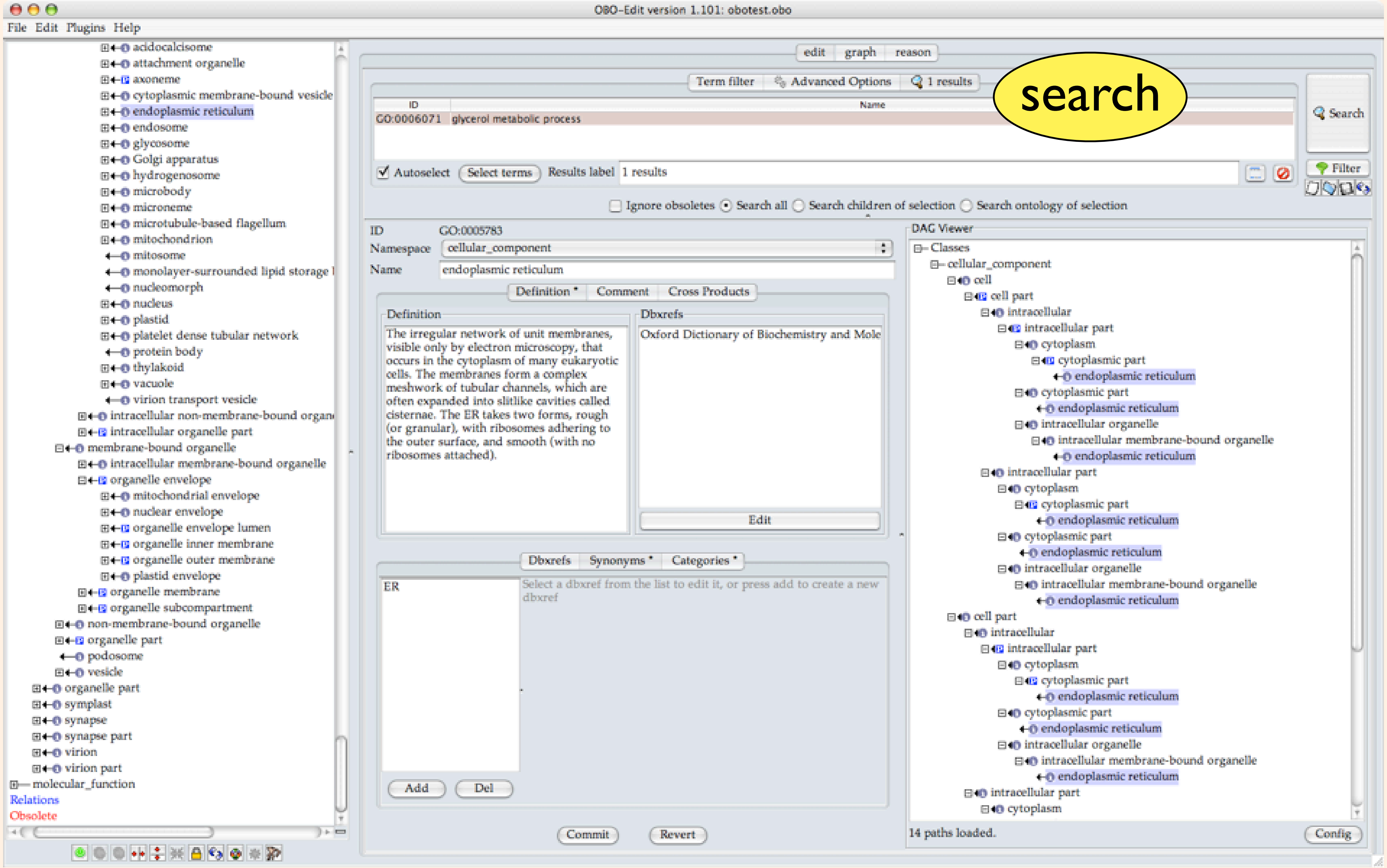

search

GO:0006071 glycerol metabolic process

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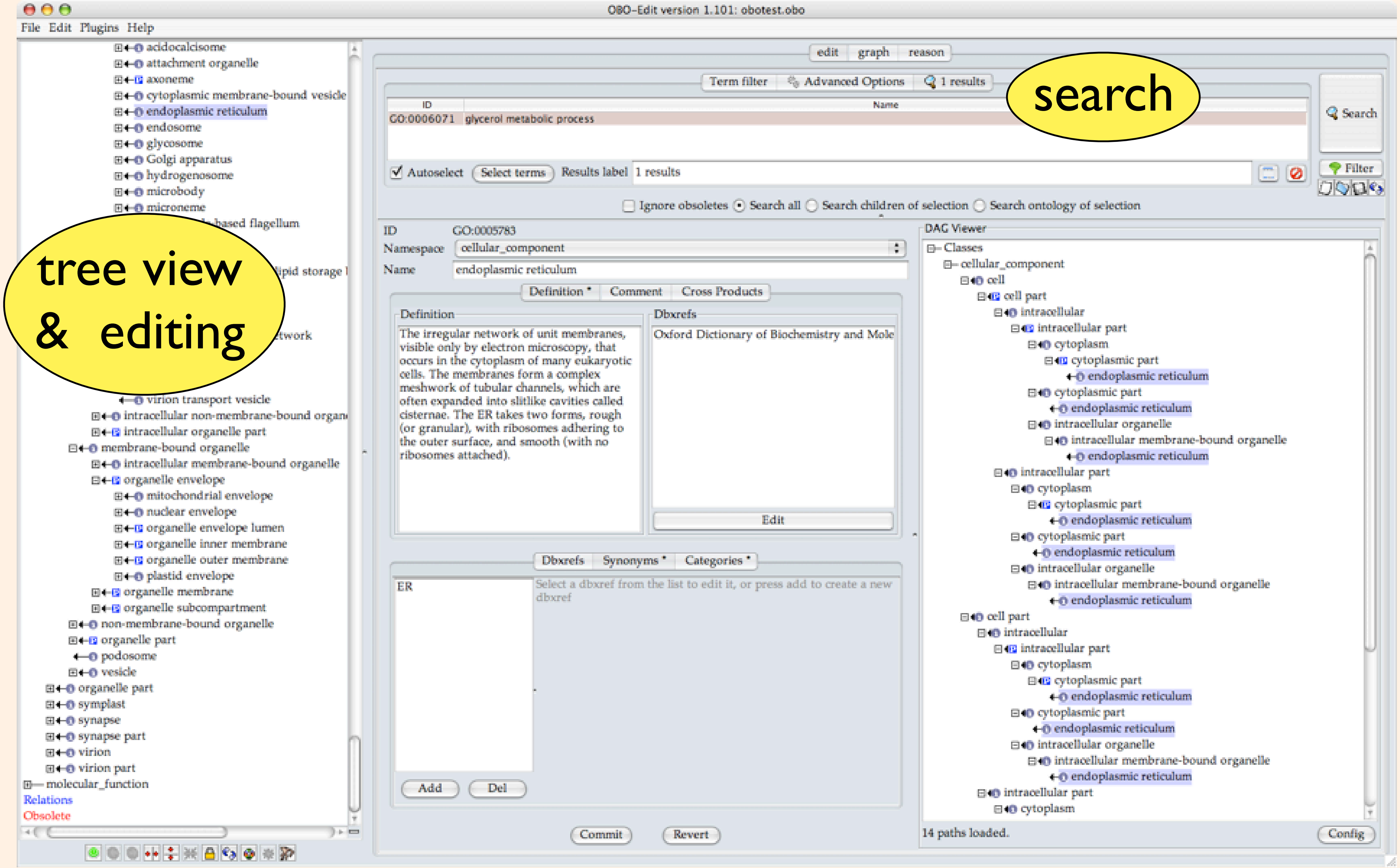

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- attachment organelle
- axoneme
- cytoplasmic membrane-bound vesicle
- endoplasmic reticulum
- endosome
- glycosome
- Golgi apparatus
- hydrogenosome
- microbody
- microneme
- -based flagellum
- lipid storage b
- twork
- virion transport vesicle
- intracellular non-membrane-bound organ
- intracellular organelle part
- membrane-bound organelle
  - intracellular membrane-bound organelle
  - organelle envelope
    - mitochondrial envelope
    - nuclear envelope
    - organelle envelope lumen
    - organelle inner membrane
    - organelle outer membrane
    - plastid envelope
  - organelle membrane
  - organelle subcompartment
- non-membrane-bound organelle
- organelle part
- podosome
- vesicle
- organelle part
- symplast
- synapse
- synapse part
- virion
- virion part
- molecular_function
- Relations
- Obsolete

ID	Name
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Definition * Comment Cross Products

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Dbxrefs

Oxford Dictionary of Biochemistry and Mole

Edit

Dbxrefs Synonyms * Categories *

ER

Select a dbxref from the list to edit it, or press add to create a new dbxref

Add Del

Commit Revert

DAG Viewer

- Classes
  - cellular_component
    - cell
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        - intracellular
          - intracellular part
            - cytoplasm
              - cytoplasmic part
                - endoplasmic reticulum
            - cytoplasmic part
              - endoplasmic reticulum
            - intracellular organelle
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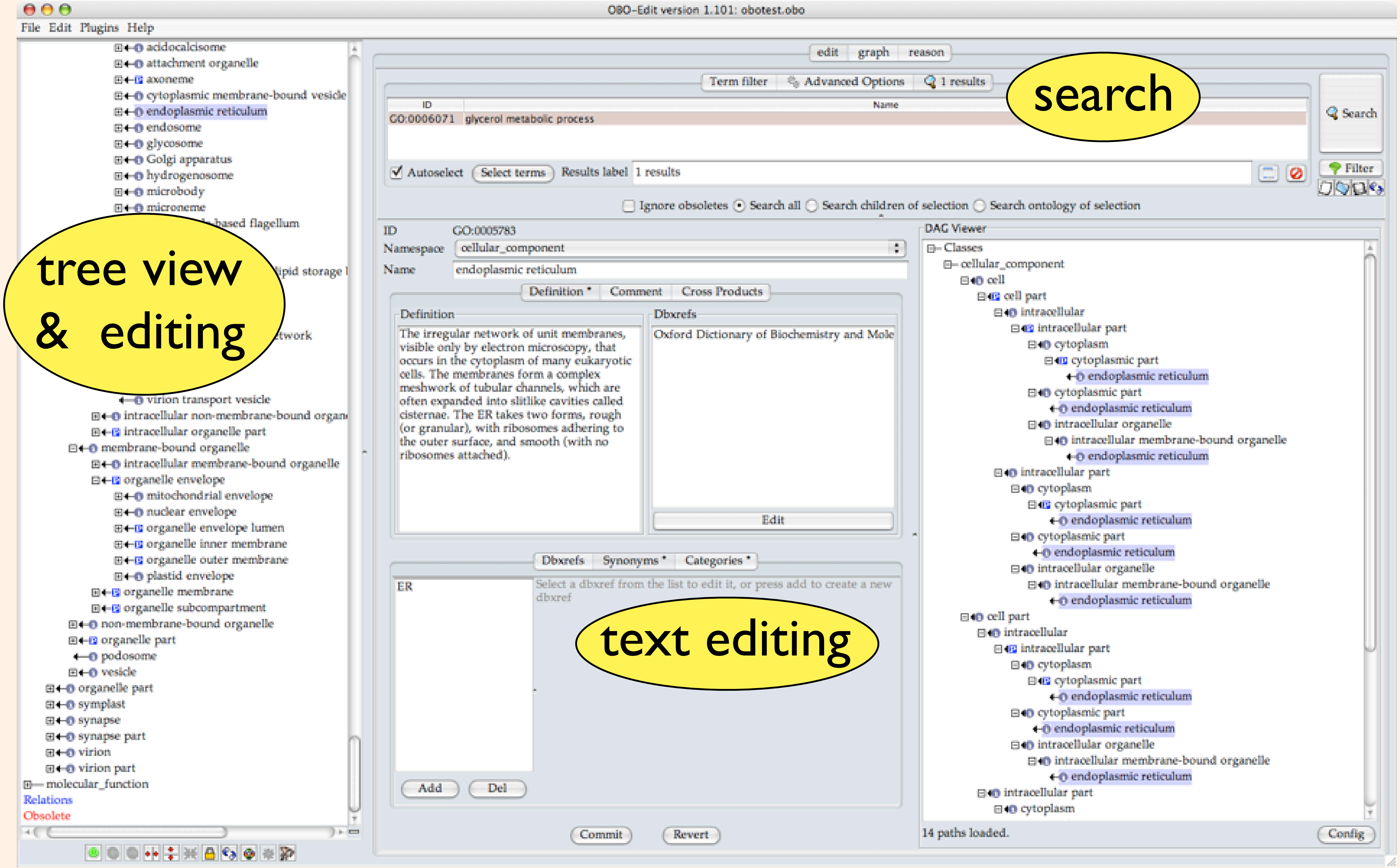
tree view
& editing

OBO-Edit


search

tree view & editing

text editing

OBO-Edit version 1.101: obotest.obo

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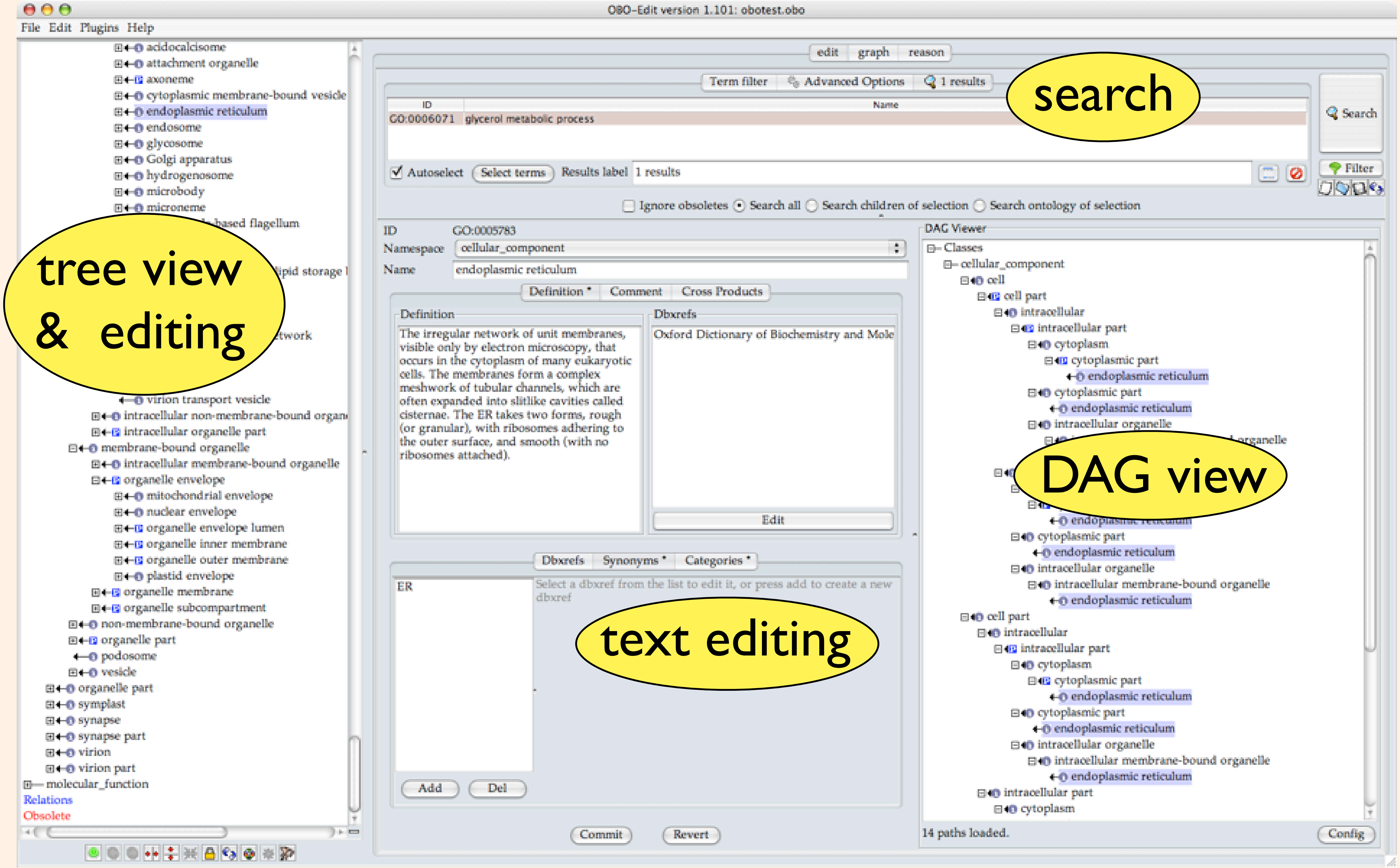

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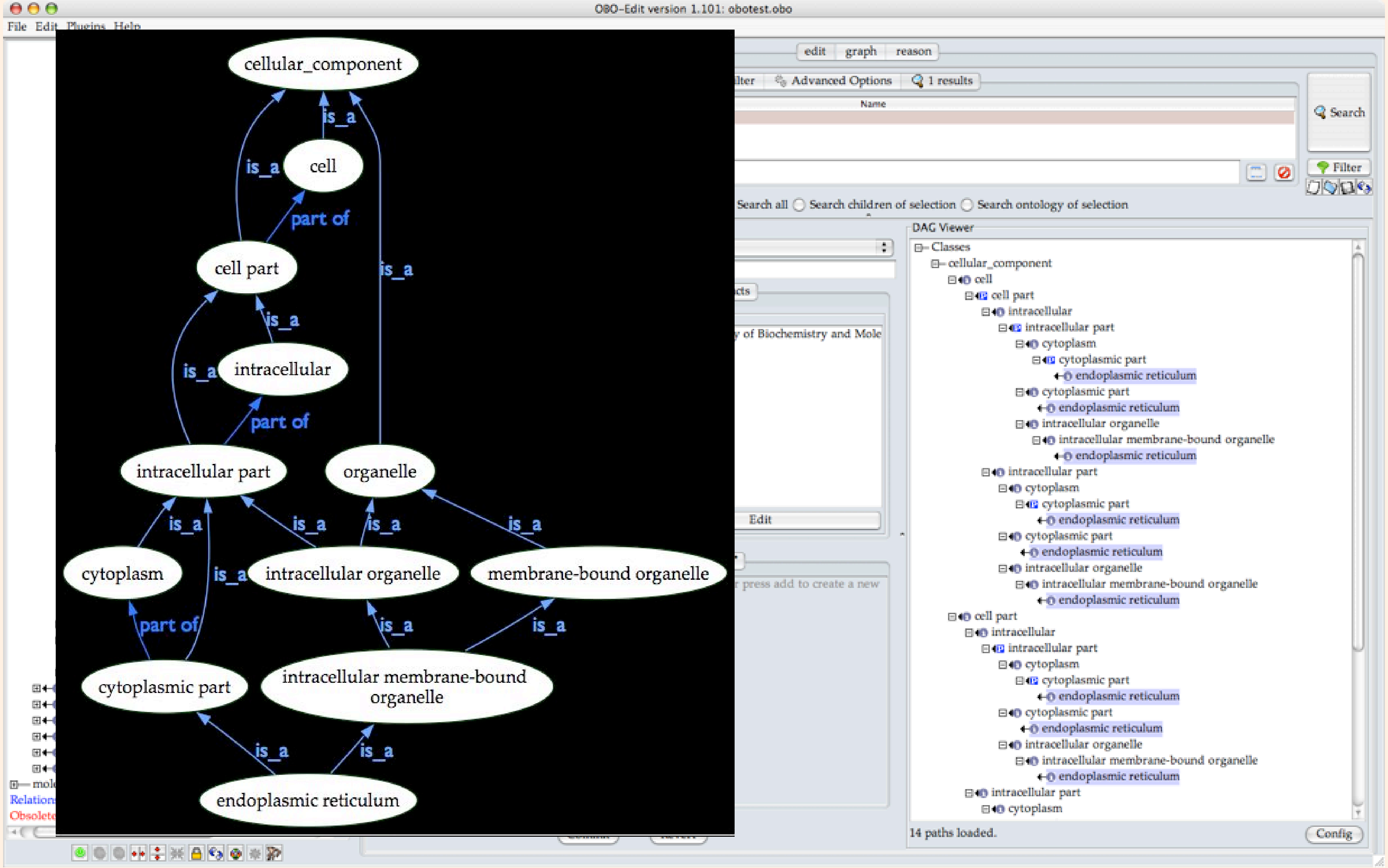

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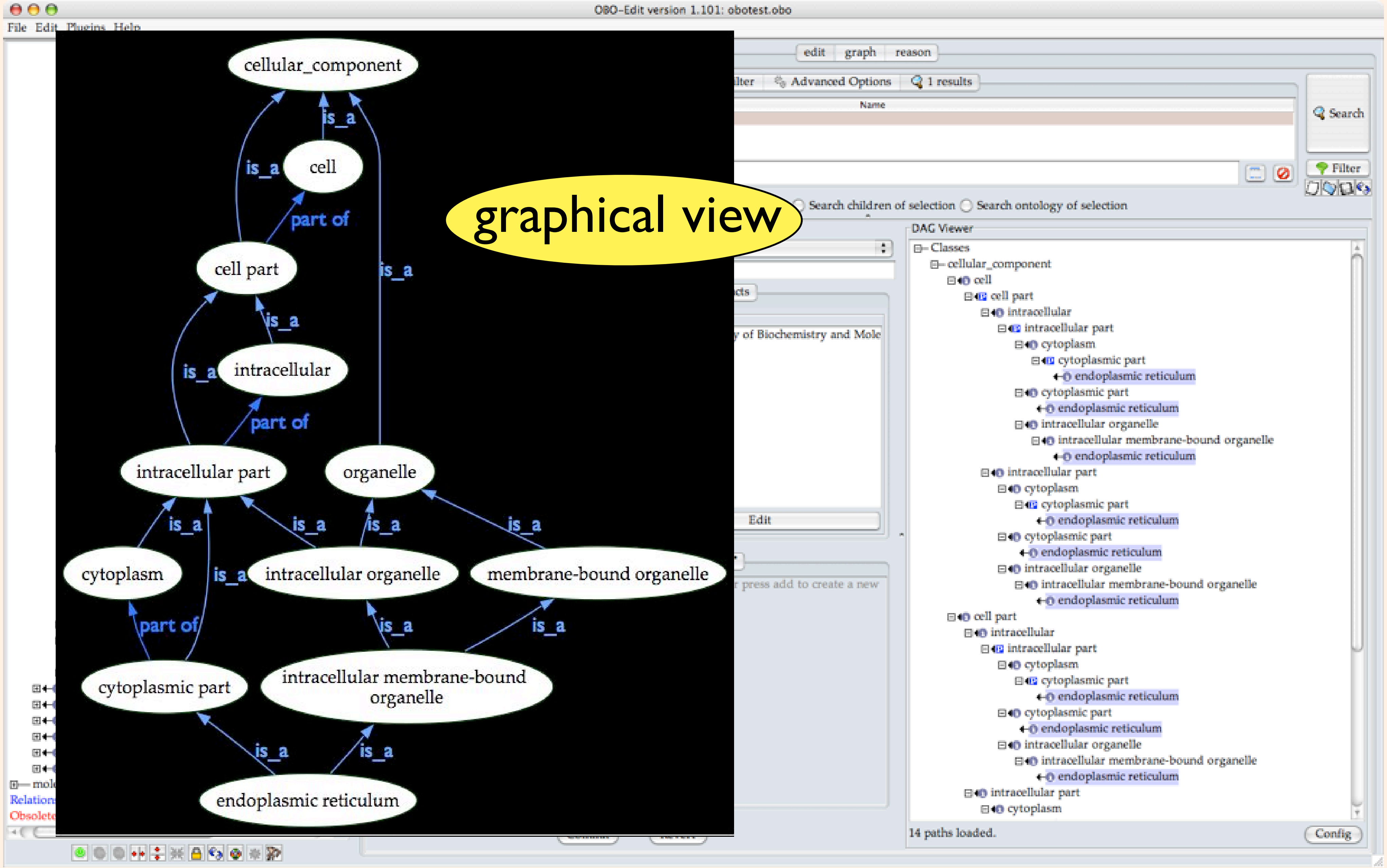
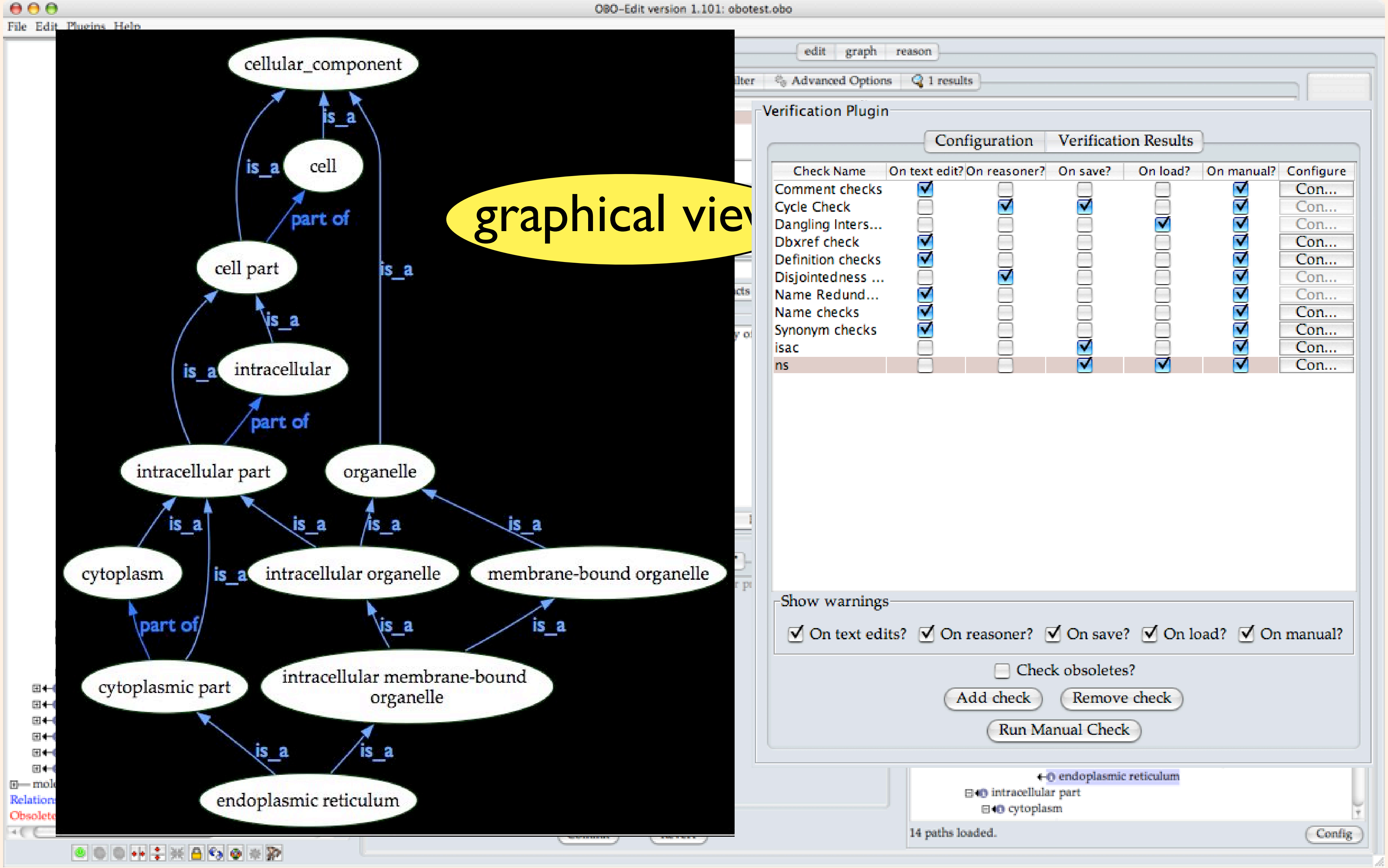


graphical view

OBO-Edit



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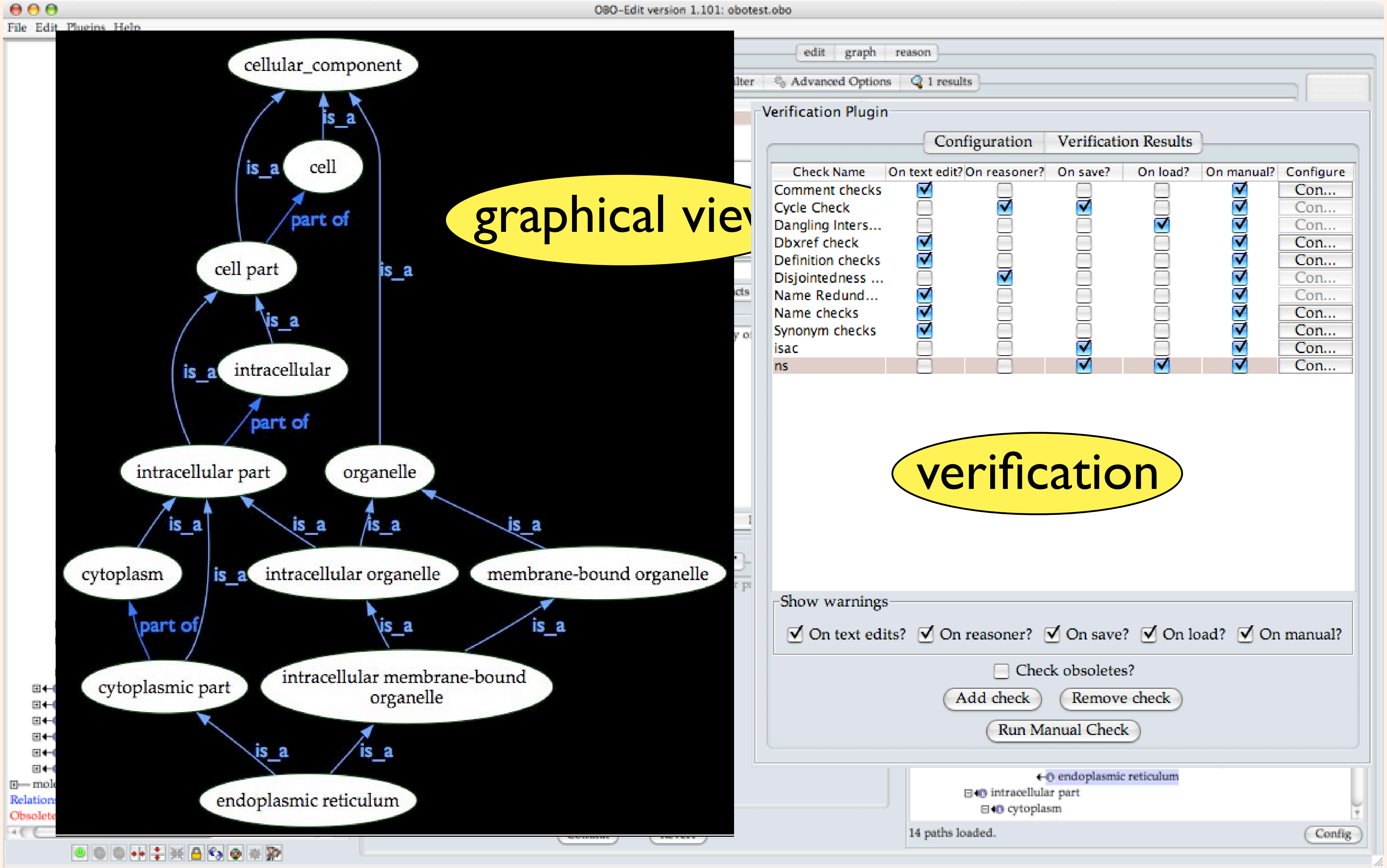
Cycle Check	☐	☑	☑	☐	☑	Con...
Dangling Inters...	☐	☐	☐	☑	☑	Con...
Dbxref check	☑	☐	☐	☐	☑	Con...
Definition checks	☑	☐	☐	☐	☑	Con...
Disjointedness ...	☐	☑	☐	☐	☑	Con...
Name Redund...	☑	☐	☐	☐	☑	Con...
Name checks	☑	☐	☐	☐	☑	Con...
Synonym checks	☑	☐	☐	☐	☑	Con...
isac	☐	☐	☑	☐	☑	Con...
ns	☐	☐	☑	☑	☑	Con...

Show warnings

☑ On text edits? ☑ On reasoner? ☑ On save? ☑ On load? ☑ On manual?

☐ Check obsoletes?

OBO-Edit



Verification Plugin

Configuration | Verification Results

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Cycle Check	☐	☑	☑	☐	☑	Con...
Dangling Inters...	☐	☐	☐	☑	☑	Con...
Dbxref check	☑	☐	☐	☐	☑	Con...
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Show warnings

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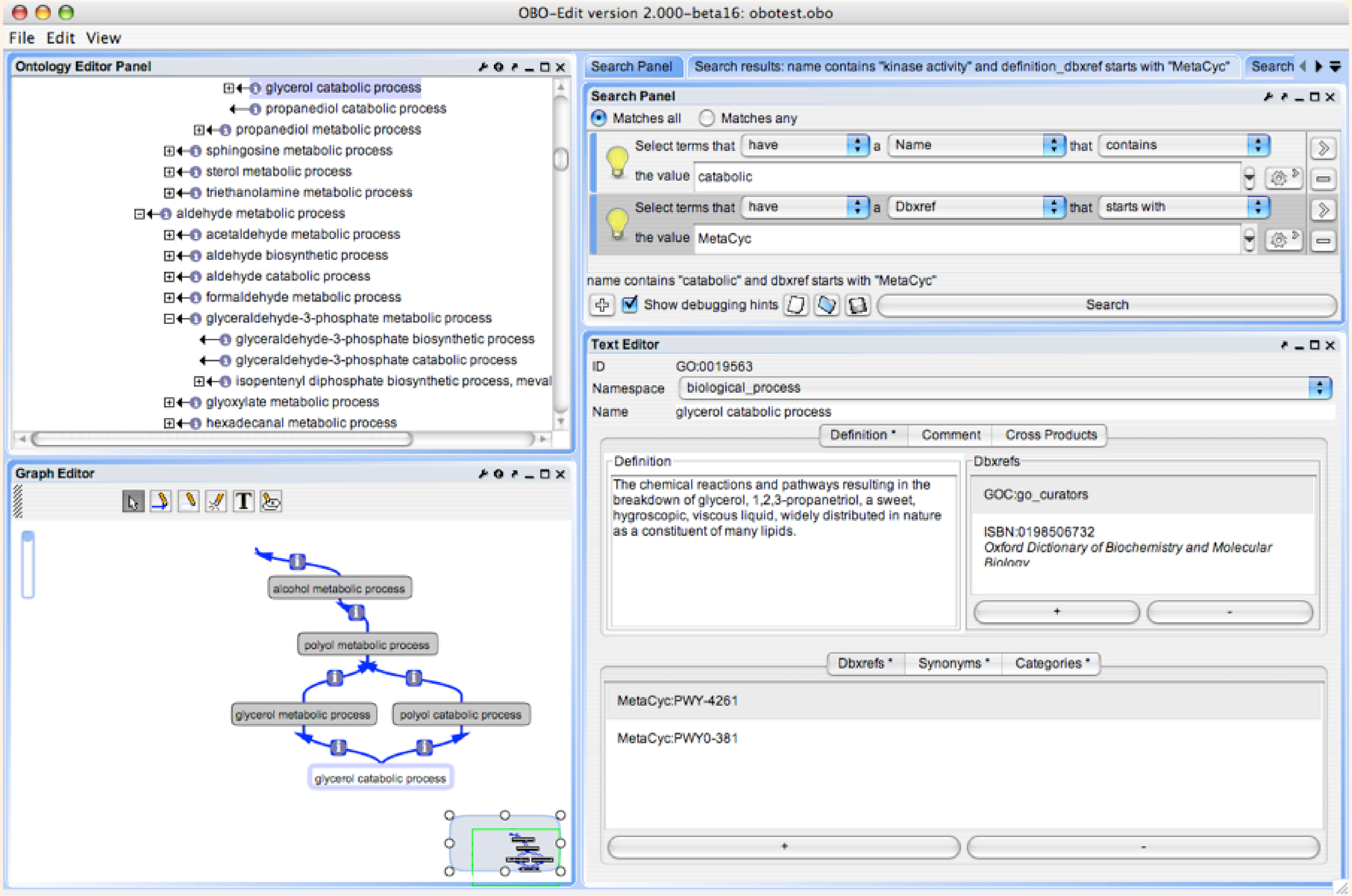
☐ Check obsoletes?

Add check | Remove check

Run Manual Check

14 paths loaded.

OBO-Edit 2.0



OBO-Edit version 2.000-beta16: obotest.obo

File Edit View

Ontology Editor Panel

- glycerol catabolic process
- propanediol catabolic process
- propanediol metabolic process
- sphingosine metabolic process
- sterol metabolic process
- triethanolamine metabolic process
- aldehyde metabolic process
  - acetaldehyde metabolic process
  - aldehyde biosynthetic process
  - aldehyde catabolic process
  - formaldehyde metabolic process
  - glyceraldehyde-3-phosphate metabolic process
    - glyceraldehyde-3-phosphate biosynthetic process
    - glyceraldehyde-3-phosphate catabolic process
    - isopentenyl diphosphate biosynthetic process, meval
  - glyoxylate metabolic process
  - hexadecanal metabolic process

Graph Editor

Search Panel | Search results: name contains "kinase activity" and definition_dbxref starts with "MetaCyc" | Search

Search Panel

● Matches all ○ Matches any

Select terms that: have a Name that contains
the value: catabolic

Select terms that: have a Dbxref that starts with
the value: MetaCyc

name contains "catabolic" and dbxref starts with "MetaCyc"

☑ Show debugging hints

Text Editor

ID: GO:0019563

Namespace: biological_process

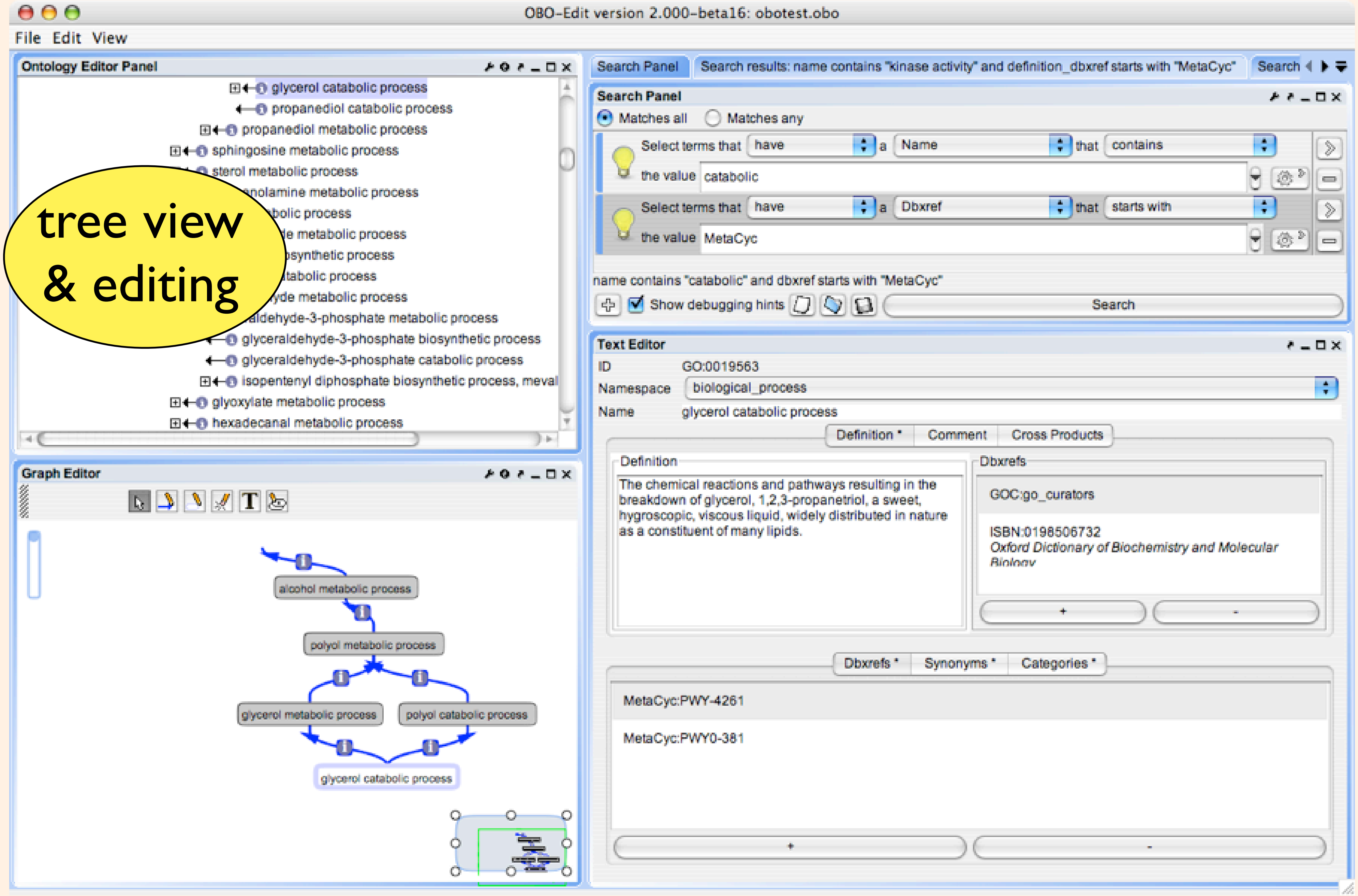
Name: glycerol catabolic process

Definition: The chemical reactions and pathways resulting in the breakdown of glycerol, 1,2,3-propanetriol, a sweet, hygroscopic, viscous liquid, widely distributed in nature as a constituent of many lipids.

Dbxrefs:
GOC:go_curators
ISBN:0198506732 *Oxford Dictionary of Biochemistry and Molecular Biology*

MetaCyc:PWY-4261
MetaCyc:PWY0-381

OBO-Edit 2.0



OBO-Edit version 2.000-beta16: obotest.obo

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- propanediol catabolic process
- propanediol metabolic process
- sphingosine metabolic process
- sterol metabolic process
- …nolamine metabolic process
- …bolic process
- …e metabolic process
- …synthetic process
- …tabolic process
- …yde metabolic process
- …aldehyde-3-phosphate metabolic process
- glyceraldehyde-3-phosphate biosynthetic process
- glyceraldehyde-3-phosphate catabolic process
- isopentenyl diphosphate biosynthetic process, meval
- glyoxylate metabolic process
- hexadecanal metabolic process

Search results: name contains "kinase activity" and definition_dbxref starts with "MetaCyc"

● Matches all ○ Matches any

Select terms that have a Name that contains the value catabolic

Select terms that have a Dbxref that starts with the value MetaCyc

name contains "catabolic" and dbxref starts with "MetaCyc"

☑ Show debugging hints

alcohol metabolic process → polyol metabolic process → glycerol metabolic process, polyol catabolic process → glycerol catabolic process

ID GO:0019563

Namespace biological_process

Name glycerol catabolic process

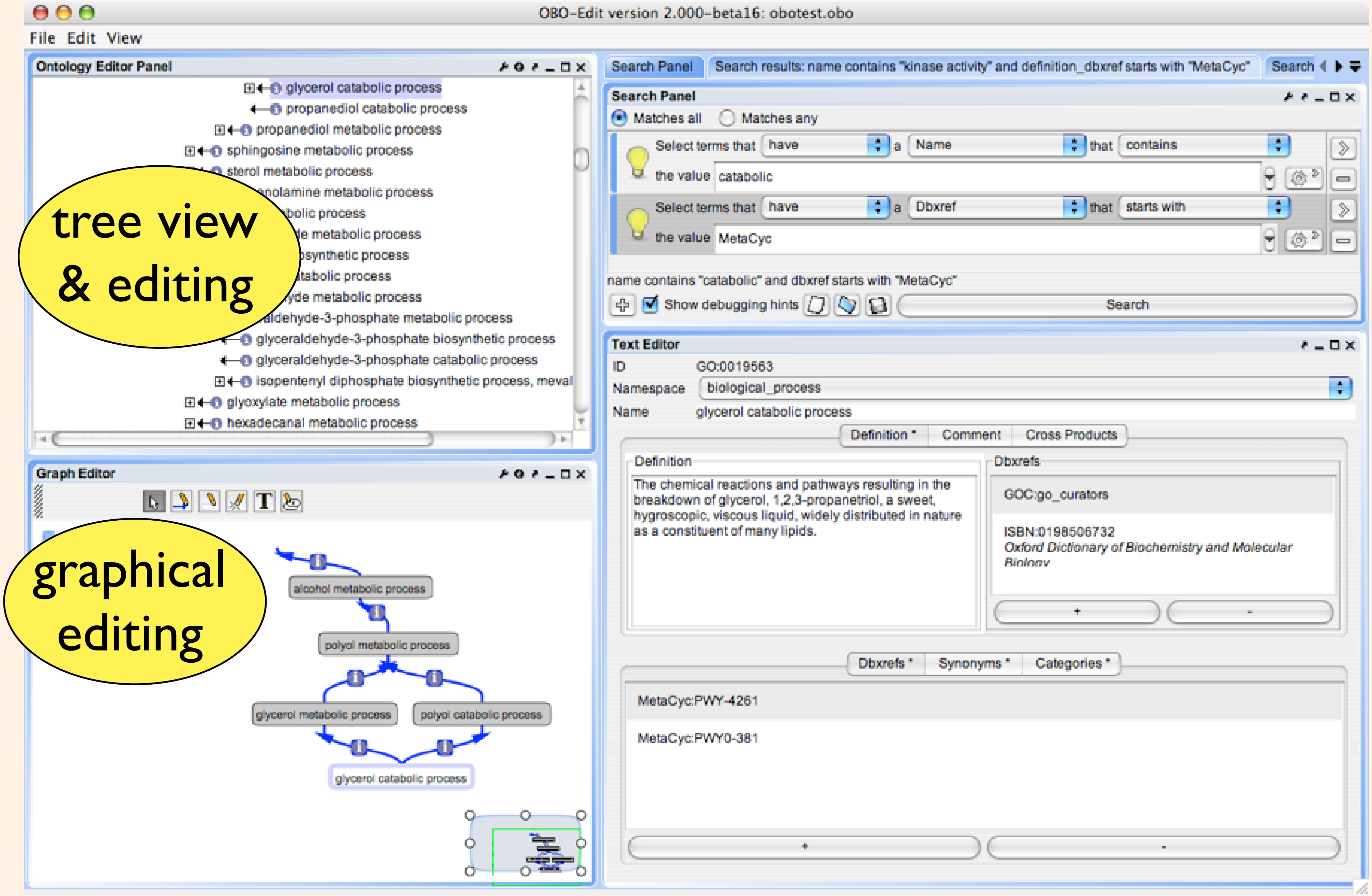
Definition: The chemical reactions and pathways resulting in the breakdown of glycerol, 1,2,3-propanetriol, a sweet, hygroscopic, viscous liquid, widely distributed in nature as a constituent of many lipids.

Dbxrefs:
GOC:go_curators
ISBN:0198506732 *Oxford Dictionary of Biochemistry and Molecular Biology*

MetaCyc:PWY-4261
MetaCyc:PWY0-381

tree view & editing

OBO-Edit 2.0



OBO-Edit version 2.000-beta16: obotest.obo

Search results: name contains "kinase activity" and definition_dbxref starts with "MetaCyc"

Search Panel

● Matches all ○ Matches any

Select terms that: have a Name that contains

the value: catabolic

Select terms that: have a Dbxref that starts with

the value: MetaCyc

name contains "catabolic" and dbxref starts with "MetaCyc"

☑ Show debugging hints

Text Editor

ID: GO:0019563

Namespace: biological_process

Name: glycerol catabolic process

Definition: The chemical reactions and pathways resulting in the breakdown of glycerol, 1,2,3-propanetriol, a sweet, hygroscopic, viscous liquid, widely distributed in nature as a constituent of many lipids.

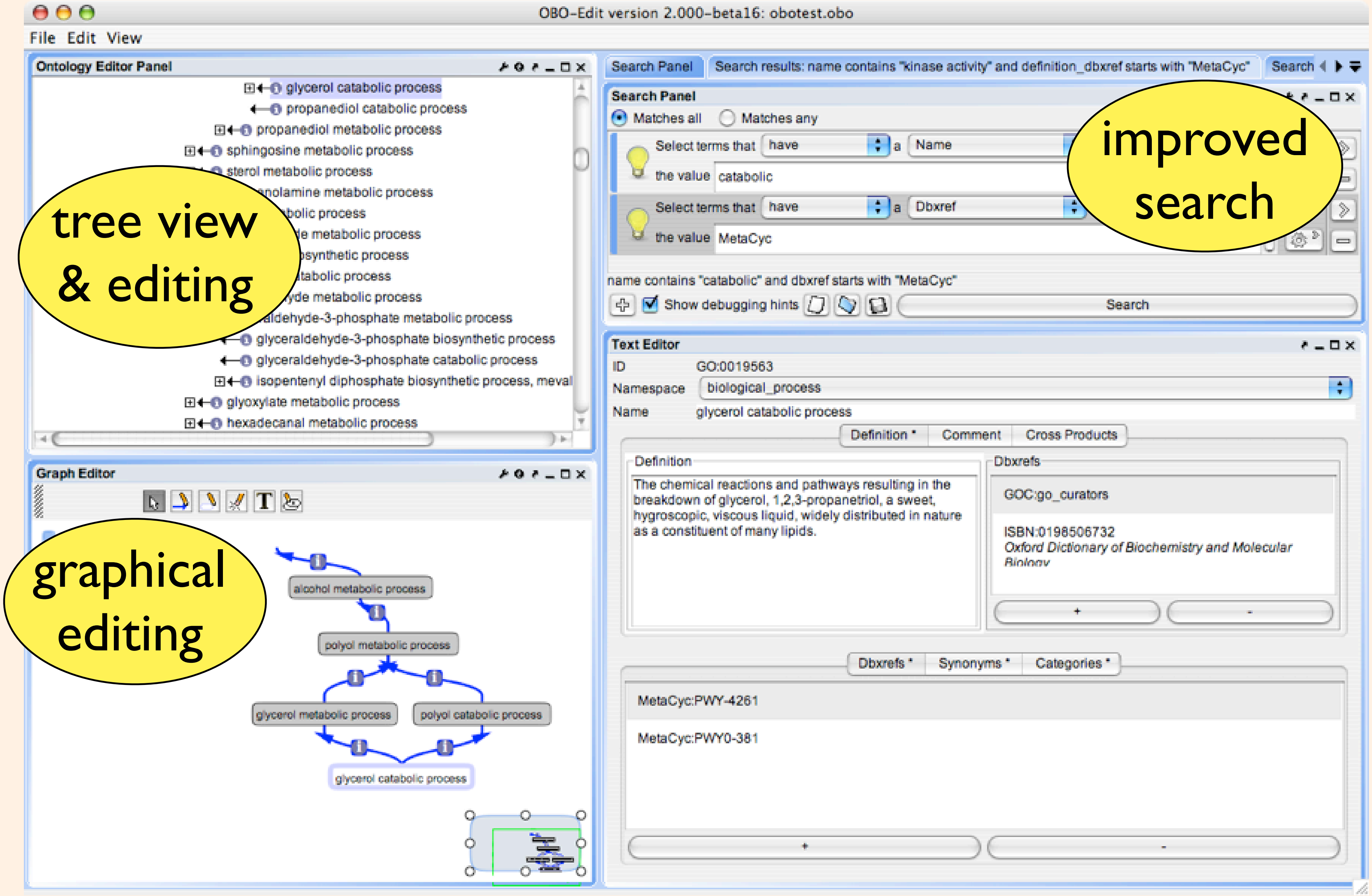
Dbxrefs: GOC:go_curators; ISBN:0198506732 *Oxford Dictionary of Biochemistry and Molecular Biology*

Dbxrefs: MetaCyc:PWY-4261; MetaCyc:PWY0-381

tree view & editing

graphical editing

OBO-Edit 2.0



OBO-Edit version 2.000-beta16: obotest.obo

- glycerol catabolic process
- propanediol catabolic process
- propanediol metabolic process
- sphingosine metabolic process
- sterol metabolic process
- …nolamine metabolic process
- …bolic process
- …e metabolic process
- …synthetic process
- …tabolic process
- …yde metabolic process
- …aldehyde-3-phosphate metabolic process
- glyceraldehyde-3-phosphate biosynthetic process
- glyceraldehyde-3-phosphate catabolic process
- isopentenyl diphosphate biosynthetic process, meval
- glyoxylate metabolic process
- hexadecanal metabolic process

Search results: name contains "kinase activity" and definition_dbxref starts with "MetaCyc"

● Matches all ○ Matches any

Select terms that: have a Name

the value: catabolic

Select terms that: have a Dbxref

the value: MetaCyc

name contains "catabolic" and dbxref starts with "MetaCyc"

☑ Show debugging hints

ID: GO:0019563

Namespace: biological_process

Name: glycerol catabolic process

Definition * Comment Cross Products

Definition: The chemical reactions and pathways resulting in the breakdown of glycerol, 1,2,3-propanetriol, a sweet, hygroscopic, viscous liquid, widely distributed in nature as a constituent of many lipids.

Dbxrefs:
- GOC:go_curators
- ISBN:0198506732 *Oxford Dictionary of Biochemistry and Molecular Biology*

Dbxrefs * Synonyms * Categories *

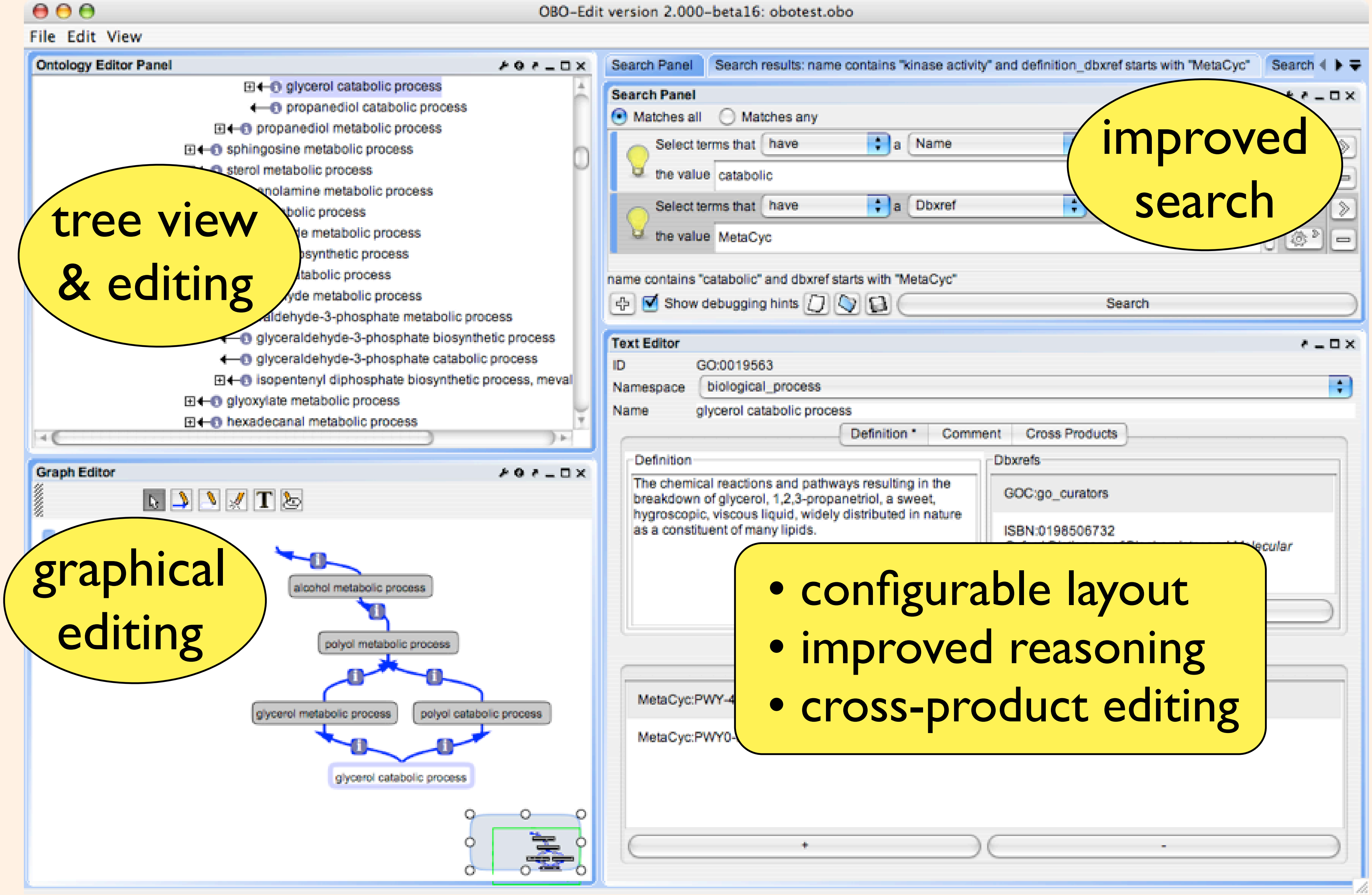
- MetaCyc:PWY-4261
- MetaCyc:PWY0-381

tree view & editing

improved search

graphical editing

OBO-Edit 2.0



- glycerol catabolic process
- propanediol catabolic process
- propanediol metabolic process
- sphingosine metabolic process
- sterol metabolic process
- glyceraldehyde-3-phosphate biosynthetic process
- glyceraldehyde-3-phosphate catabolic process
- isopentenyl diphosphate biosynthetic process, meval
- glyoxylate metabolic process
- hexadecanal metabolic process

Search results: name contains "kinase activity" and definition_dbxref starts with "MetaCyc"

● Matches all ○ Matches any

Select terms that have a Name the value catabolic

Select terms that have a Dbxref the value MetaCyc

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☑ Show debugging hints Search

ID GO:0019563

Namespace biological_process

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Dbxrefs: GOC:go_curators; ISBN:0198506732

MetaCyc:PWY-4

MetaCyc:PWY0-

tree view & editing

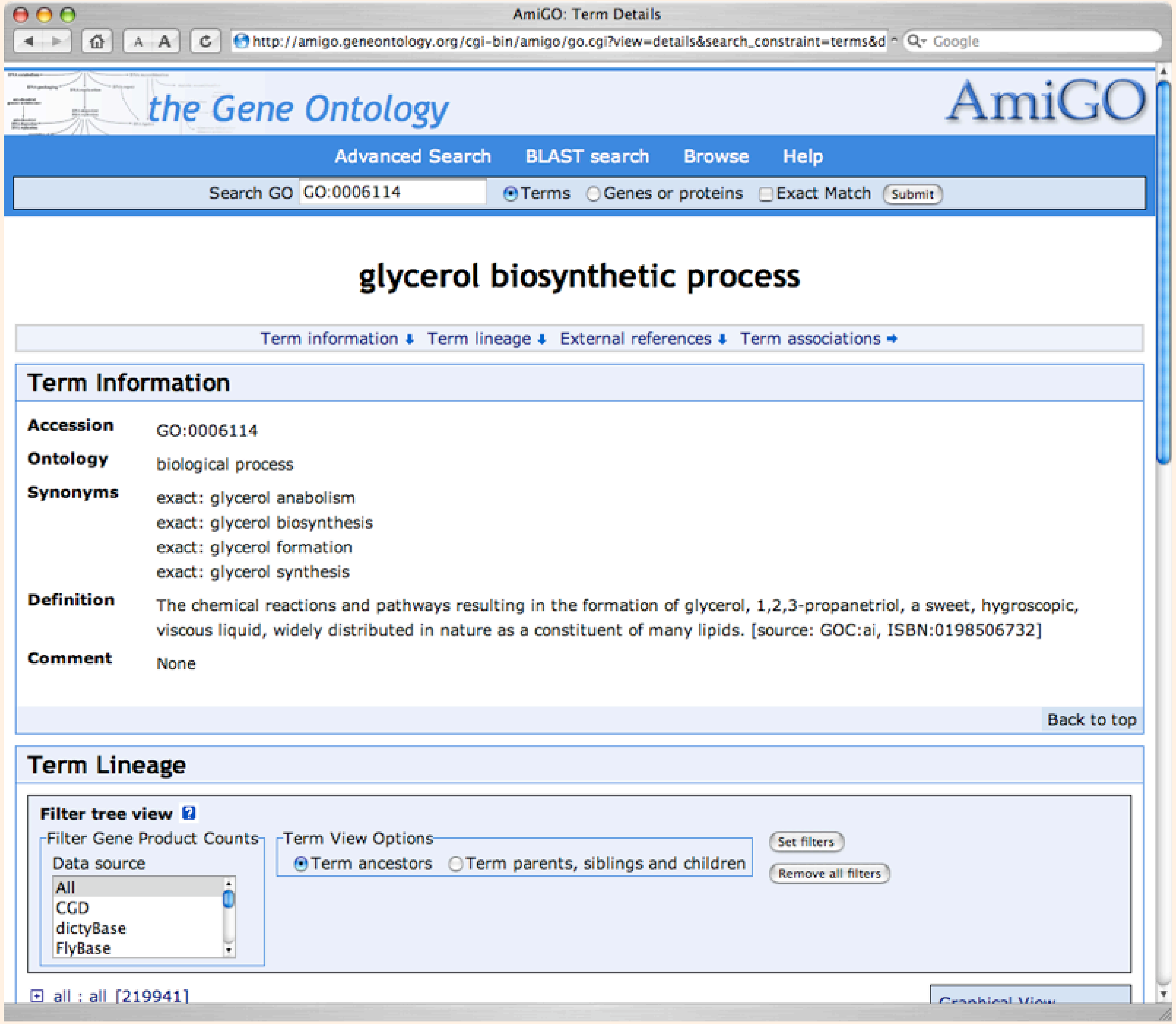
improved search

graphical editing

- configurable layout
- improved reasoning
- cross-product editing

AmiGO

AmiGO



Search GO GO:0006114 ● Terms ○ Genes or proteins ☐ Exact Match

glycerol biosynthetic process

Term Information

Accession	GO:0006114
Ontology	biological process
Synonyms	exact: glycerol anabolism exact: glycerol biosynthesis exact: glycerol formation exact: glycerol synthesis
Definition	The chemical reactions and pathways resulting in the formation of glycerol, 1,2,3-propanetriol, a sweet, hygroscopic, viscous liquid, widely distributed in nature as a constituent of many lipids. [source: GOC:ai, ISBN:0198506732]
Comment	None

Term Lineage

Filter tree view

Filter Gene Product Counts

Data source

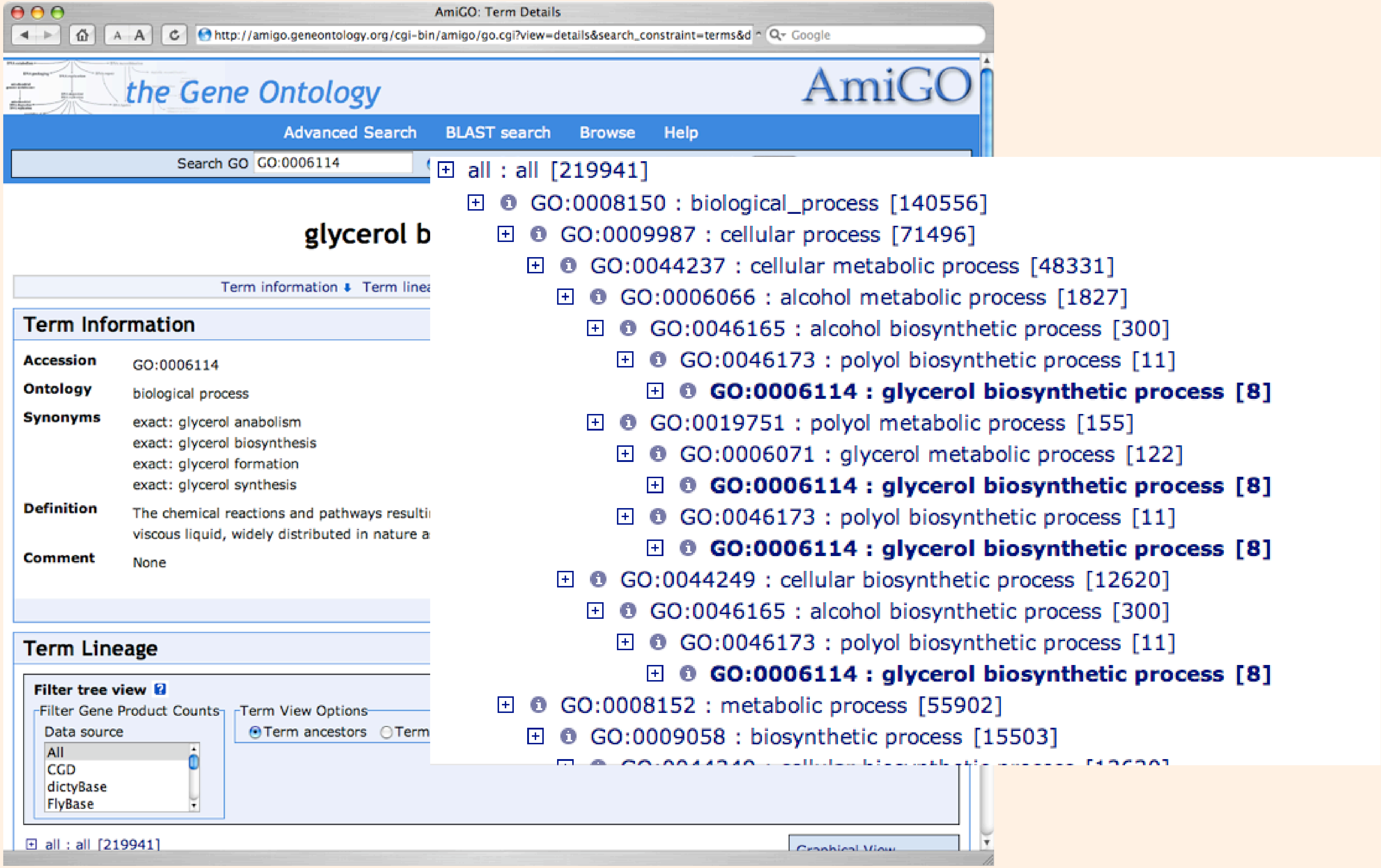
- All
- CGD
- dictyBase
- FlyBase

Term View Options

● Term ancestors ○ Term parents, siblings and children

all : all [219941]

AmiGO



Search GO GO:0006114

glycerol b

Term Information

Accession	GO:0006114
Ontology	biological process
Synonyms	exact: glycerol anabolism exact: glycerol biosynthesis exact: glycerol formation exact: glycerol synthesis
Definition	The chemical reactions and pathways resulti viscous liquid, widely distributed in nature a
Comment	None

Term Lineage

Filter tree view

Filter Gene Product Counts

Data source

- All
- CGD
- dictyBase
- FlyBase

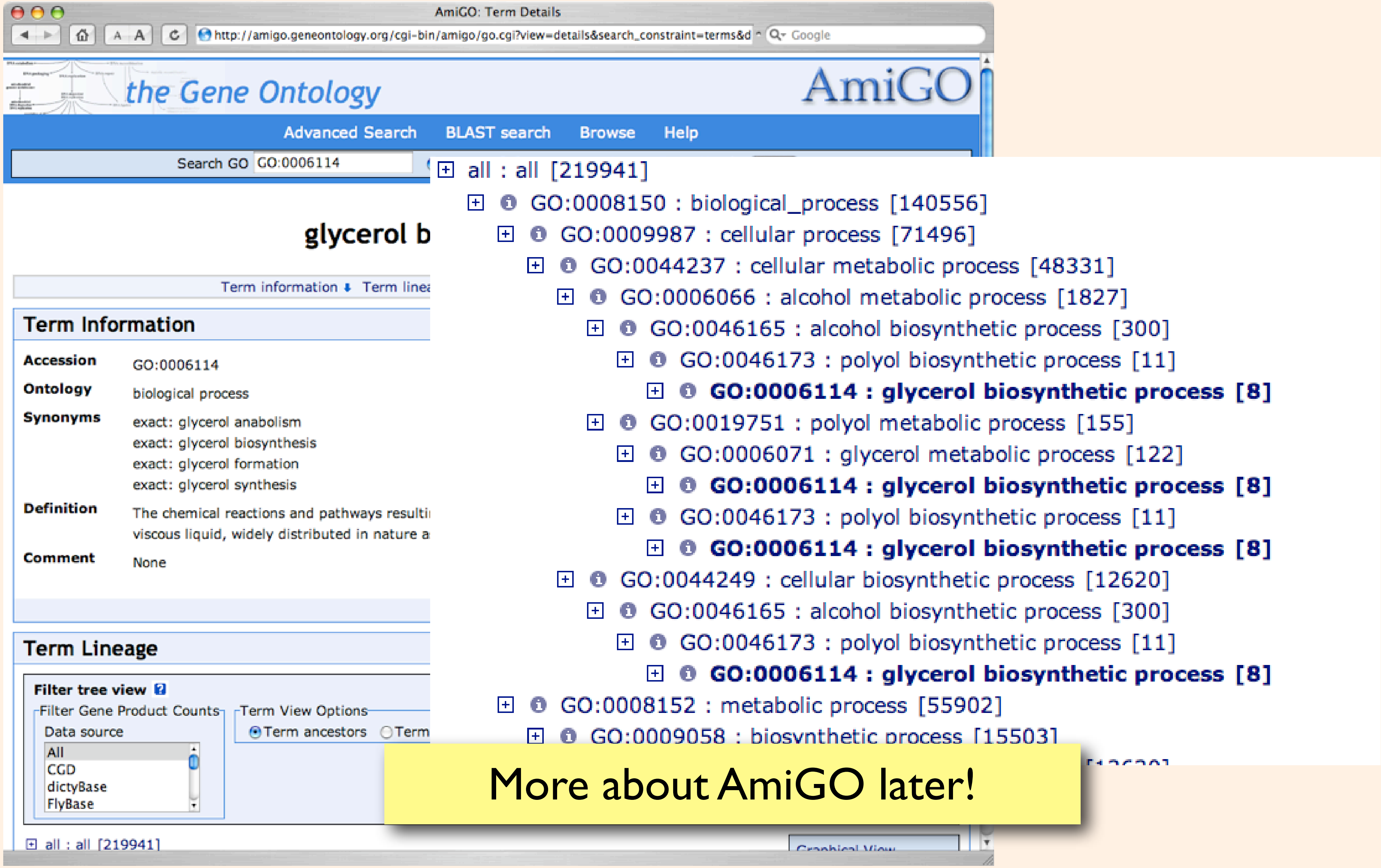
Term View Options

● Term ancestors ○ Term

⊞ all : all [219941]

```
⊞ all : all [219941]
  ⊞ GO:0008150 : biological_process [140556]
    ⊞ GO:0009987 : cellular process [71496]
      ⊞ GO:0044237 : cellular metabolic process [48331]
        ⊞ GO:0006066 : alcohol metabolic process [1827]
          ⊞ GO:0046165 : alcohol biosynthetic process [300]
            ⊞ GO:0046173 : polyol biosynthetic process [11]
              ⊞ GO:0006114 : glycerol biosynthetic process [8]
          ⊞ GO:0019751 : polyol metabolic process [155]
            ⊞ GO:0006071 : glycerol metabolic process [122]
              ⊞ GO:0006114 : glycerol biosynthetic process [8]
            ⊞ GO:0046173 : polyol biosynthetic process [11]
              ⊞ GO:0006114 : glycerol biosynthetic process [8]
        ⊞ GO:0044249 : cellular biosynthetic process [12620]
          ⊞ GO:0046165 : alcohol biosynthetic process [300]
            ⊞ GO:0046173 : polyol biosynthetic process [11]
              ⊞ GO:0006114 : glycerol biosynthetic process [8]
    ⊞ GO:0008152 : metabolic process [55902]
      ⊞ GO:0009058 : biosynthetic process [15503]
```

AmiGO



GO:0006114

glycerol b

Term information ⬇ Term linea

Term Information

Accession GO:0006114

Ontology biological process

Synonyms exact: glycerol anabolism
exact: glycerol biosynthesis
exact: glycerol formation
exact: glycerol synthesis

Definition The chemical reactions and pathways resulti
viscous liquid, widely distributed in nature a

Comment None

Term Lineage

Filter tree view

Filter Gene Product Counts
Data source
All
CGD
dictyBase
FlyBase

Term View Options
● Term ancestors ○ Term

all : all [219941]

```
all : all [219941]
  GO:0008150 : biological_process [140556]
    GO:0009987 : cellular process [71496]
      GO:0044237 : cellular metabolic process [48331]
        GO:0006066 : alcohol metabolic process [1827]
          GO:0046165 : alcohol biosynthetic process [300]
            GO:0046173 : polyol biosynthetic process [11]
              GO:0006114 : glycerol biosynthetic process [8]
          GO:0019751 : polyol metabolic process [155]
            GO:0006071 : glycerol metabolic process [122]
              GO:0006114 : glycerol biosynthetic process [8]
            GO:0046173 : polyol biosynthetic process [11]
              GO:0006114 : glycerol biosynthetic process [8]
        GO:0044249 : cellular biosynthetic process [12620]
          GO:0046165 : alcohol biosynthetic process [300]
            GO:0046173 : polyol biosynthetic process [11]
              GO:0006114 : glycerol biosynthetic process [8]
    GO:0008152 : metabolic process [55902]
      GO:0009058 : biosynthetic process [15503]
```

More about AmiGO later!

GO and Other Ontologies

Ontologies and Experiments

human lung fibroblasts
treated with cycloheximide

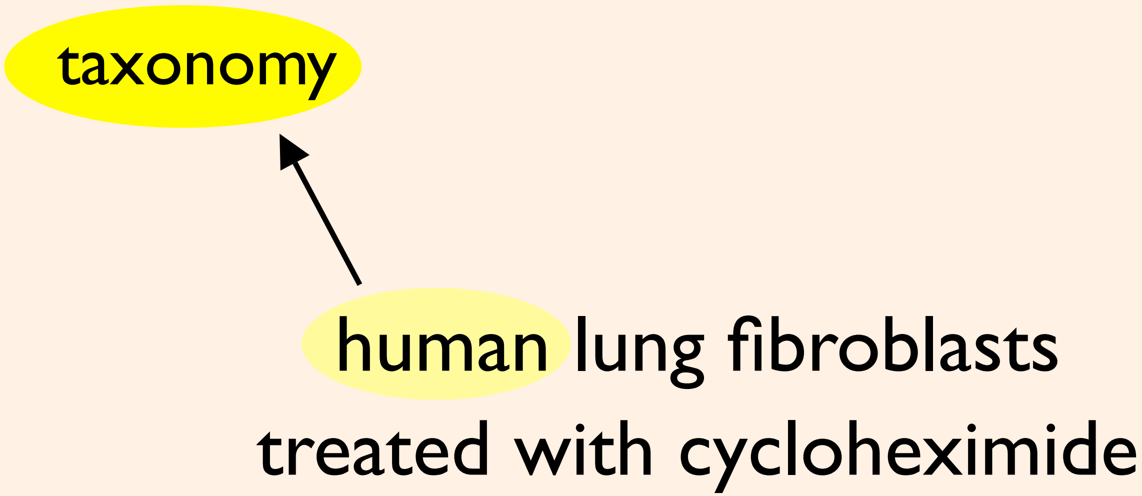
Ontologies and Experiments

human lung fibroblasts
treated with cycloheximide

Ontologies and Experiments

human lung fibroblasts
treated with cycloheximide

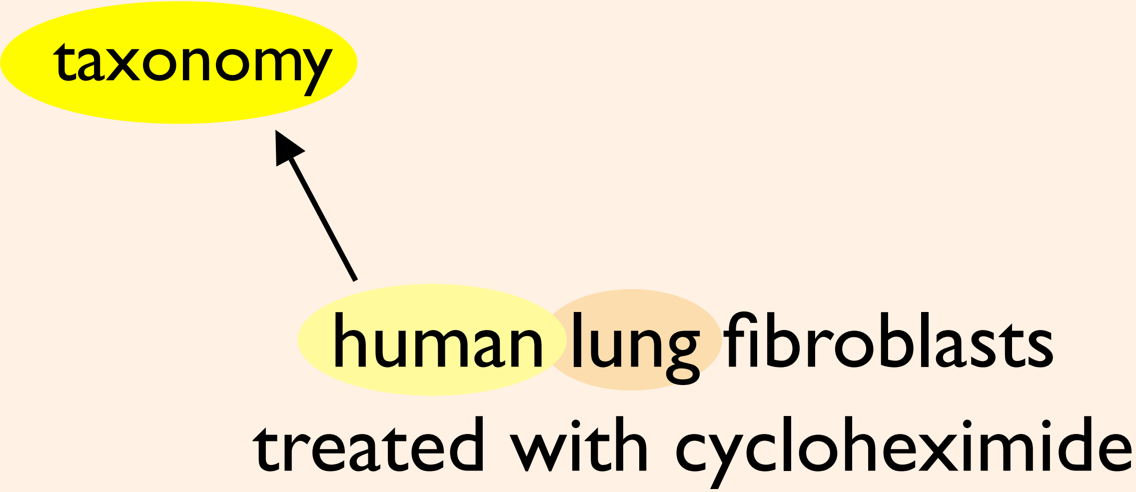
Ontologies and Experiments



taxonomy

human lung fibroblasts
treated with cycloheximide

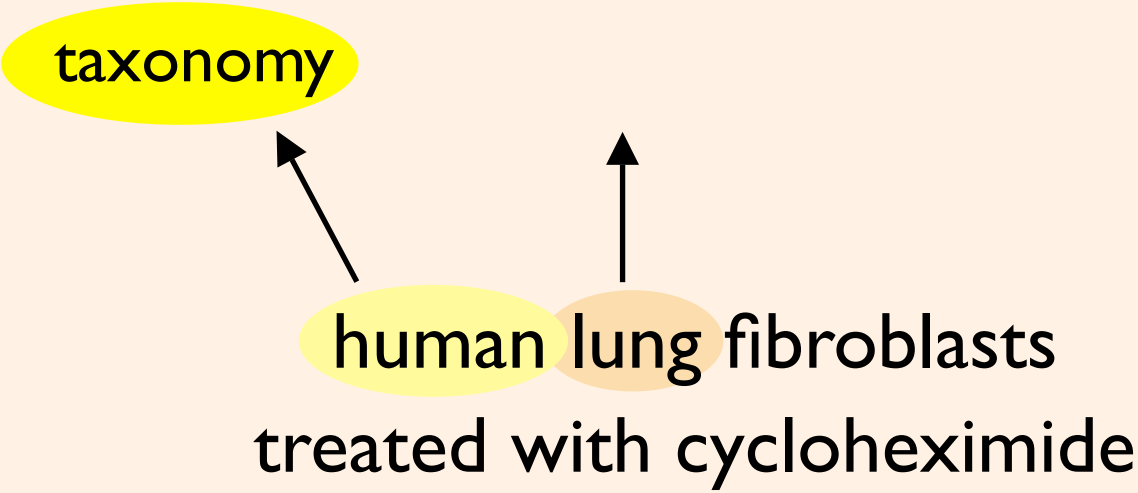
Ontologies and Experiments



taxonomy

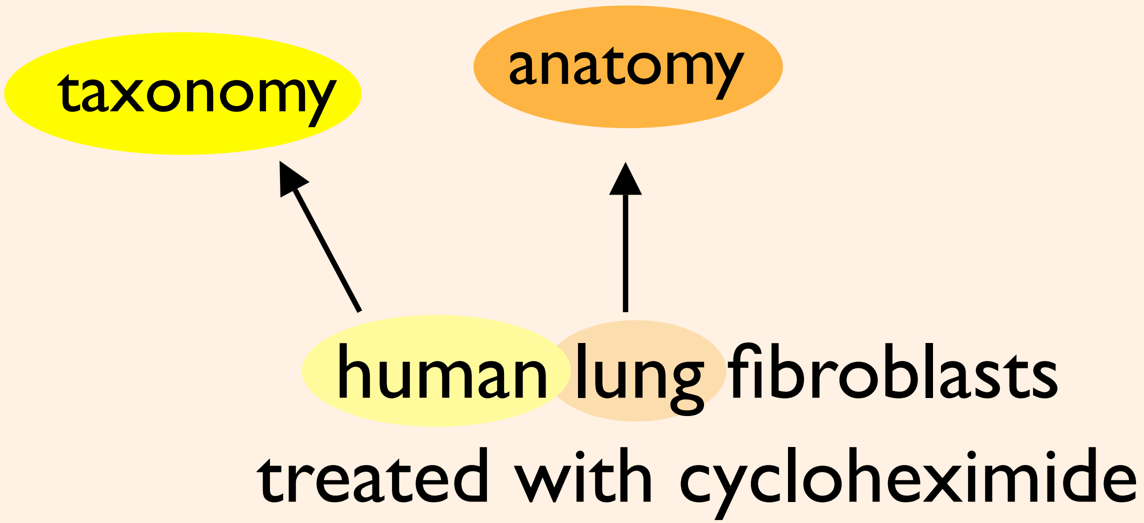
human lung fibroblasts
treated with cycloheximide

Ontologies and Experiments



human lung fibroblasts treated with cycloheximide

Ontologies and Experiments



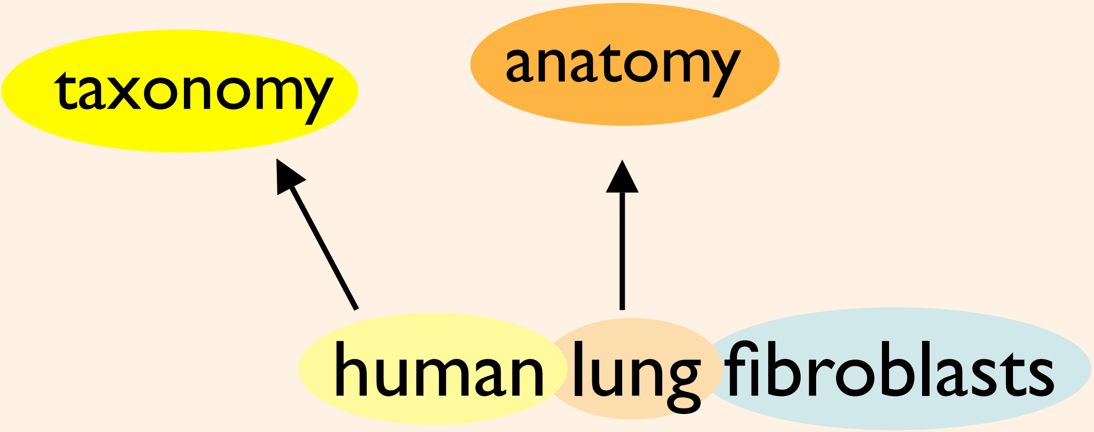
taxonomy

anatomy

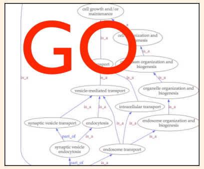
human lung fibroblasts

treated with cycloheximide

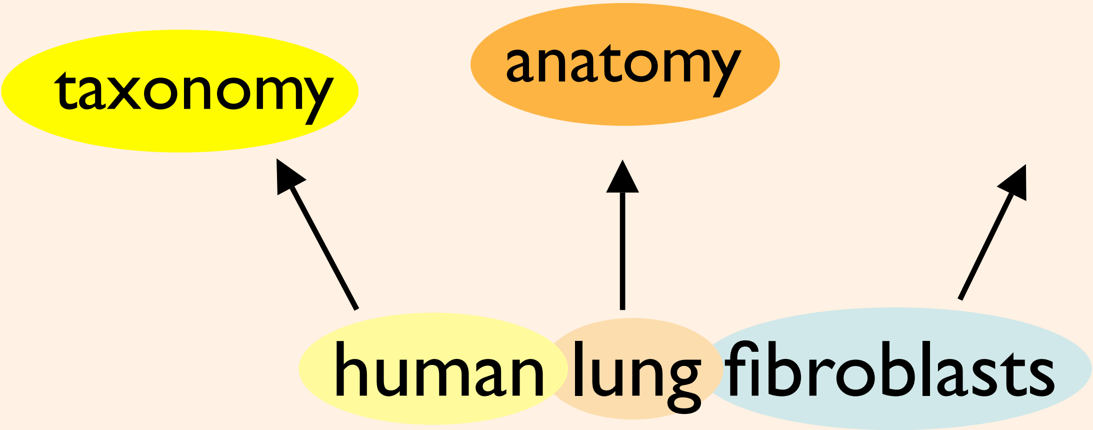
Ontologies and Experiments



human lung fibroblasts
treated with cycloheximide



Ontologies and Experiments

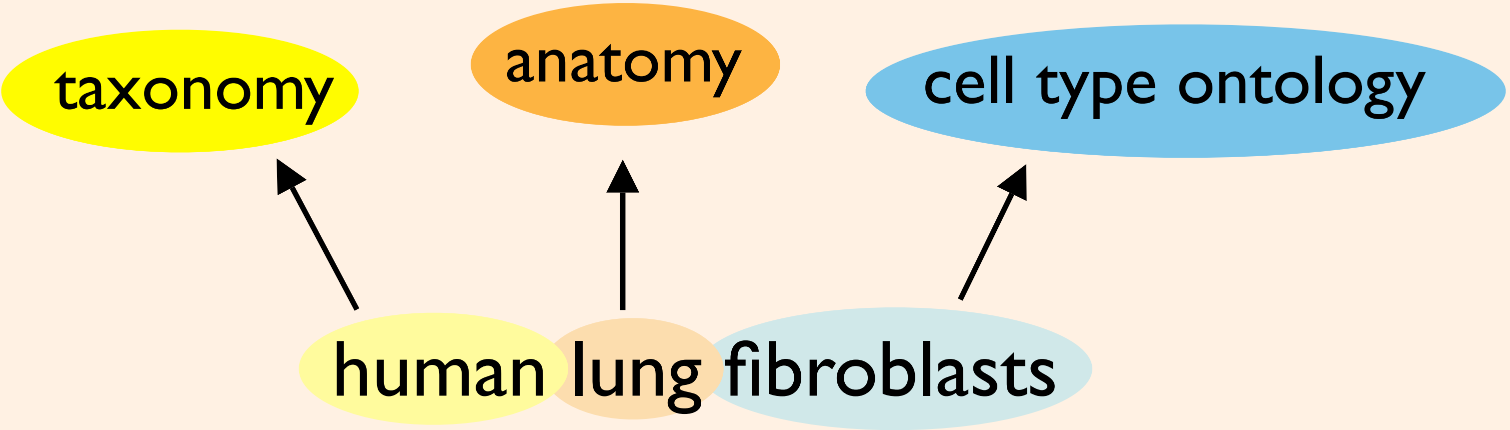

taxonomy

anatomy

human lung fibroblasts

treated with cycloheximide

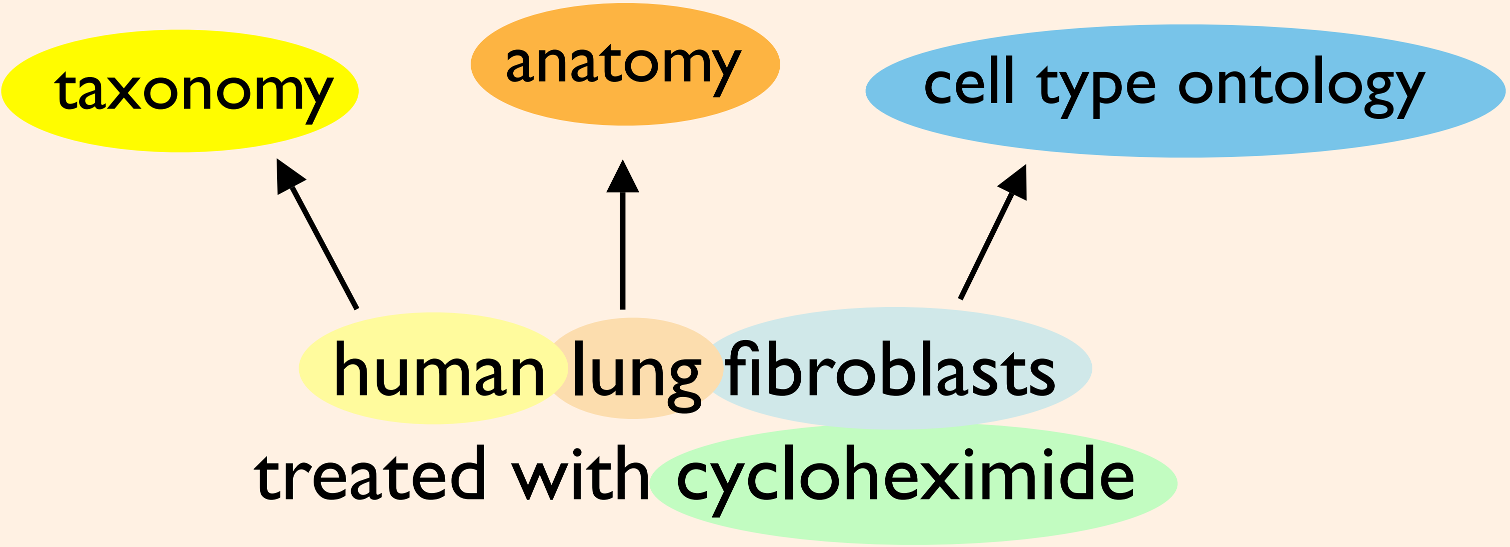
Ontologies and Experiments



human lung fibroblasts

treated with cycloheximide

Ontologies and Experiments



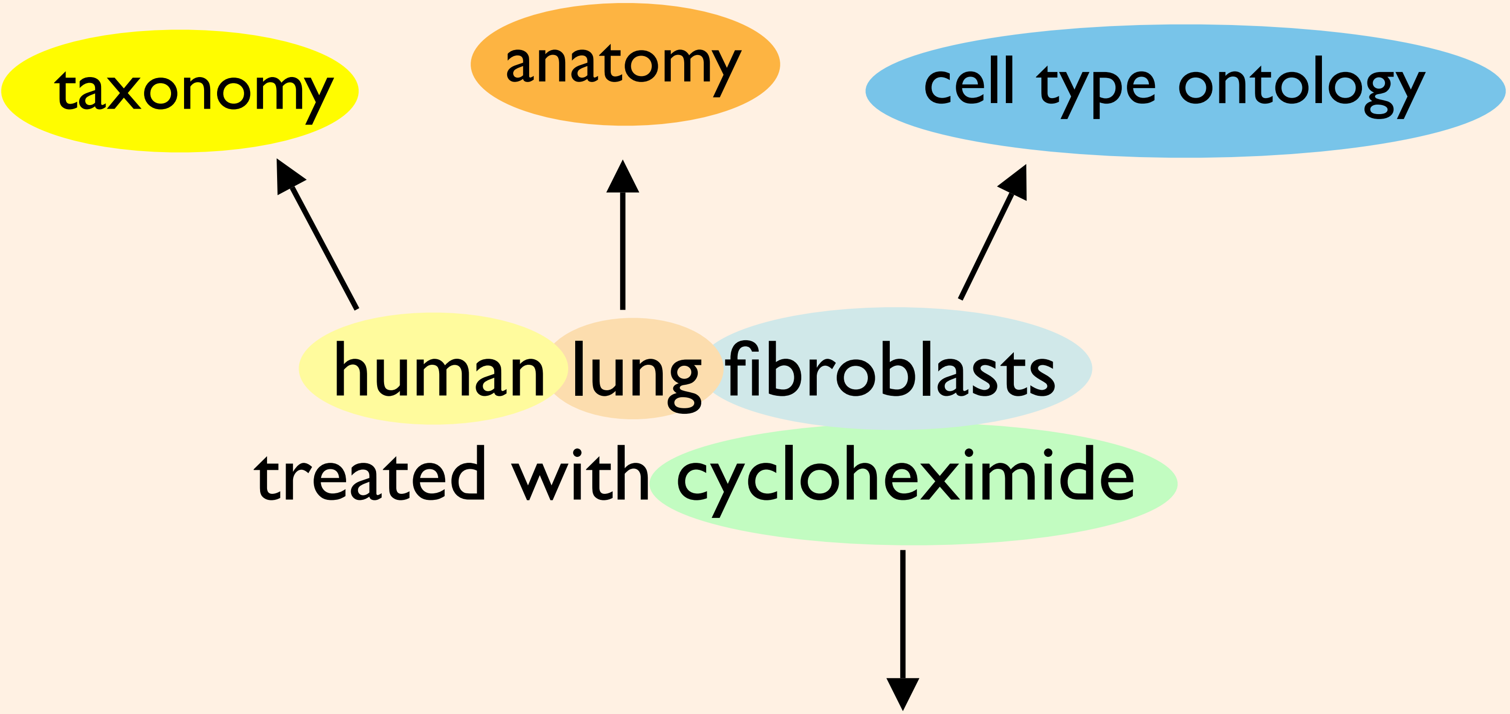
taxonomy

anatomy

cell type ontology

human lung fibroblasts
treated with cycloheximide

Ontologies and Experiments



taxonomy

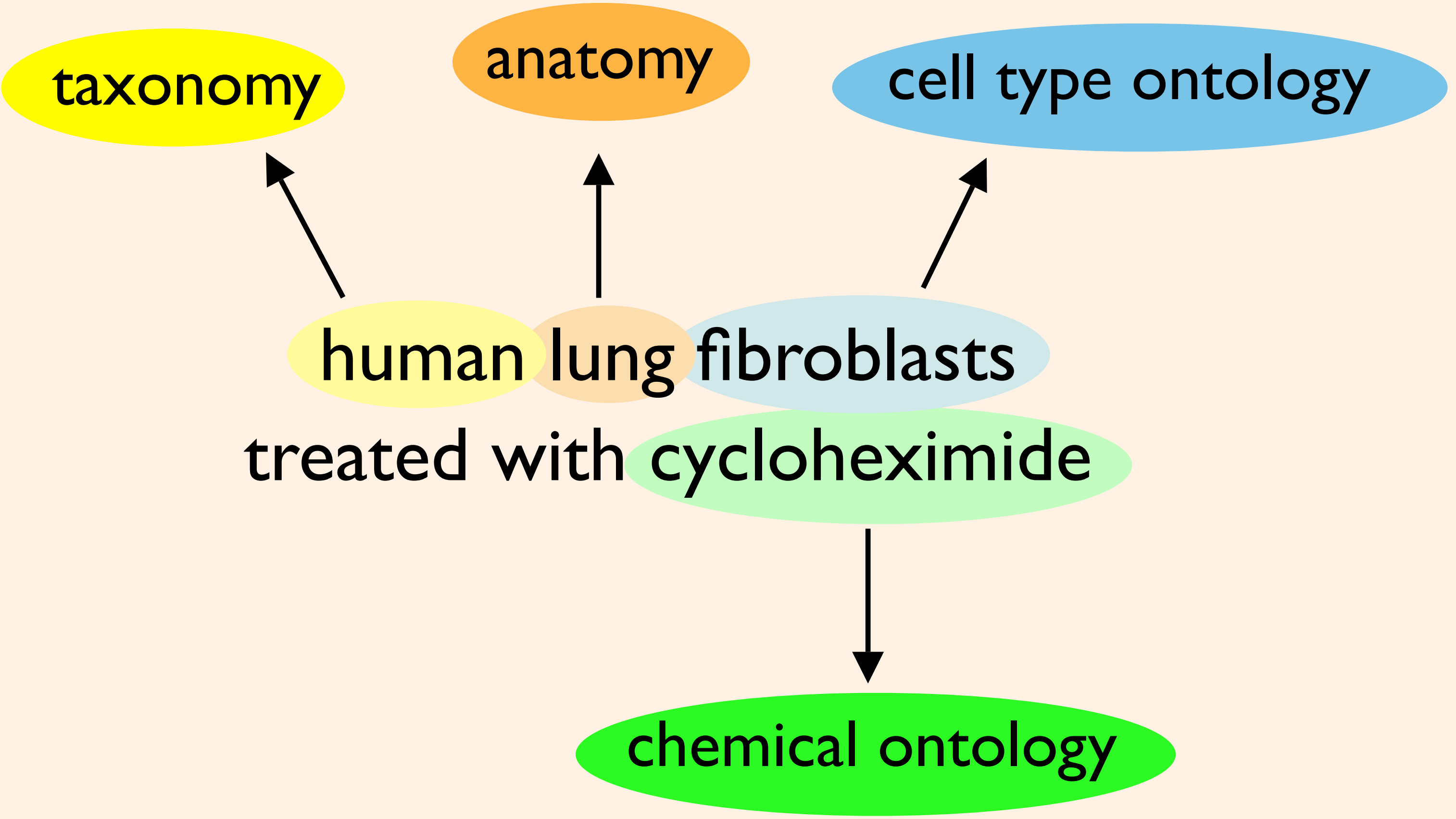
anatomy

cell type ontology

human lung fibroblasts

treated with cycloheximide

Ontologies and Experiments



taxonomy

anatomy

cell type ontology

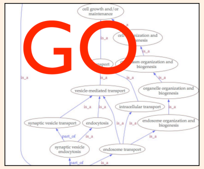
human lung fibroblasts

treated with cycloheximide

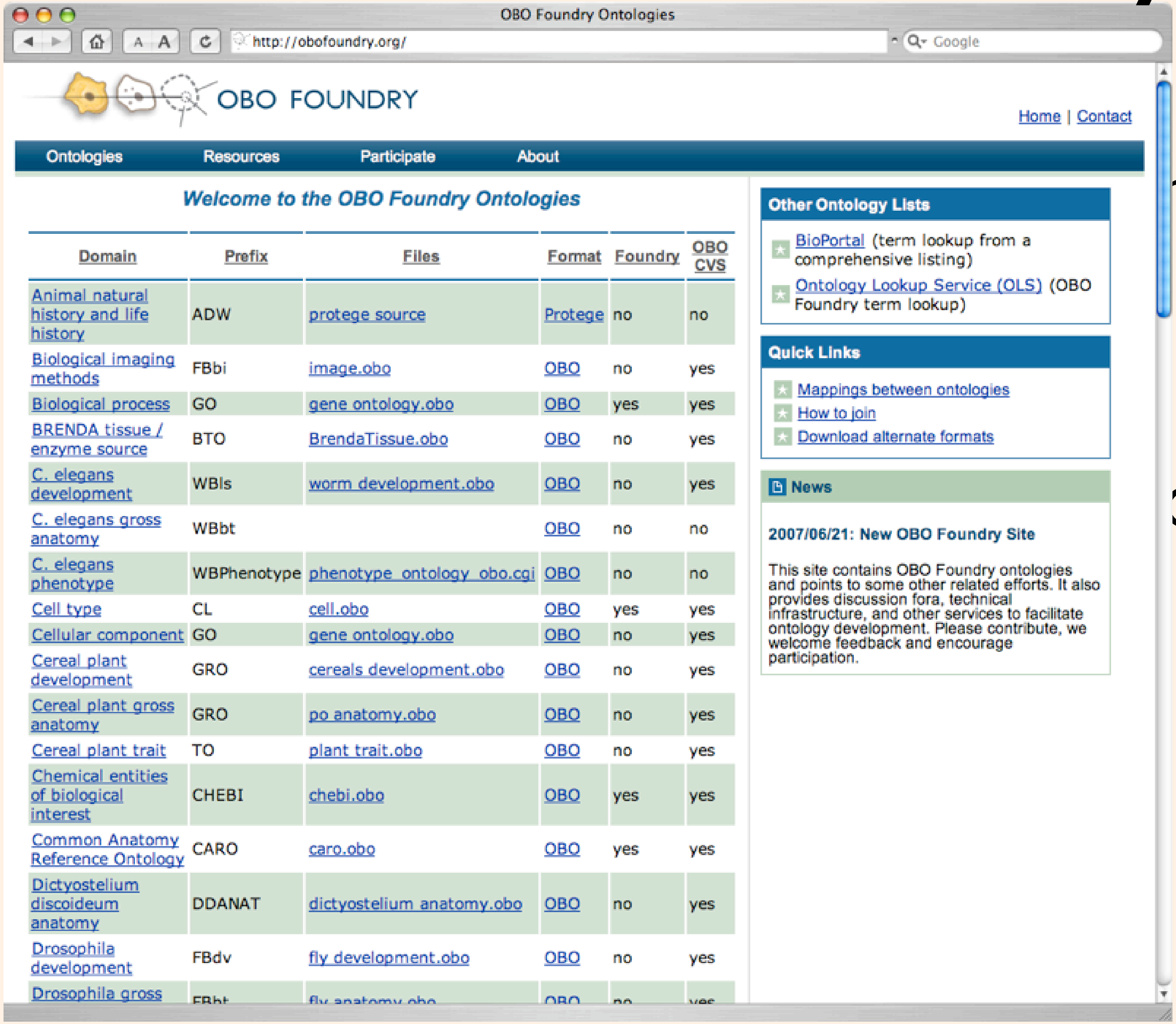
chemical ontology

OBO Foundry

- Open Biomedical Ontologies
  - Collection of shared genomics and proteomics vocabularies
  - Principles:
    - Open source
    - Common syntax (e.g. OWL or OBO)
    - Orthogonal; avoid duplication
    - Share ID space
    - Define terms
    - Involve the community



OBO Foundry



Welcome to the OBO Foundry Ontologies

Domain	Prefix	Files	Format	Foundry	OBO CVS
Animal natural history and life history	ADW	protege source	Protege	no	no
Biological imaging methods	FBbi	image.obo	OBO	no	yes
Biological process	GO	gene_ontology.obo	OBO	yes	yes
BRENDA tissue / enzyme source	BTO	BrendaTissue.obo	OBO	no	yes
C. elegans development	WBls	worm_development.obo	OBO	no	yes
C. elegans gross anatomy	WBbt		OBO	no	no
C. elegans phenotype	WBPhenotype	phenotype_ontology_obo.cgi	OBO	no	no
Cell type	CL	cell.obo	OBO	yes	yes
Cellular component	GO	gene_ontology.obo	OBO	no	yes
Cereal plant development	GRO	cereals_development.obo	OBO	no	yes
Cereal plant gross anatomy	GRO	po_anatomy.obo	OBO	no	yes
Cereal plant trait	TO	plant_trait.obo	OBO	no	yes
Chemical entities of biological interest	CHEBI	chebi.obo	OBO	yes	yes
Common Anatomy Reference Ontology	CARO	caro.obo	OBO	yes	yes
Dictyostelium discoideum anatomy	DDANAT	dictyostelium_anatomy.obo	OBO	no	yes
Drosophila development	FBdv	fly_development.obo	OBO	no	yes
Drosophila gross	FBbt	fly_anatomy.obo	OBO	no	yes

Other Ontology Lists

- BioPortal (term lookup from a comprehensive listing)
- Ontology Lookup Service (OLS) (OBO Foundry term lookup)

Quick Links

- Mappings between ontologies
- How to join
- Download alternate formats

News

2007/06/21: New OBO Foundry Site

This site contains OBO Foundry ontologies and points to some other related efforts. It also provides discussion fora, technical infrastructure, and other services to facilitate ontology development. Please contribute, we welcome feedback and encourage participation.

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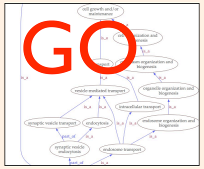

OBO Foundry

OBO FOUNDRY

Home | Contact

Ontologies | Resources | Participate | About

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C. elegans gross anatomy	WBbt		OBO	no	no
C. elegans phenotype	WBPhenotype	phenotype ontology obo.cgi	OBO	no	no
Cell type	CL	cell.obo	OBO	yes	yes
Cellular component	GO	gene ontology.obo	OBO	no	yes
Cereal plant development	GRO	cereals development.obo	OBO	no	yes
Cereal plant gross anatomy	GRO	po anatomy.obo	OBO	no	yes
Cereal plant trait	TO	plant trait.obo	OBO	no	yes
Chemical entities of biological interest	CHEBI	chebi.obo	OBO	yes	yes
Common Anatomy Reference Ontology	CARO	caro.obo			
Dictyostelium discoideum anatomy	DDANAT	dictyostelium anatomy.obo	OBO	no	yes
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obofoundry.org

OBO Foundry

Welcome to the OBO Fou

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Dictyostelium discoideum anatomy	DDANAT	dictyostelium a
Drosophila development	FBdv	fly developmen
Drosophila gross	FBbt	fly anatomy.ob

OBO Download Matrix

This page is for downloading OBO ontologies in a variety of formats, and for downloading reports from each of the ontologies. The data below is derived automatically from the primary sources, available from the main OBO website. The status of this page is EXPERIMENTAL. See below for caveats.

This page will be retired when the current functionality is subsumed by the BioPortal resource developed by the NCBO

- Ontologies
- Logical Definitions
- Mappings
- Format Guide
- Metadata
- Reports
- FTP Archive
- Statistics

Ontologies

Ontology	ID Prefix	Files
adult_mouse_anatomy:**Mouse adult gross anatomy**	MA	obo_xml[595 kb] -- obo[285 kb] -- go_ont[169 kb] -- owl[1311 kb] -- chadoxml[1614 kb] -- godb_prestore[1498 kb] -- rdf[830 kb] -- tbl[93 kb] -- prolog[676 kb] -- validation_report[2 kb] -- stats[2 kb] -- pellet_report[0 kb] -- obo.html[1737 kb] --
animalhist:**Animal natural history and life history**	ADW	
biological_process:**Biological process**	GO	obo_xml[19969 kb] -- obo[6909 kb] -- obo_xml[12263 kb] -- obo[6909 kb] -- go_ont[2987 kb] -- owl[25382 kb] -- chadoxml[31966 kb] -- godb_prestore[26749 kb] -- rdf[11461 kb] -- tbl[4304 kb] -- prolog[13859 kb] - validation_report[0 kb] -- stats[2 kb] -- pellet_report[0 kb] -- o.html[26513 kb] --
bre		o_xml[0 kb] -- obo[2 kb] -- owl[2 kb] -- chadoxml[2 kb] -- odb_prestore[1 kb] -- rdf[0 kb] -- prolog[0 kb] -- validation_report[0 kb] -- ats[0 kb] -- pellet_report[0 kb] -- obo.html[0 kb] --
caro:**Common Anatomy Reference Ontology**	CARO	obo_xml[18 kb] -- obo[10 kb] -- go_ont[2 kb] -- owl[54 kb] -- chadoxml[47 kb] -- godb_prestore[37 kb] -- rdf[20 kb] -- tbl[6 kb] -- prolog[18 kb] -- validation_report[0 kb] -- stats[2 kb] -- pellet_report[14 kb] -- obo.html[44 kb] --
cell:**Cell type**	CL	obo_xml[351 kb] -- obo[200 kb] -- go_ont[66 kb] -- owl[783 kb] -- chadoxml[978 kb] -- godb_prestore[830 kb] -- rdf[409 kb] -- tbl[102 kb] --

obofoundry.org

OBO Foundry

Welcome to the OBO Fou

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Drosophila gross	FBbt	fly anatomy.ob

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Ontologies
Logical Definitions
Mappings
Format Guide
Metadata
Reports
FTP Archive
Statistics

Ontologies

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animalhist:Animal natural history and life history	ADW	
biological_process:Biological process	GO	obo_xml[19969 kb] -- obo[6909 kb] -- obo_xml[12263 kb] -- obo[6909 kb] -- go_ont[2987 kb] -- owl[25382 kb] -- chadoxml[31966 kb] -- godb_prestore[26749 kb] -- rdf[11461 kb] -- tbl[4304 kb] -- prolog[13859 kb] - validation_report[0 kb] -- stats[2 kb] -- pellet_report[0 kb] -- bo.html[26513 kb] --
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caro:Common Anatomy Reference Ontology	CARO	obo_xml[18 kb] -- obo[10 kb] -- go_ont[2 kb] -- owl[54 kb] -- chadoxml[47 kb] -- godb_prestore[37 kb] -- rdf[20 kb] -- tbl[6 kb] -- prolog[18 kb] -- validation_report[0 kb] -- stats[2 kb] -- pellet_report[14 kb] -- obo.html[44 kb] --
cell:Cell type	CL	obo_xml[351 kb] -- obo[200 kb] -- go_ont[66 kb] -- owl[783 kb] -- chadoxml[978 kb] -- godb_prestore[830 kb] -- rdf[409 kb] -- tbl[102 kb] --

obofoundry.org

Cross-products in GO

- Explicit links between one GO term and another ontology term

Many GO terms are *implicitly* composite

- cysteine biosynthesis
- myoblast fusion
- ATP synthesis coupled proton transport (ChEBI)
- snoRNA catabolism
- wing disc pattern formation
- epidermal cell differentiation
- regulation of flower development
- interleukin-18 receptor complex
- B-cell differentiation
- dorsal ectoderm

slide credit: Chris Mungall, BBOP

Opaque references to other **OBO** ontologies

- cysteine biosynthesis (ChEBI)
- myoblast fusion (Cell Type Ontology)
- ATP synthesis coupled proton transport (ChEBI)
- snoRNA catabolism (Sequence Ontology)
- wing disc pattern formation (*Drosophila* anatomy)
- epidermal cell differentiation (Cell Type Ontology)
- regulation of flower development (*Plant* anatomy)
- interleukin-18 receptor complex (not yet in OBO)
- B-cell differentiation (Cell Type Ontology)
- dorsal ectoderm (Spatial Qualifiers)

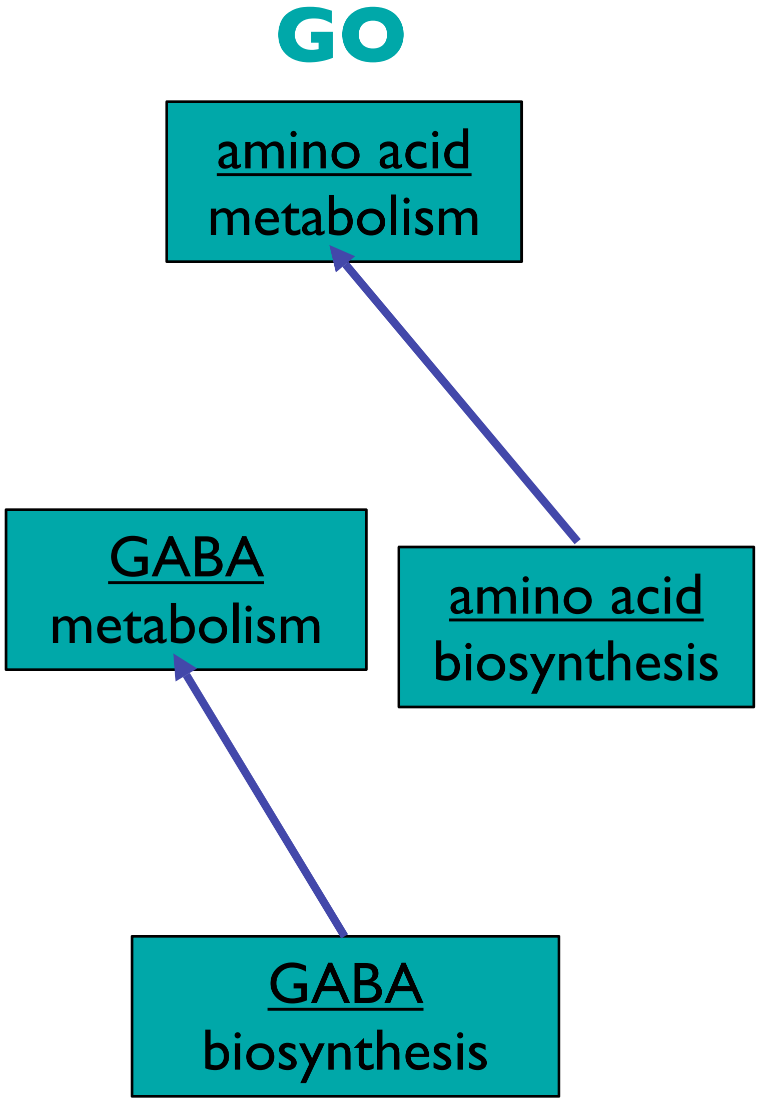
slide credit: Chris Mungall, BBOP

Methodology

- Make opaque references explicit
- Use OBO ontologies
  - ChEBI
  - Cell
  - PATO (qualities)
  - Anatomy/CARO

slide credit: Chris Mungall, BBOP

GO: now



GO

amino acid metabolism

GABA metabolism

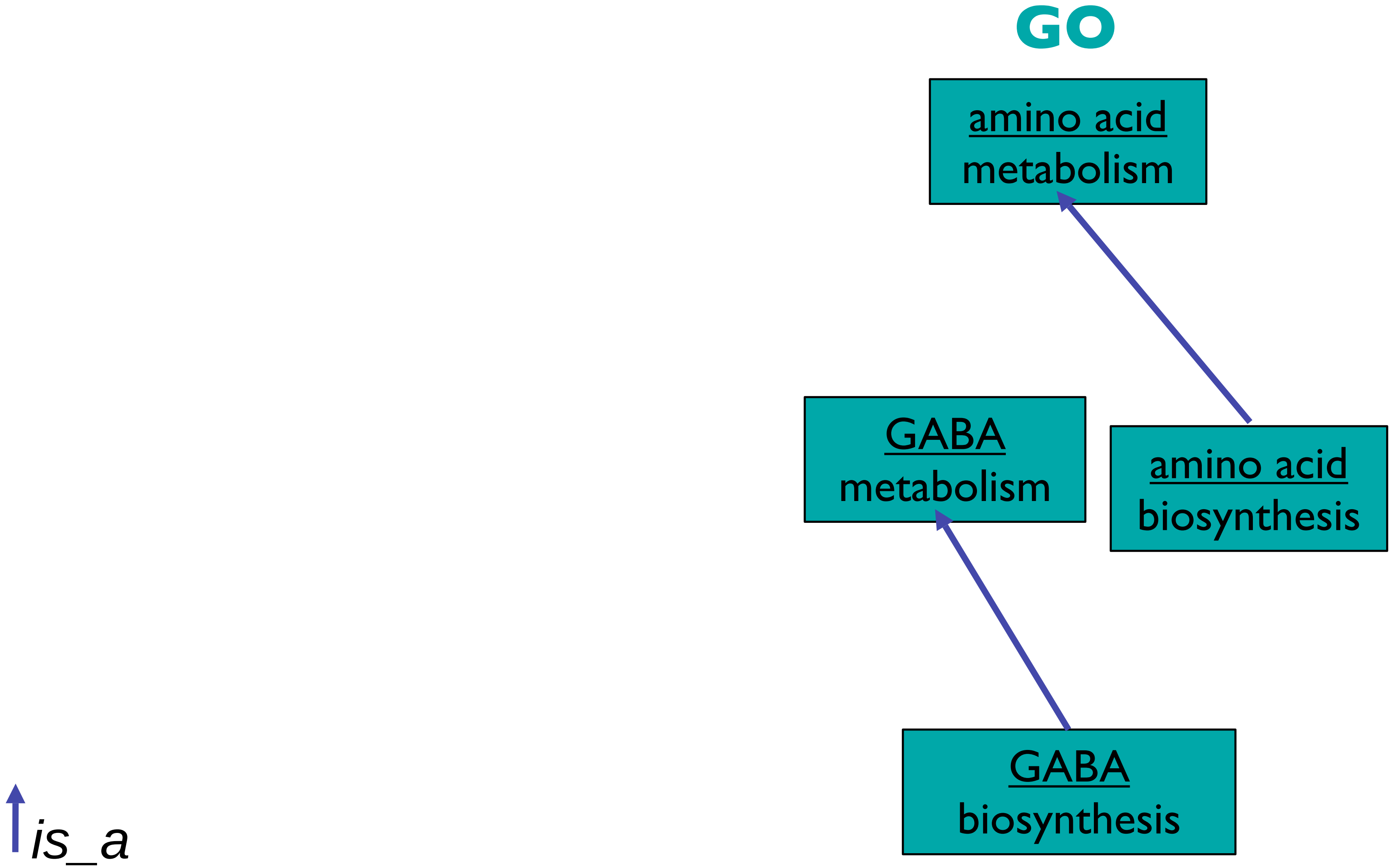
amino acid biosynthesis

GABA biosynthesis

↑ *is_a*

slide credit: Chris Mungall, BBOP

GO+ChEBI: now



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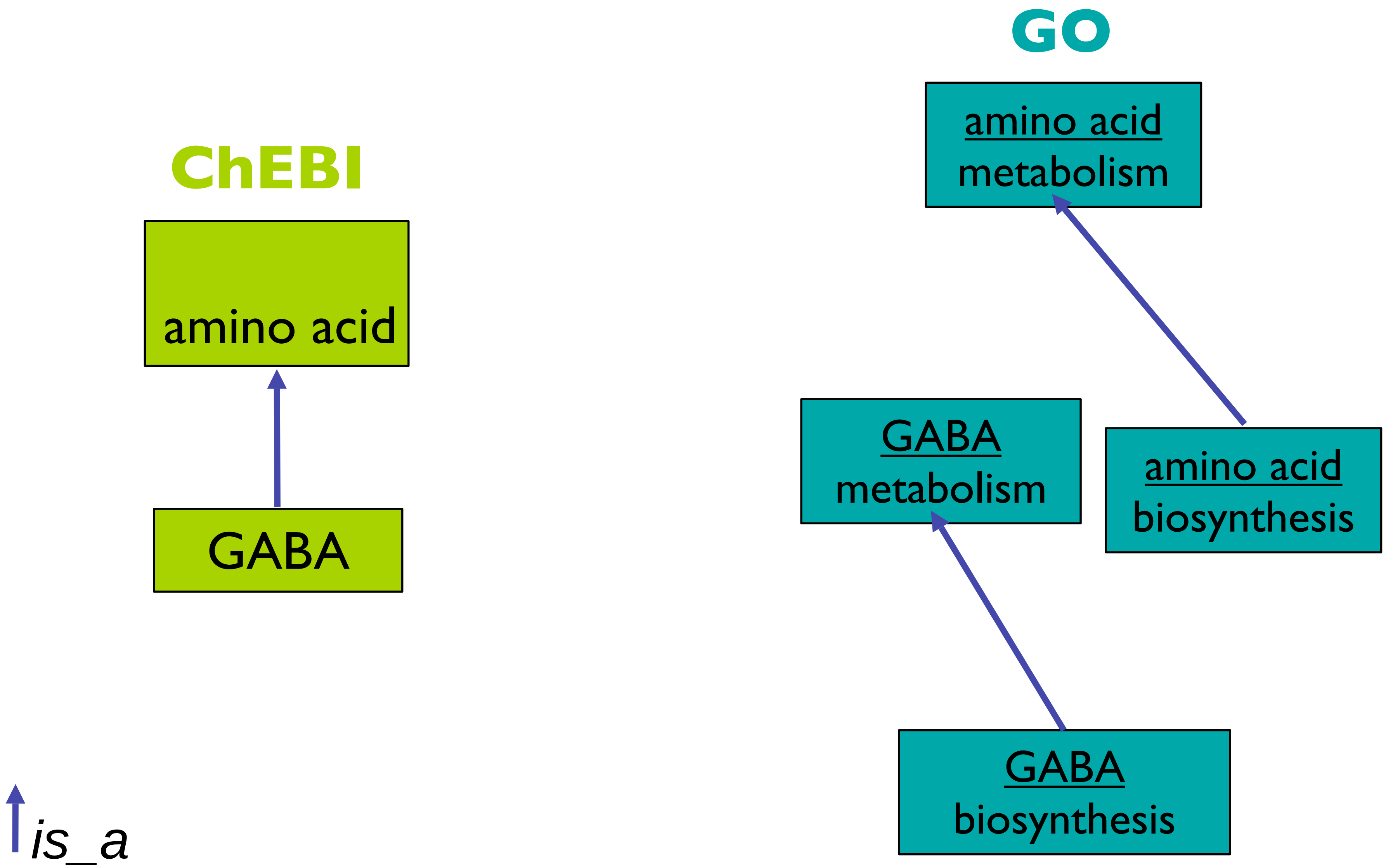
GO+ChEBI: now



ChEBI

amino acid

GABA

GO

amino acid metabolism

GABA metabolism

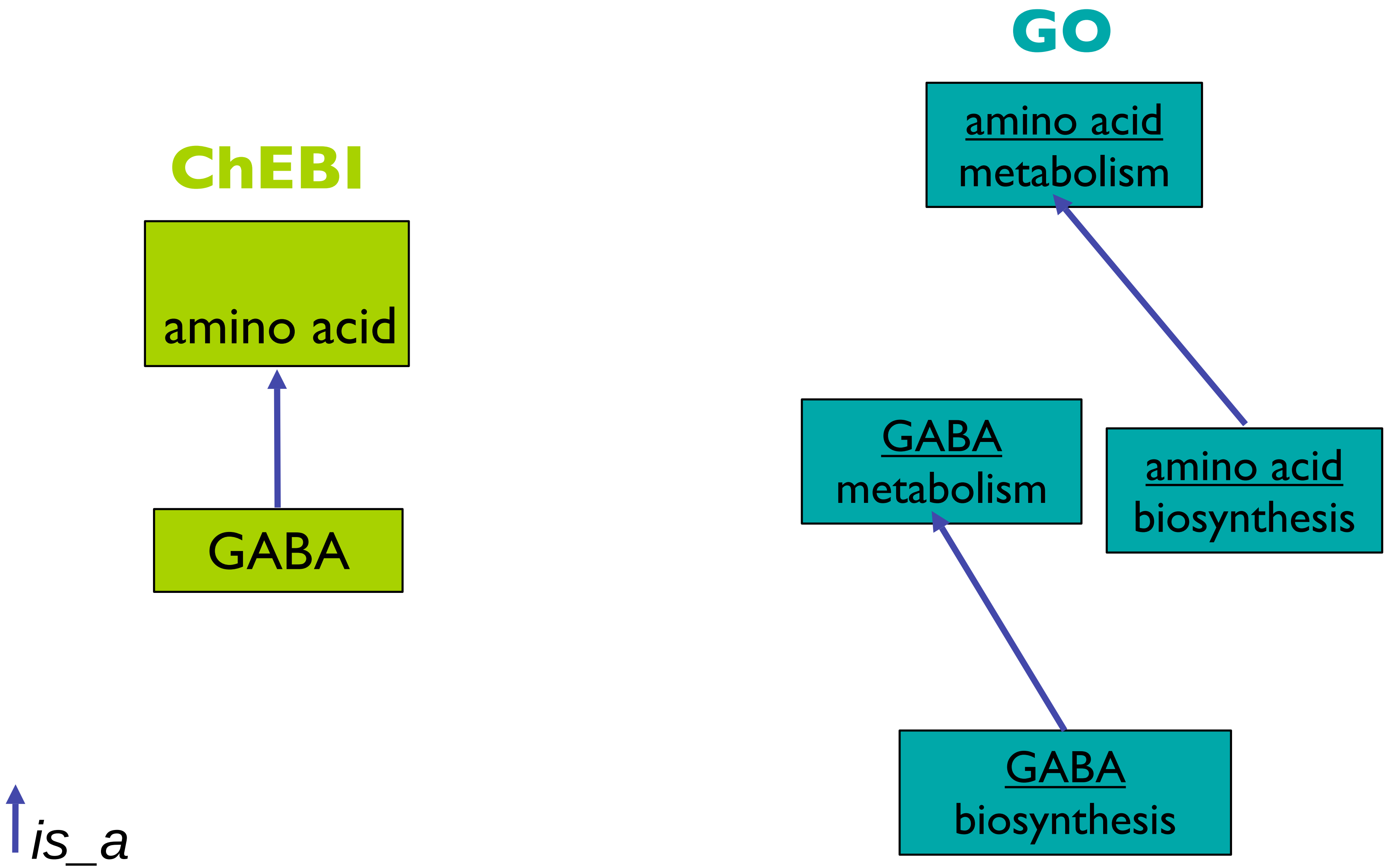
amino acid biosynthesis

GABA biosynthesis

is_a

slide credit: Chris Mungall, BBOP

Linking GO to ChEBI



GO

amino acid metabolism

GABA metabolism

amino acid biosynthesis

GABA biosynthesis

ChEBI

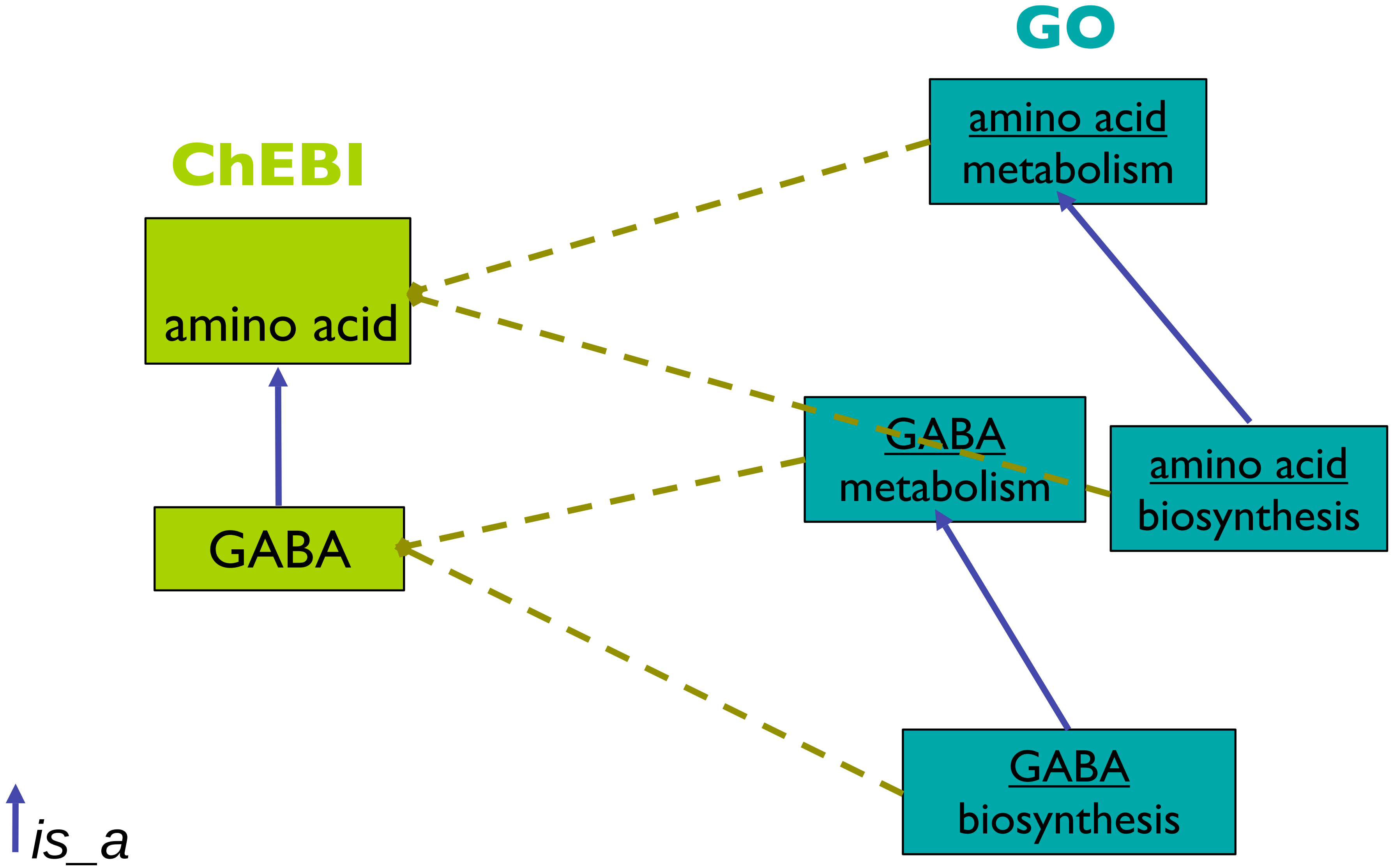
amino acid

GABA

is_a

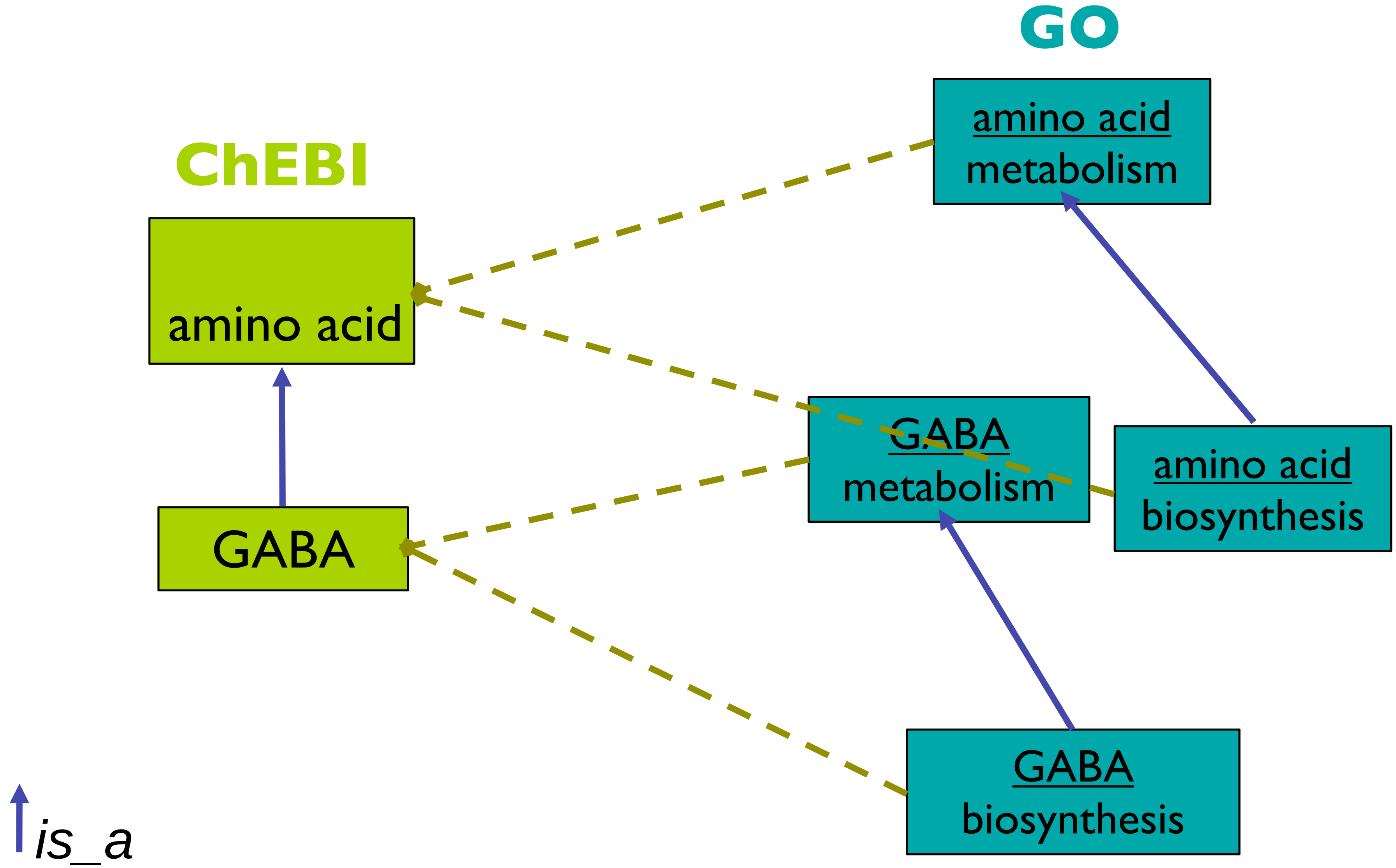
slide credit: Chris Mungall, BBOP

Linking GO to ChEBI



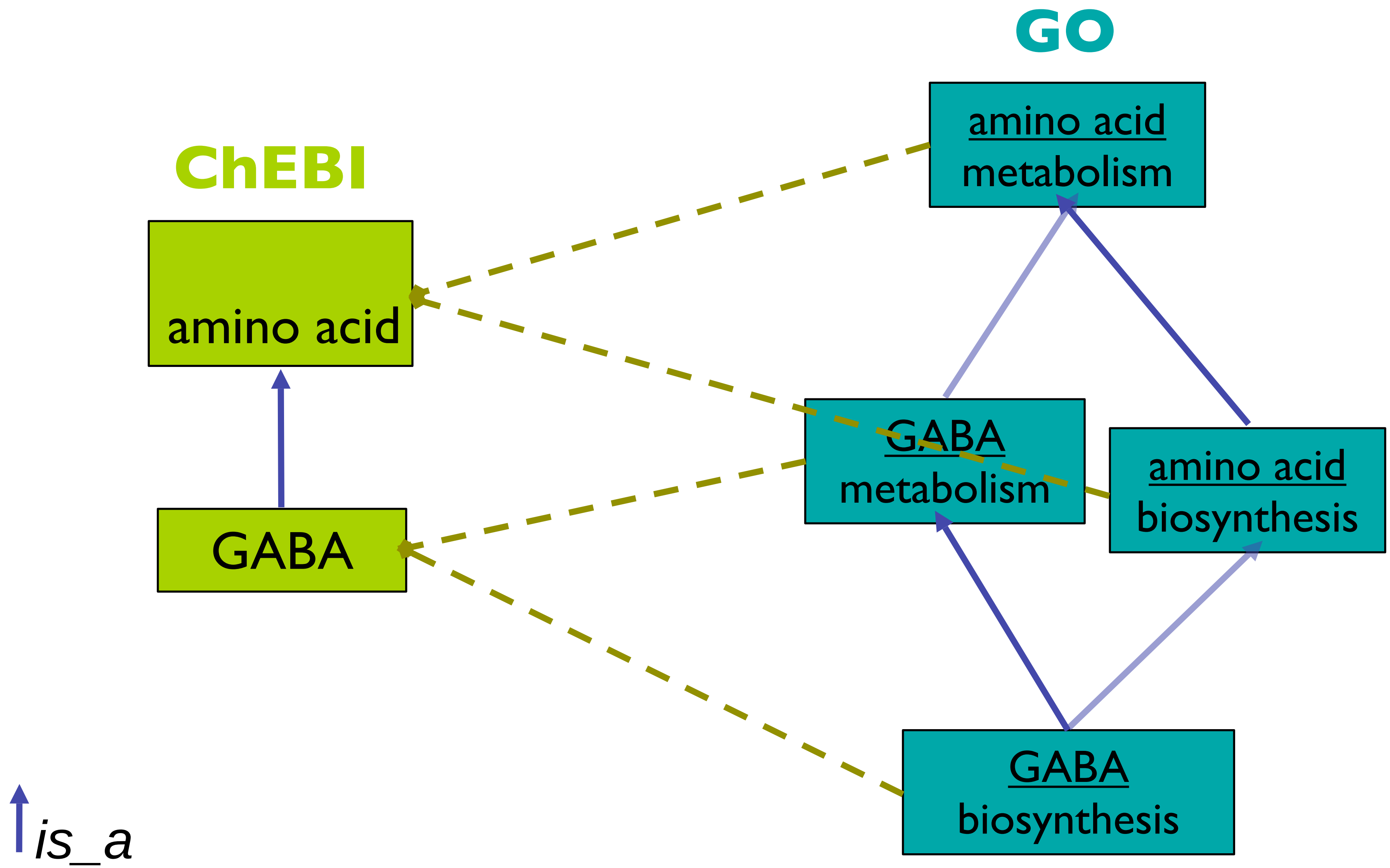
slide credit: Chris Mungall, BBOP

Can we find missing links?



slide credit: Chris Mungall, BBOP

Can we find missing links?



GO

amino acid metabolism

ChEBI

amino acid

GABA metabolism

amino acid biosynthesis

GABA

GABA biosynthesis

is_a

slide credit: Chris Mungall, BBOP

Simple DAGs are not enough

- We want to define GO terms using ChEBI terms
- DAGs give necessary conditions
- We want to give necessary <u>and sufficient</u> conditions
- Solution:
  - Genus-differentia definitions
  - Aka cross-products, aka intersections

slide credit: Chris Mungall, BBOP

Example genus-differentia definition

- A **GABA metabolic process** is:
  - A **metabolic process**
    - That *has_participant* **GABA**
- This is saying more than simply adding a DAG link between GABA metabolic process and GABA
  - It allows us to do powerful reasoning
    - Keeps ontologies in sync

slide credit: Chris Mungall, BBOP

```
[Term]
id: GO:0009447
name: putrescine catabolic process
intersection_of: GO:0009056 ! catabolism
intersection_of: results_in_division_of CHEBI:17148 ! putrescine

[Term]
id: GO:0009448
name: gamma-aminobutyric acid metabolic process
intersection_of: GO:0008152 ! metabolism
intersection_of: has_participant CHEBI:16865 ! gamma-aminobutyric acid

[Term]
id: GO:0009449
name: gamma-aminobutyric acid biosynthetic process
intersection_of: GO:0009058 ! biosynthesis
intersection_of: results_in_change_to CHEBI:16865 ! gamma-aminobutyric acid

[Term]
id: GO:0009450
name: gamma-aminobutyric acid catabolic process
intersection_of: GO:0009056 ! catabolism
intersection_of: results_in_division_of CHEBI:16865 ! gamma-aminobutyric acid
```

slide credit: Chris Mungall, BBOP

GO and Phenotypes

- Phenotype annotation uses GO — and other OBO ontologies — in combination with qualities from PATO
- An *entity* (GO, anatomy, etc. term) has a *quality* (PATO term)
- Example: decreased rate of sucrose synthesis

GO and Phenotypes

- Phenotype annotation uses GO — and other OBO ontologies — in combination with qualities from PATO
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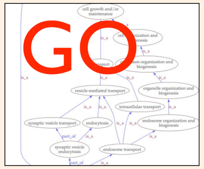
GO:0005986

GO and Phenotypes

- Phenotype annotation uses GO — and other OBO ontologies — in combination with qualities from PATO
- An *entity* (GO, anatomy, etc. term) has a *quality* (PATO term)
- Example: decreased rate of sucrose synthesis

PATO:0000911

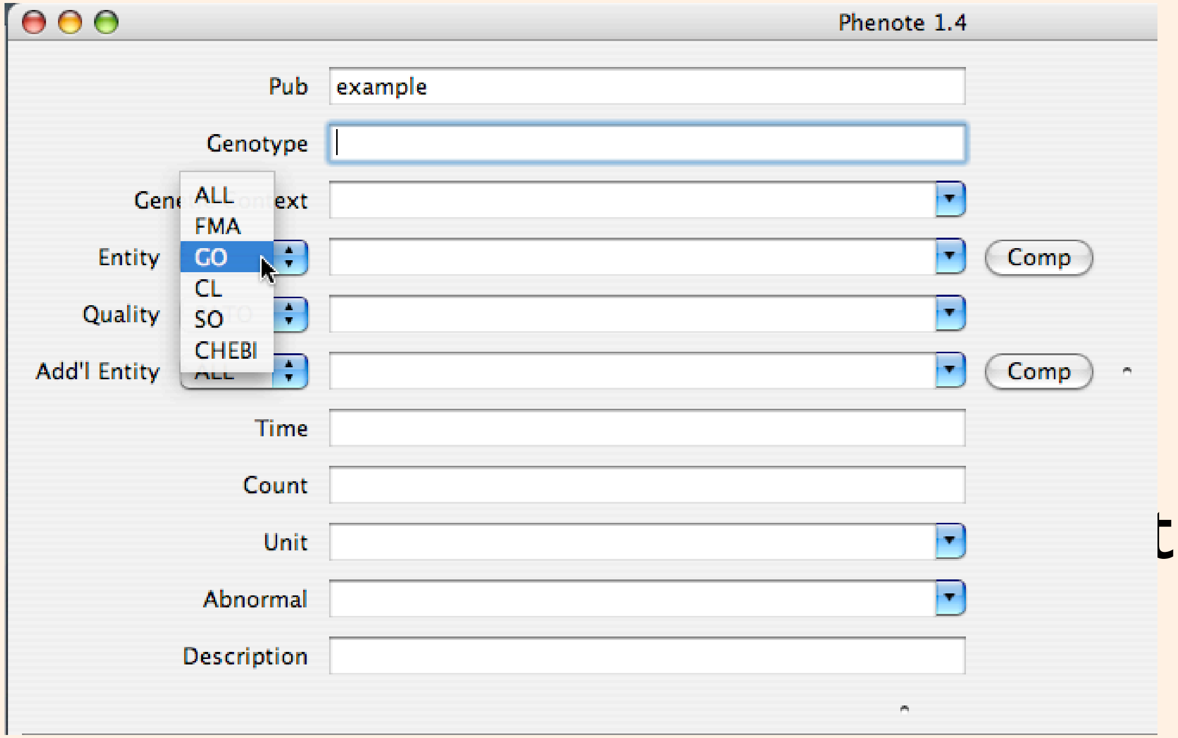
GO:0005986



Phenote

- Java application for annotating phenotypes using biological ontologies
- Uses any ontology in OBO format

Phenote

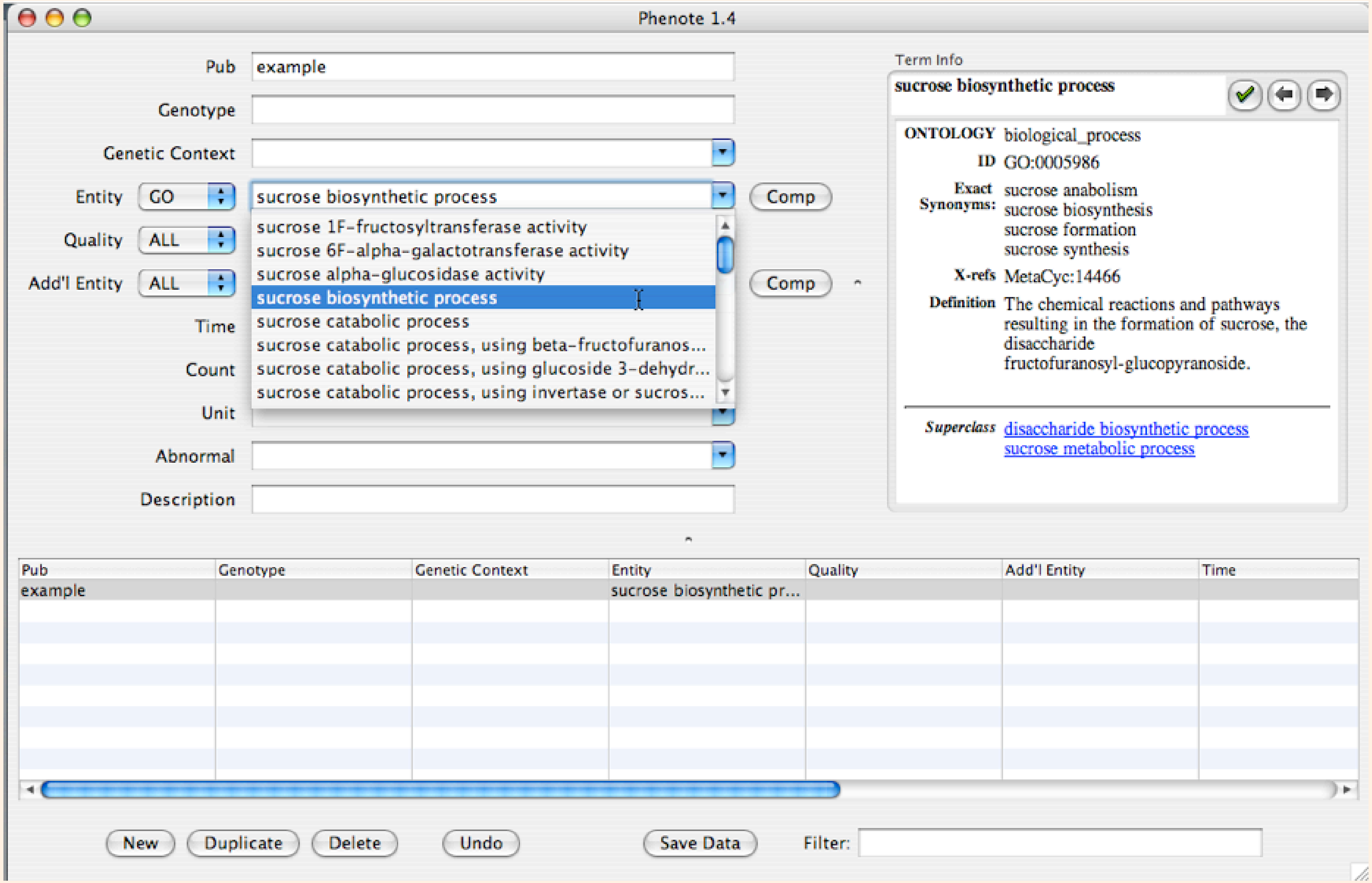


Pub: example

ting phenotypes

- Uses any ontology in OBO format

Phenote



Pub: example
Genotype:
Genetic Context:
Entity: GO — sucrose biosynthetic process
Quality: ALL
Add'l Entity: ALL
Time:
Count:
Unit:
Abnormal:
Description:

- sucrose 1F-fructosyltransferase activity
- sucrose 6F-alpha-galactotransferase activity
- sucrose alpha-glucosidase activity
- **sucrose biosynthetic process**
- sucrose catabolic process
- sucrose catabolic process, using beta-fructofuranos...
- sucrose catabolic process, using glucoside 3-dehydr...
- sucrose catabolic process, using invertase or sucros...

Term Info

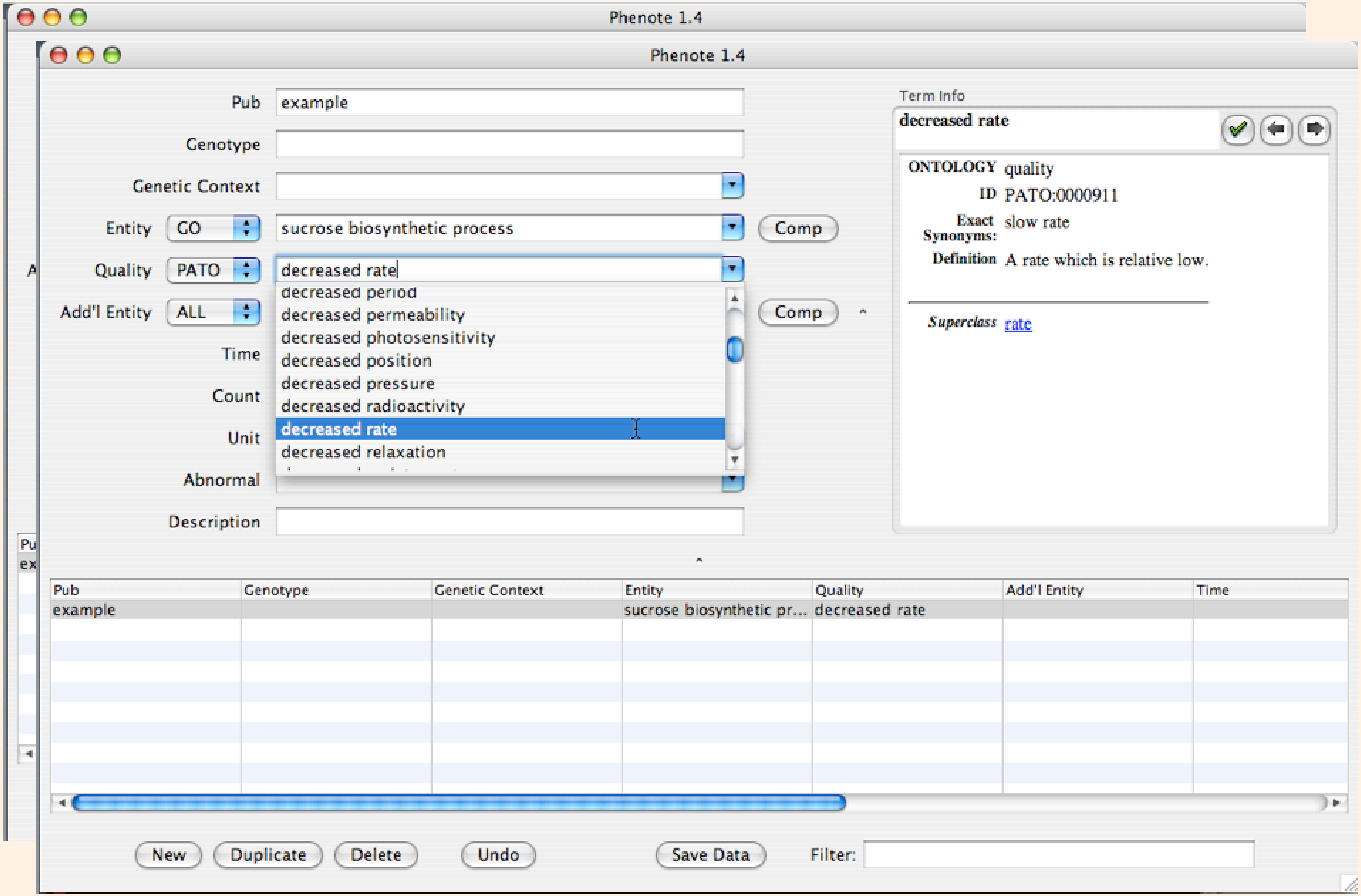
sucrose biosynthetic process

ONTOLOGY	biological_process
ID	GO:0005986
Exact Synonyms:	sucrose anabolism sucrose biosynthesis sucrose formation sucrose synthesis
X-refs	MetaCyc:14466
Definition	The chemical reactions and pathways resulting in the formation of sucrose, the disaccharide fructofuranosyl-glucopyranoside.
Superclass	disaccharide biosynthetic process sucrose metabolic process

Pub	Genotype	Genetic Context	Entity	Quality	Add'l Entity	Time
example			sucrose biosynthetic pr...			

Filter:

Phenote



Phenote 1.4

Pub: example

Genotype:

Genetic Context:

Entity: GO — sucrose biosynthetic process

Quality: PATO — decreased rate

- decreased period
- decreased permeability
- decreased photosensitivity
- decreased position
- decreased pressure
- decreased radioactivity
- decreased rate
- decreased relaxation

Add'l Entity: ALL

Time:

Count:

Unit:

Abnormal:

Description:

Term Info

decreased rate

ONTOLOGY quality

ID PATO:0000911

Exact Synonyms: slow rate

Definition A rate which is relative low.

Superclass rate

Pub	Genotype	Genetic Context	Entity	Quality	Add'l Entity	Time
example			sucrose biosynthetic pr...	decreased rate		

Filter:

Acknowledgements

- The GO Editorial Office
    - Jane Lomax
    - Amelia Ireland
    - Jennifer Deegan
- GO Consortium members
- Funding:

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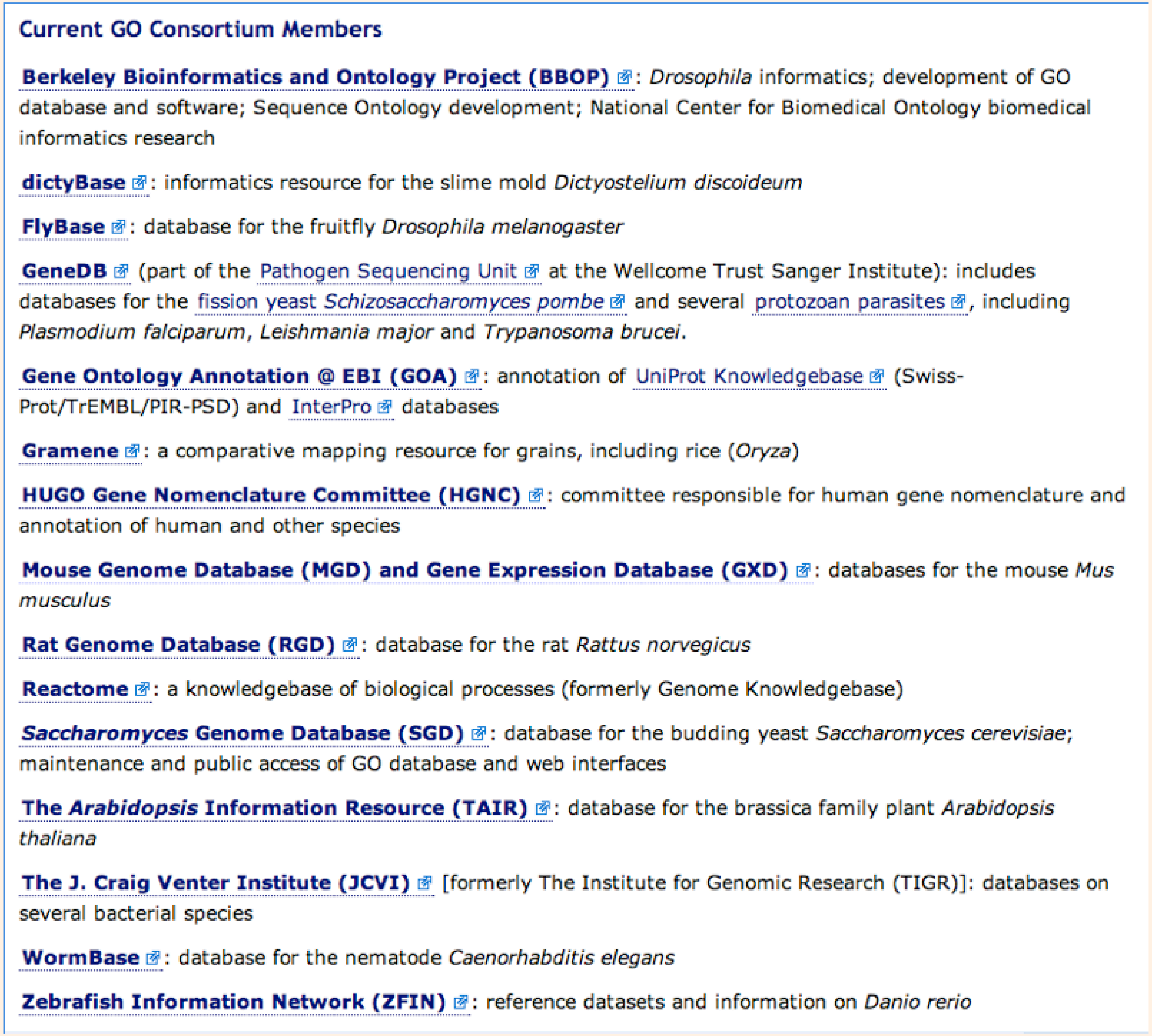


Current GO Consortium Members

Berkeley Bioinformatics and Ontology Project (BBOP): *Drosophila* informatics; development of GO database and software; Sequence Ontology development; National Center for Biomedical Ontology biomedical informatics research

dictyBase: informatics resource for the slime mold *Dictyostelium discoideum*

FlyBase: database for the fruitfly *Drosophila melanogaster*

GeneDB (part of the Pathogen Sequencing Unit at the Wellcome Trust Sanger Institute): includes databases for the fission yeast *Schizosaccharomyces pombe* and several protozoan parasites, including *Plasmodium falciparum*, *Leishmania major* and *Trypanosoma brucei*.

Gene Ontology Annotation @ EBI (GOA): annotation of UniProt Knowledgebase (Swiss-Prot/TrEMBL/PIR-PSD) and InterPro databases

Gramene: a comparative mapping resource for grains, including rice (*Oryza*)

HUGO Gene Nomenclature Committee (HGNC): committee responsible for human gene nomenclature and annotation of human and other species

Mouse Genome Database (MGD) and Gene Expression Database (GXD): databases for the mouse *Mus musculus*

Rat Genome Database (RGD): database for the rat *Rattus norvegicus*

Reactome: a knowledgebase of biological processes (formerly Genome Knowledgebase)

***Saccharomyces* Genome Database (SGD)**: database for the budding yeast *Saccharomyces cerevisiae*; maintenance and public access of GO database and web interfaces

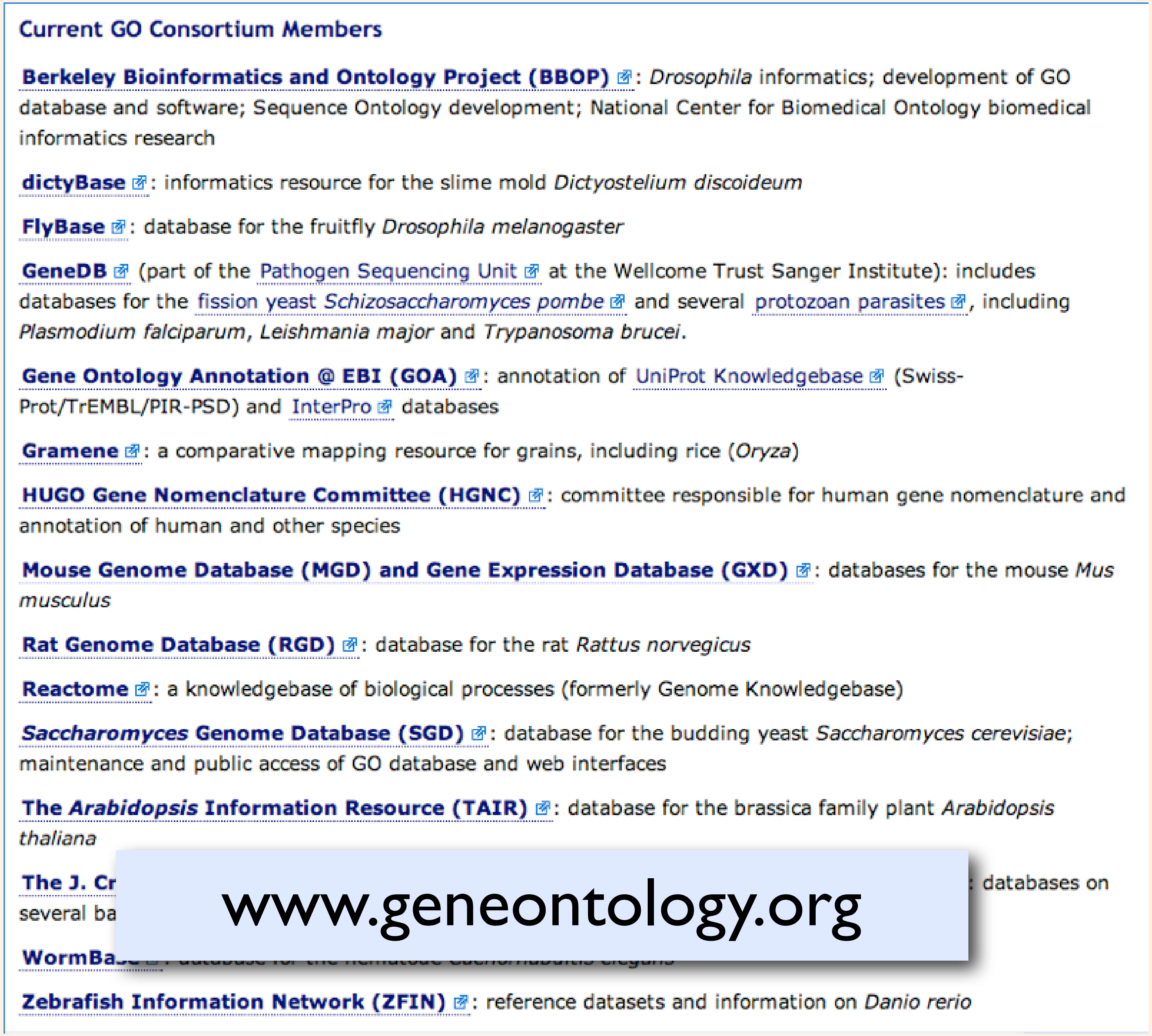
The *Arabidopsis* Information Resource (TAIR): database for the brassica family plant *Arabidopsis thaliana*

The J. Craig Venter Institute (JCVI) [formerly The Institute for Genomic Research (TIGR)]: databases on several bacterial species

WormBase: database for the nematode *Caenorhabditis elegans*

Zebrafish Information Network (ZFIN): reference datasets and information on *Danio rerio*

The GO Consortium



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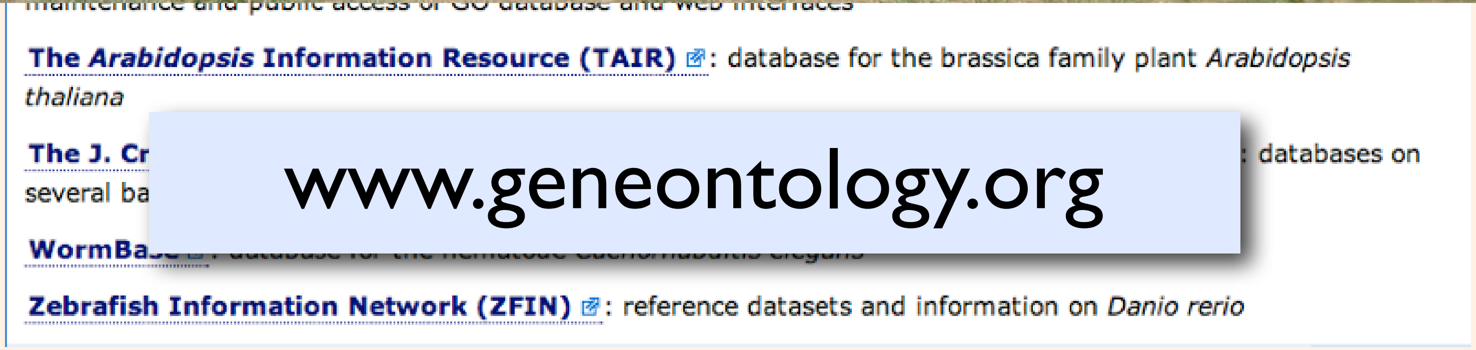

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