Gene Ontology Consortium

http://www.geneontology.org/

Ontology (for our purposes)

- "an explicit specification of some topic" –
  Stanford Knowledge Systems Lab

- Includes:
  - a vocabulary of terms (names for concepts)
  - defined logical relationships to each
  - definitions

Tactition

Taction

Tactile sense

Tactition

Taction

Tactile sense

perception of touch ; GO:0050975

What GO is not:

- Not a way of unifying databases!
- Not a dictated standard
- Additional ontologies needed to model biology and experimentation.

http://obo.sourceforge.net/

The Three Ontologies

- *Molecular Function*: elemental activity or task
- *Biological Process*: broad objective or goal
- *Cellular Component*: location or complex

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  DNA binding, catalysis of a reaction
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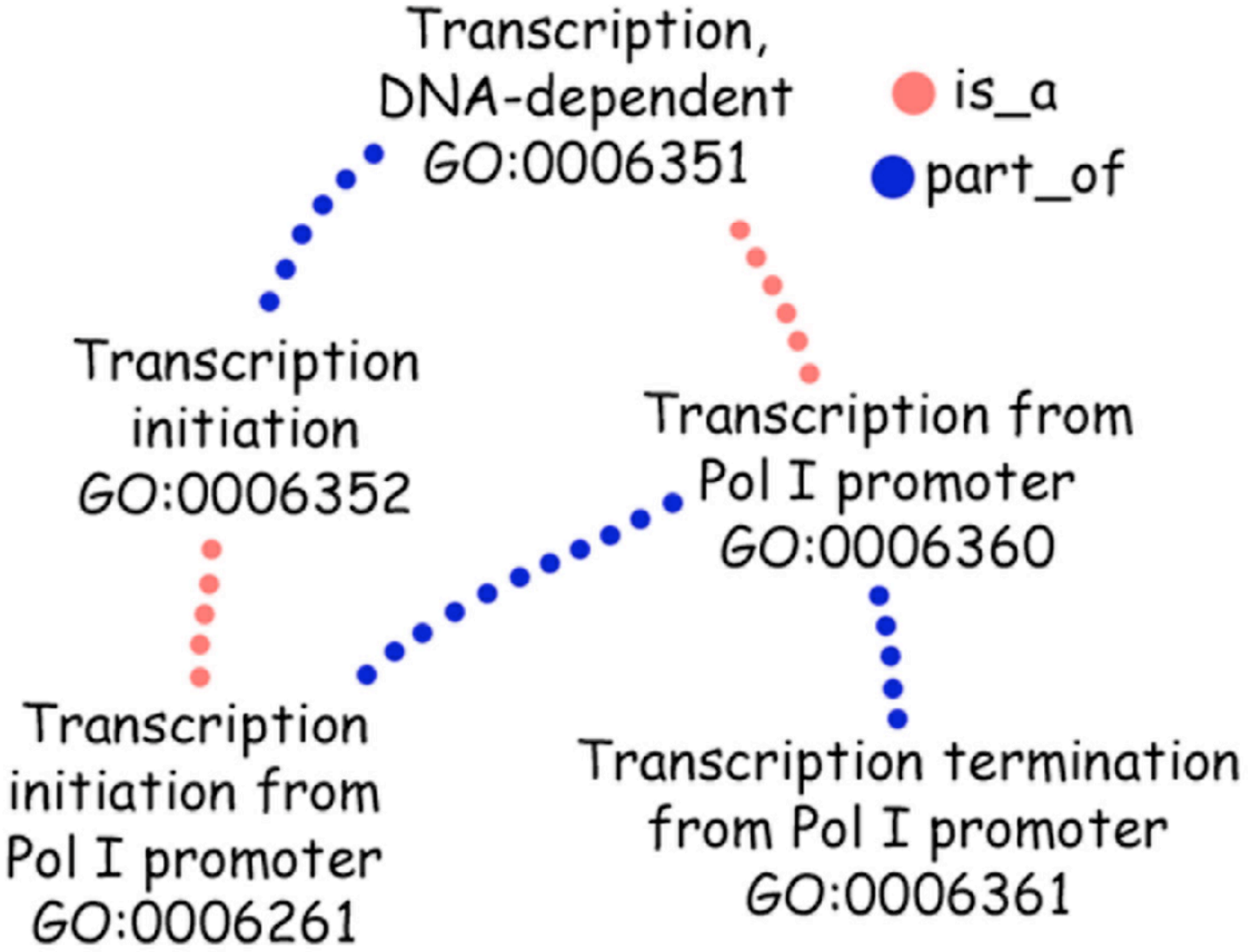
- *Biological Process*: broad objective or goal
  mitosis, signal transduction, metabolism
- *Cellular Component*: location or complex
  nucleus, ribosome

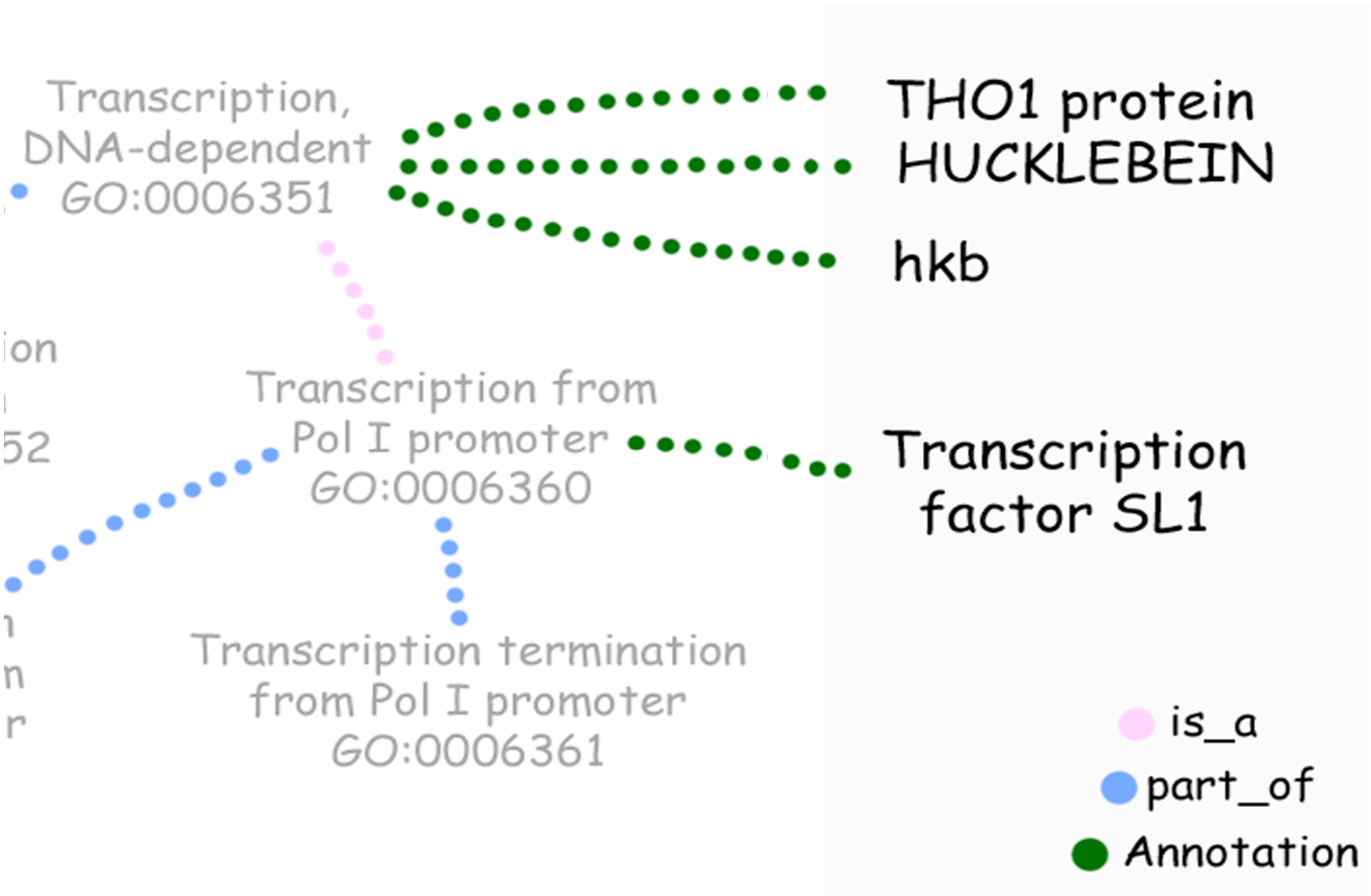

Transcription,
DNA-dependent
GO:0006351
is_a
part_of
Transcription
initiation
GO:0006352
Transcription from
Pol I promoter
GO:0006360
Transcription
initiation from
Pol I promoter
GO:0006261
Transcription termination
from Pol I promoter
GO:0006361

What's in a GO term?

term: transcription initiation

id: GO:0006352

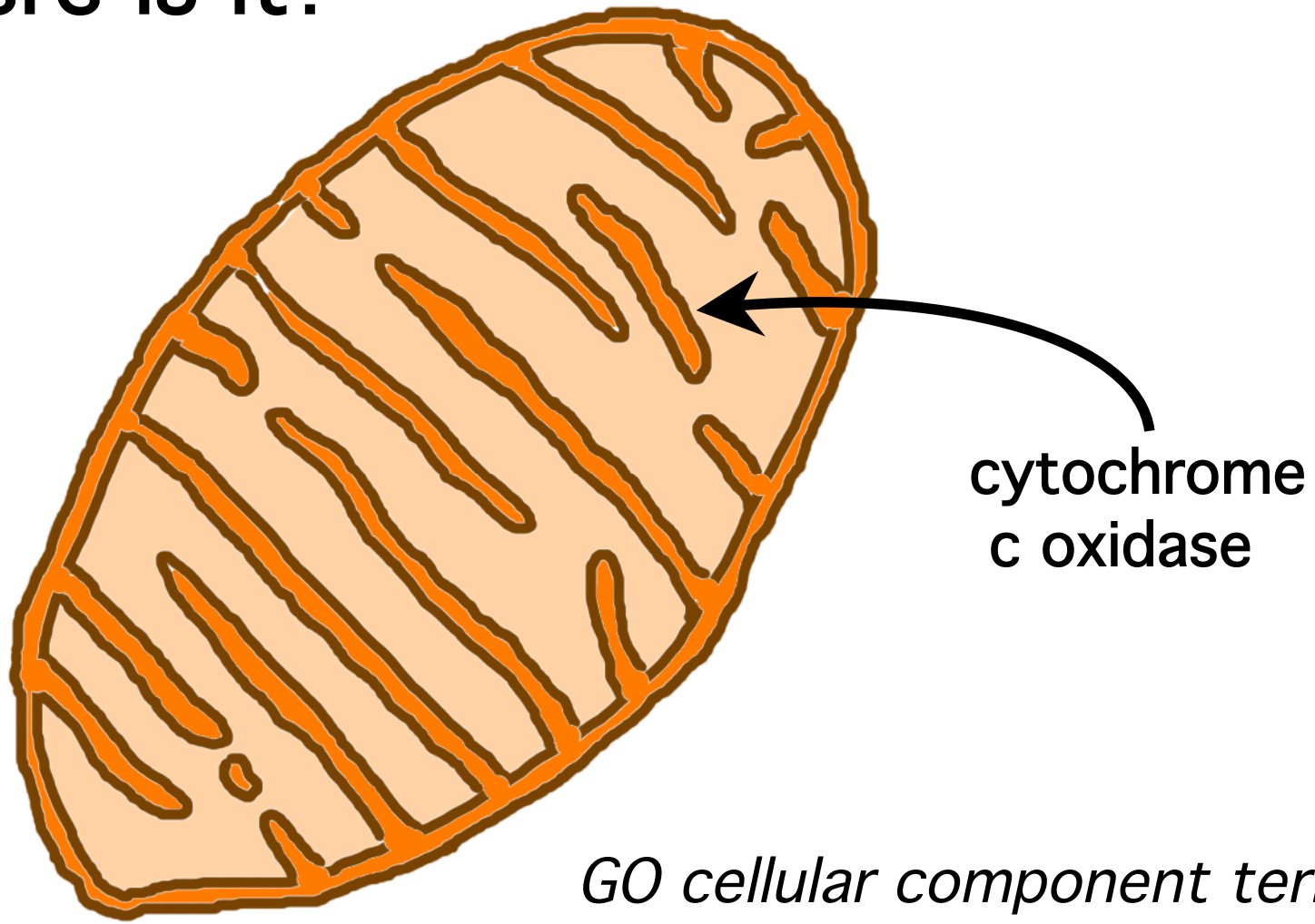
definition: Processes involved in starting transcription, where transcription is the synthesis of RNA by RNA polymerases using a DNA template.


Transcription,
DNA-dependent
GO:0006351
THO1 protein
HUCKLEBEIN
hkb
ion
52
Transcription from
Pol I promoter
GO:0006360
Transcription
factor SL1
Transcription termination
from Pol I promoter
GO:0006361
is_a
part_of
Annotation

Annotation

cytochrome c oxidase

Where is it?



GO cellular component term:
mitochondrial inner membrane ; GO:0005743

What does it do?

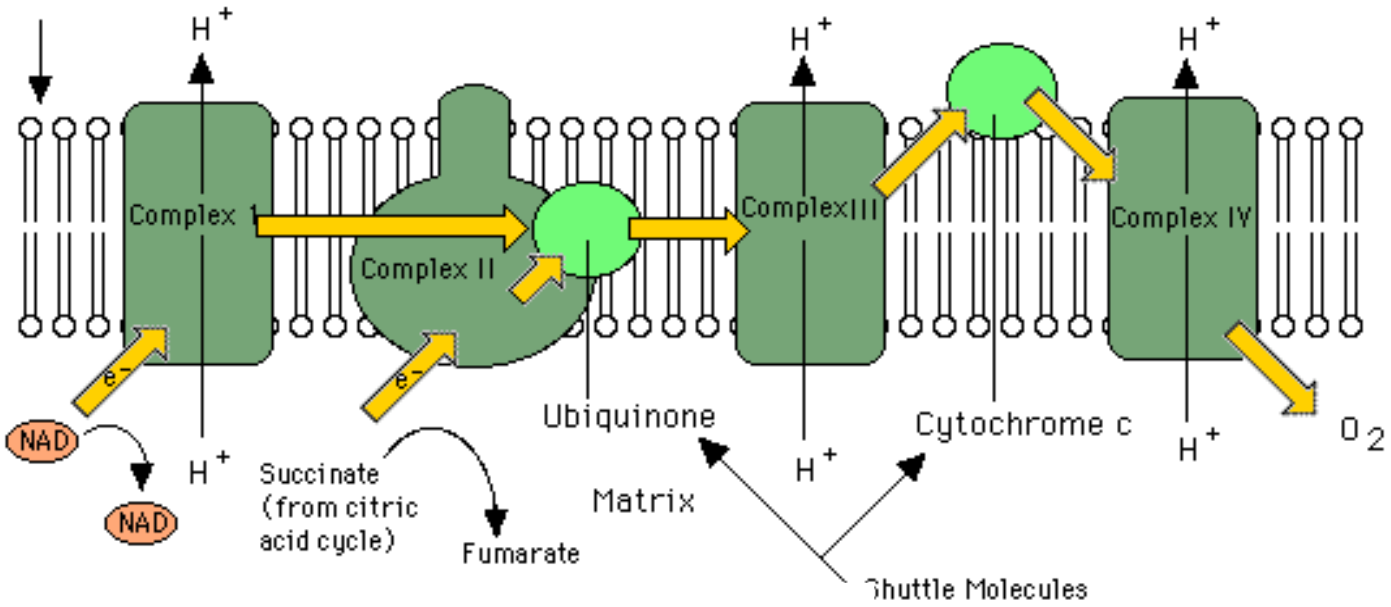
4 ferrocytochrome c + O2

=

4 ferricytochrome c + 2 H2O

GO molecular function term:
cytochrome-c oxidase activity; GO:0004497

Which process is this?



GO biological process term:
electron transport ; GO:0006118

http://ntri.tamuk.edu/cell/mitochondrion/krebpic.html

cytochrome-c oxidase activity

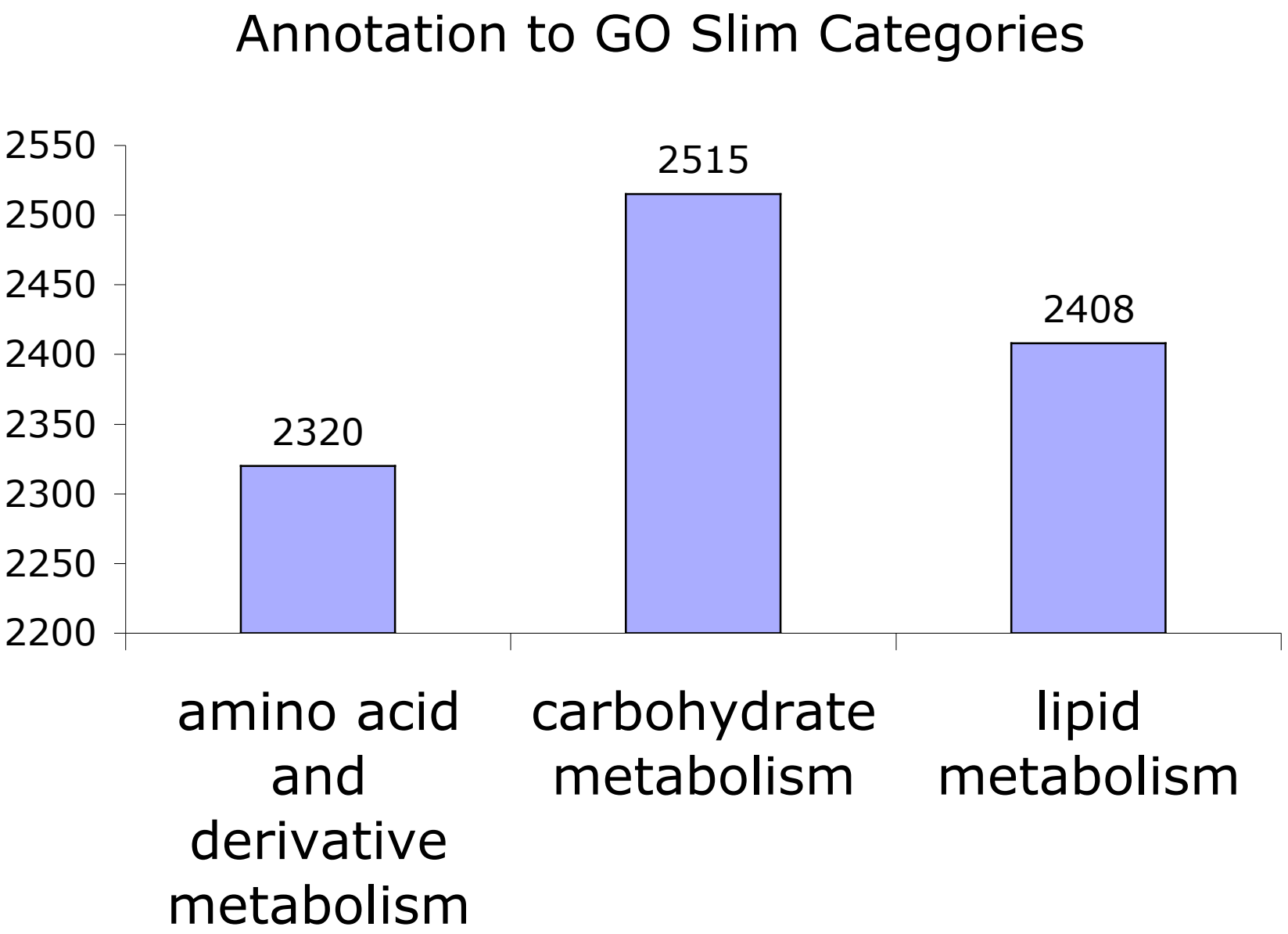
Accession: GO:0004129
Synonyms: None
Definition:
Catalysis of the reaction: 4 ferrocytochrome c + O2 = 4 ferricytochrome c + 2 H2O.
Term Lineage Graphical View

GO:0003673 : Gene_Ontology (146200)
Ⓟ GO:0003674 : molecular_function (97507)
① GO:0003824 : catalytic activity (32256)
① GO:0016491 : oxidoreductase activity (4721)
① GO:0015002 : heme-copper terminal oxidase activity (123)
① **GO:0004129 : cytochrome-c oxidase activity (118)**

☐	COX2_CAERE		UniProt	ISS	Cytochrome c oxidase polypeptide II (Fragment)
☐	COX2_HUMAN	GOst	UniProt	NAS	Cytochrome c oxidase polypeptide II
☐	COX2_YEAST	GOst	UniProt	IDA	Cytochrome c oxidase polypeptide II precursor
☐	COX3		TAIR	TAS	None
☐	COX3_CAEEL	GOst	UniProt	ISS	Cytochrome c oxidase polypeptide III
☐	COX3_HUMAN	GOst	UniProt	NAS	Cytochrome c oxidase polypeptide III
☐	COX3_YEAST	GOst	UniProt	IDA	Cytochrome c oxidase polypeptide III
☐	COX4	GOst	SGD	IDA	cytochrome c oxidase subunit IV
☐	Cox4b		RGD	IPI	cytochromecoxidase,subunit4b
☐	Cox4i2	GOst	MGI	ISS	cytochrome c oxidase subunit IV isoform 2
☐	COX5A	GOst	SGD	IDA	cytochrome c oxidase chain Va
☐	Cox5a		RGD	IDA	cytochromecoxidase,subunitVa
☐	Cox5b		RGD	TAS	cytochromecoxidasesubunitVb
☐	COX5B	GOst	SGD	IDA	cytochrome c oxidase chain Vb
☐	COX6	GOst	SGD	IDA	cytochrome c oxidase subunit

GO Slim

- **GO:0008152 : metabolism (34935)**
  - GO:0006066 : alcohol metabolism (1043)
  - GO:0006081 : aldehyde metabolism (67)
  - GO:0009308 : amine metabolism (2232)
  - **GO:0006519 : amino acid and derivative metabolism (2320)**
    - **GO:0006575 : amino acid derivative metabolism (659)**
      - GO:0018902 : 1,3-dichloro-2-propanol metabolism (0)
      - GO:0018871 : 1-aminocyclopropane-1-carboxylate metabolism (1)
      - GO:0019471 : 4-hydroxyproline metabolism (35)
      - GO:0046442 : aerobactin metabolism (0)
      - GO:0042398 : amino acid derivative biosynthesis (351)
      - GO:0042219 : amino acid derivative catabolism (70)
      - GO:0009448 : aminobutyrate metabolism (14)
      - GO:0006576 : biogenic amine metabolism (278)
      - GO:0009692 : ethylene metabolism (38)
      - GO:0046516 : hypusine metabolism (7)
      - GO:0046418 : nopaline metabolism (0)
      - GO:0046419 : octopine metabolism (1)


Annotation to GO Slim Categories
2550
2500
2450
2400
2350
2300
2250
2200
2320
2515
2408
amino acid and derivative metabolism
carbohydrate metabolism
lipid metabolism

13 May 2004:

Total terms = 17400

90% have definitions

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GO Tools
Mappings to GO

Archives

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Terms/Annotations

GENE ONTOLOGY CONSORTIUM

What is the Gene Ontology? *Download the Ontologies*

The goal of the Gene Ontology™ (GO) Consortium is to produce a controlled vocabulary that can be applied to all organisms even as knowledge of gene and protein roles in cells is accumulating and changing. GO provides three structured networks of defined terms to describe gene product attributes. GO is one of the controlled vocabularies of the Open Biological Ontologies.

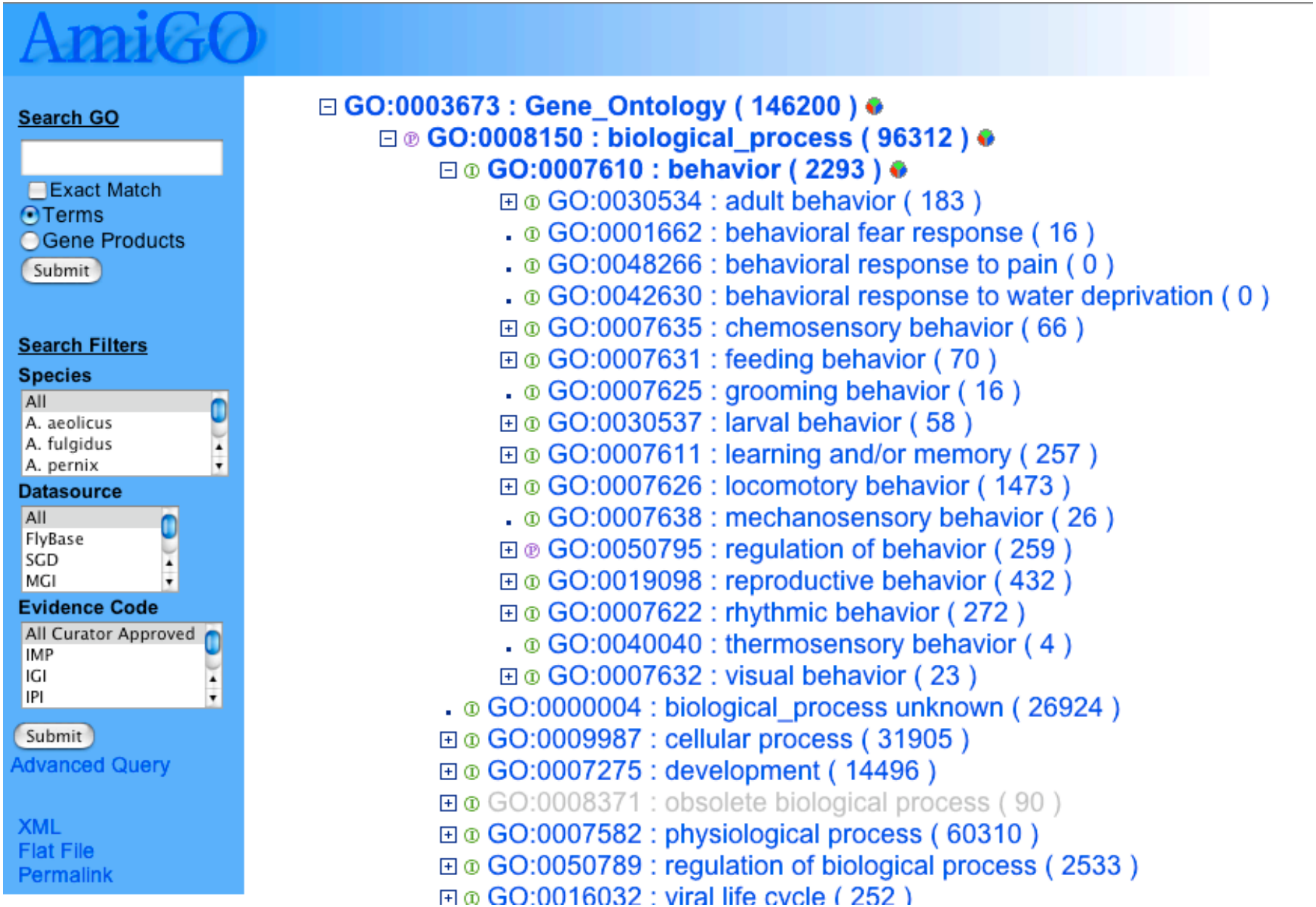
- Submit new GO term suggestions via the Curator Requests Tracker at SourceForge. Help with new term submission is available.
- Send comments and questions to go@geneontology.org.

Search Terms and Annotations

go

This search uses the AmiGO browser. You can also use one of the many other GO Browsers

http://www.geneontology.org/



http://www.godatabase.org/cgi-bin/amigo/go.cgi

AmiGO

mechanosensory behavior

Accession: GO:0007638
Synonyms: None
Definition:
Behavior that is dependent upon the sensation of movement.
Term Lineage Graphical View

GO:0003673 : Gene_Ontology (146200)
Ⓟ GO:0008150 : biological_process (96312)
① GO:0007610 : behavior (2293)
① **GO:0007638 : mechanosensory behavior (26)**

External References

⊞ SP_KW (1)

Direct Gene Product Associations

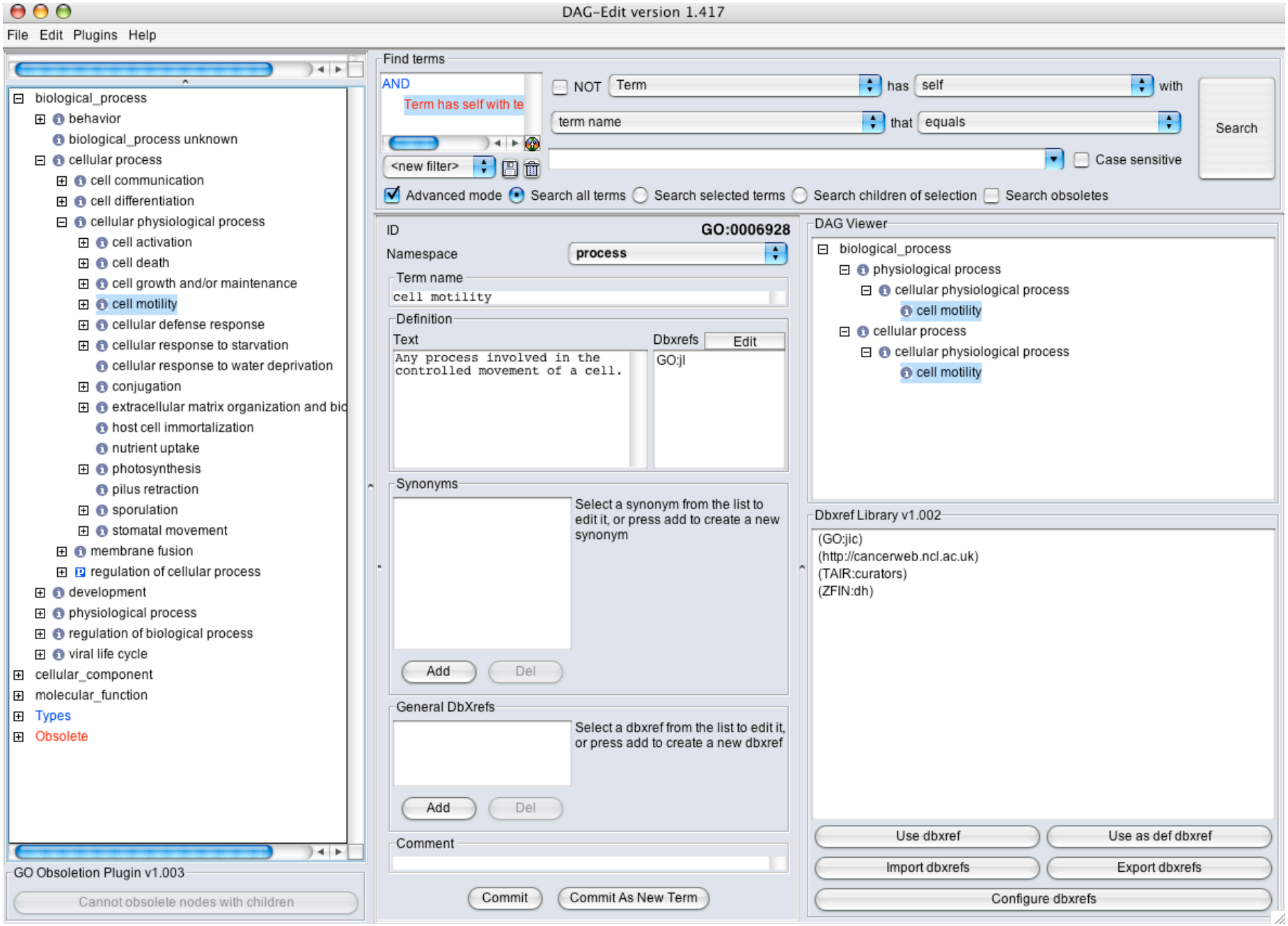
Direct Associations Submit

Filter Associations

Datasource	Evidence Code	Species
All	All Curator Approved	All
FlyBase	IMP	A. aeolicus
SGD	IGI	A. fulgidus
MGI	IPI	A. pernix

Submit

Gene Symbol	Datasource	Evidence	Full Name
bas	FlyBase	IMP	bas
bss	FlyBase	IMP	bss
C50H2.3	Wormbase	IMP	None
C53C9.3	Wormbase	IMP	None
eas	FlyBase	IMP	eas
EAS_DROME GOst	UniProt	IMP	Ethanolamine kinase


DAG-Edit version 1.417
File Edit Plugins Help
biological_process
behavior
biological_process unknown
cellular process
cell communication
cell differentiation
cellular physiological process
cell activation
cell death
cell growth and/or maintenance
cell motility
cellular defense response
cellular response to starvation
cellular response to water deprivation
conjugation
extracellular matrix organization and bio
host cell immortalization
nutrient uptake
photosynthesis
pilus retraction
sporulation
stomatal movement
membrane fusion
regulation of cellular process
development
physiological process
regulation of biological process
viral life cycle
cellular_component
molecular_function
Types
Obsolete
GO Obsoletion Plugin v1.003
Cannot obsolete nodes with children
Find terms
AND
Term has self with te
<new filter>
NOT
Term
has
self
with
term name
that
equals
Case sensitive
Search
Advanced mode
Search all terms
Search selected terms
Search children of selection
Search obsoletes
ID
GO:0006928
Namespace
process
Term name
cell motility
Definition
Text
Any process involved in the controlled movement of a cell.
Dbxrefs
Edit
GO:jl
Synonyms
Select a synonym from the list to edit it, or press add to create a new synonym
Add
Del
General DbXrefs
Select a dbxref from the list to edit it, or press add to create a new dbxref
Add
Del
Comment
Commit
Commit As New Term
DAG Viewer
biological_process
physiological process
cellular physiological process
cell motility
cellular process
cellular physiological process
cell motility
Dbxref Library v1.002
(GO:jic)
(http://cancerweb.ncl.ac.uk)
(TAIR:curators)
(ZFIN:dh)
Use dbxref
Use as def dbxref
Import dbxrefs
Export dbxrefs
Configure dbxrefs

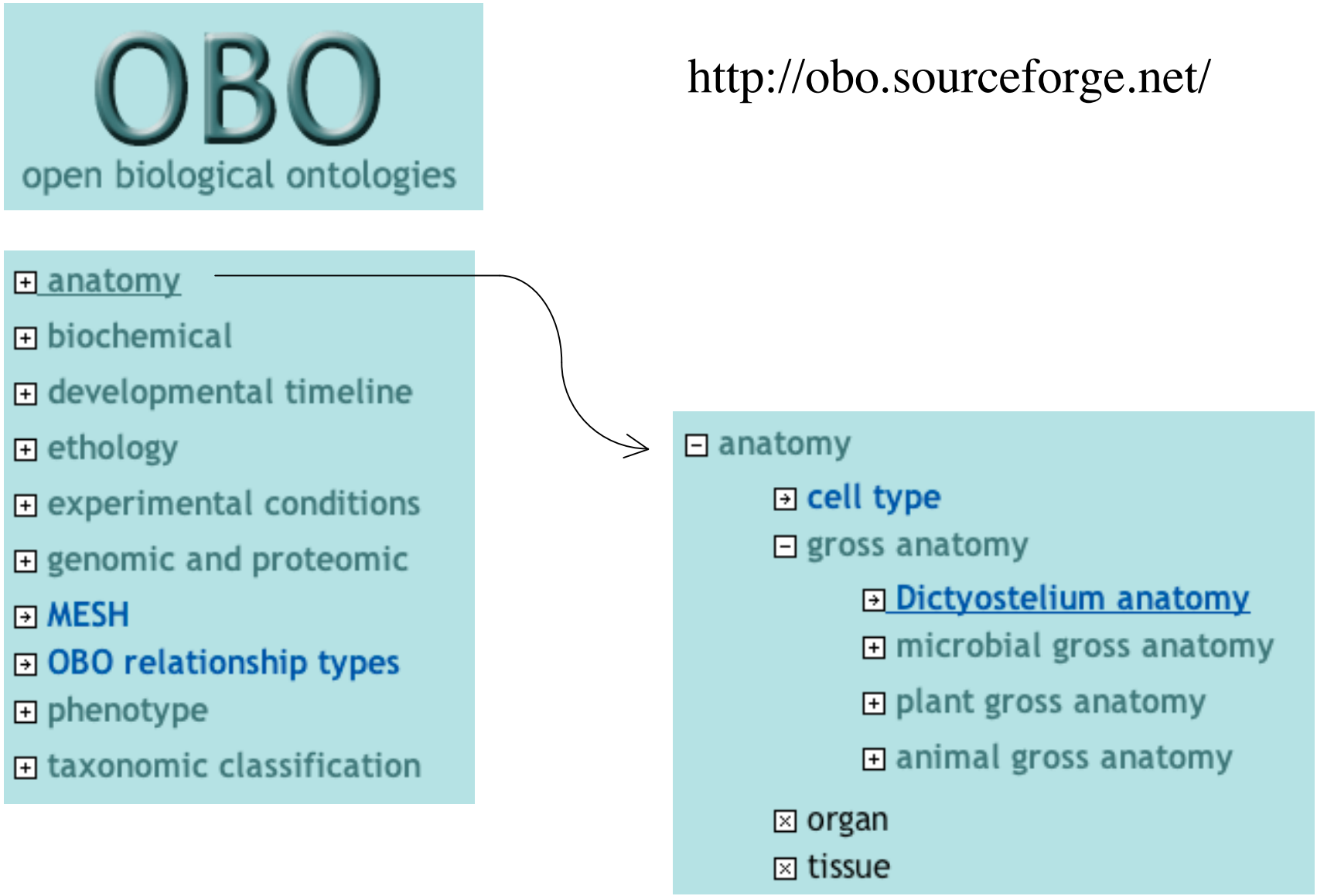
GO flatfile format

```
$Gene_Ontology ; GO:0003673
 <biological_process ; GO:0008150
  %behavior ; GO:0007610 ; synonym:behaviour
   %adult behavior ; GO:0030534
    %adult feeding behavior ; GO:0008343
    %adult locomotory behavior ; GO:0008344
     %adult walking behavior ; GO:0007628
     %flight behavior ; GO:0007629
     %jump response ; GO:0007630
```

OBO flatfile format

```
[Term]
id: GO:0042174
name: negative regulation of sporulation
namespace: process
def: "Any […] sporulation." [GO:curators]
is_a: GO:0042173

[Term]
id: GO:0030121
name: AP-1 adaptor complex
namespace: component
def: "An […] network." [GO:mah]
exact_synonym: "HA1" []
is_a: GO:0030131
relationship: part_of GO:0030130
```


OBO
open biological ontologies
http://obo.sourceforge.net/
anatomy
biochemical
developmental timeline
ethology
experimental conditions
genomic and proteomic
MESH
OBO relationship types
phenotype
taxonomic classification
anatomy
cell type
gross anatomy
Dictyostelium anatomy
microbial gross anatomy
plant gross anatomy
animal gross anatomy
organ
tissue

Conditions:

- **Open Source**
- Common shared **syntax**
- **Orthogonal** to other ontologies
- Unique **identifier space**
- Terms **defined**

Cross-products

Hill, D.P., Blake, J.A., Richardson,
J.E. and Ringwald, M. 2002.

Extension and Integration of the Gene Ontology (GO):
Combining GO vocabularies with external vocabularies.

Genome Res **12:** 1982-1991.

Heart development node

-% heart development
--< heart morphogenesis
----< heart formation
----< heart structural organization
--< heart maturation

Mus Adult Gross Anatomy

--% cardiovascular system
----< heart
--------< cardiogenic plate
--------< primitive heart tube
----------< myocardium

Biological Process Ontology

% Biological process
--% development
---< morphogenesis
----< formation
----< structural development
---< maturation

Cross product

% heart *development*
--< cardiogenic plate *development*
--< primitive heart tube *development*
----< myocardium *development*

The Full cross-product

–Generate the entire cross product of the two DAGS

–Use biological knowledge to pick and choose

–We would need to do a lot of culling.

The pick-and-choose approach

Pick out anatomical terms and combine them with appropriate developmental process terms.

OBOL

Open Bio-Ontology Language

Chris Mungall and Suzanna Lewis,
University of California, Berkeley

Many terms are standardized

Biosynthesis:
The formation from simpler components of…

Catabolism:
The breakdown into simpler components of…

Regulation:
Any process that modulates the frequency, rate or extent of…

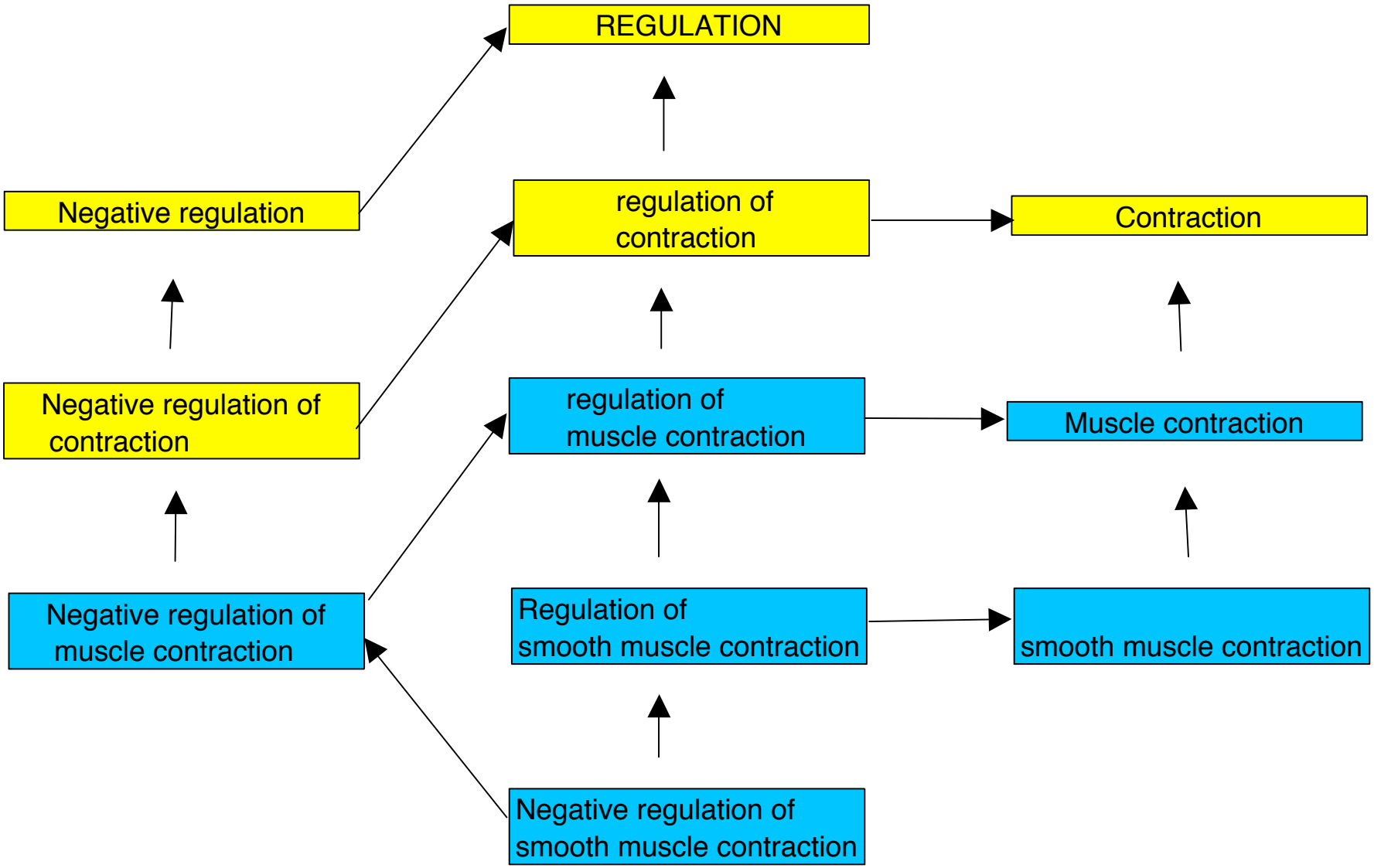
Formal Grammars

A rule system for

- parsing (decomposing)
- generating (composing)

sequences of symbols.

A Typical Fiendishly Hard Lattice



Contributors

FlyBase
DictyBase
GeneDB S. pombe
Mouse Genome Database
Genome Knowledge Base
TIGR Gramene
The Arabidopsis Information Resource
The Zebrafish Information Network
Berkeley Drosophila Genome Project
Saccharomyces Genome Database
The Institute for Genomic Research
Rat Genome Database
WormBase
Compugen
GeneDB for protozoa
EBI GOA project

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