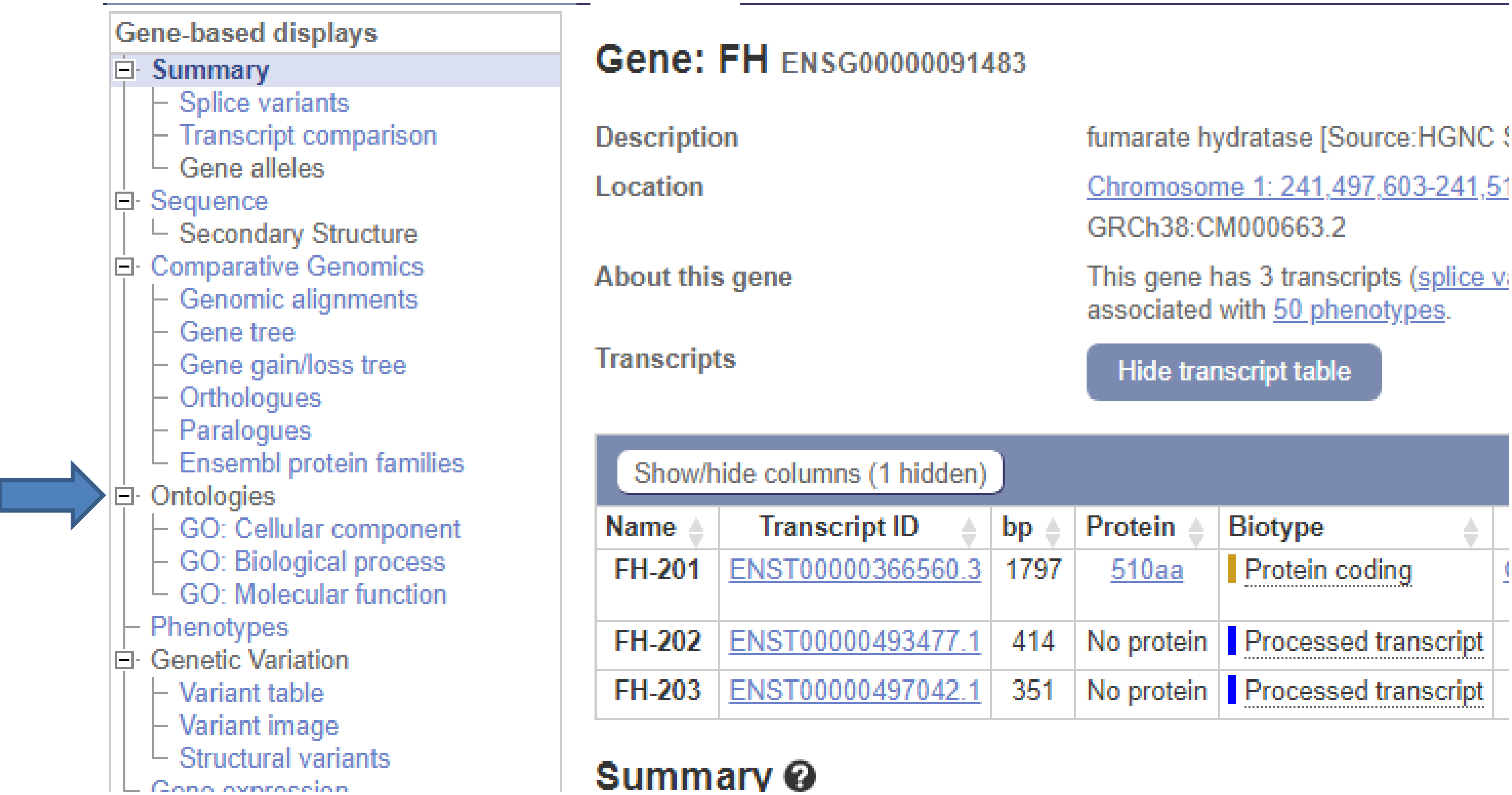
Ch2-L5.2

Accessories

GO

Gene Ontology

When you are in the [ENSEMBL browser]on some human gene, please note that on the left menu there are Gene Ontologies.



Gene: FH ENSG00000091483

Description	fumarate hydratase [Source:HGNC
Location	Chromosome 1: 241,497,603-241,5 GRCh38:CM000663.2
About this gene	This gene has 3 transcripts (splice v associated with 50 phenotypes.
Transcripts	Hide transcript table

Name	Transcript ID	bp	Protein	Biotype
FH-201	ENST00000366560.3	1797	510aa	Protein coding
FH-202	ENST00000493477.1	414	No protein	Processed transcript
FH-203	ENST00000497042.1	351	No protein	Processed transcript

Summary

Click on the three terms and have a look

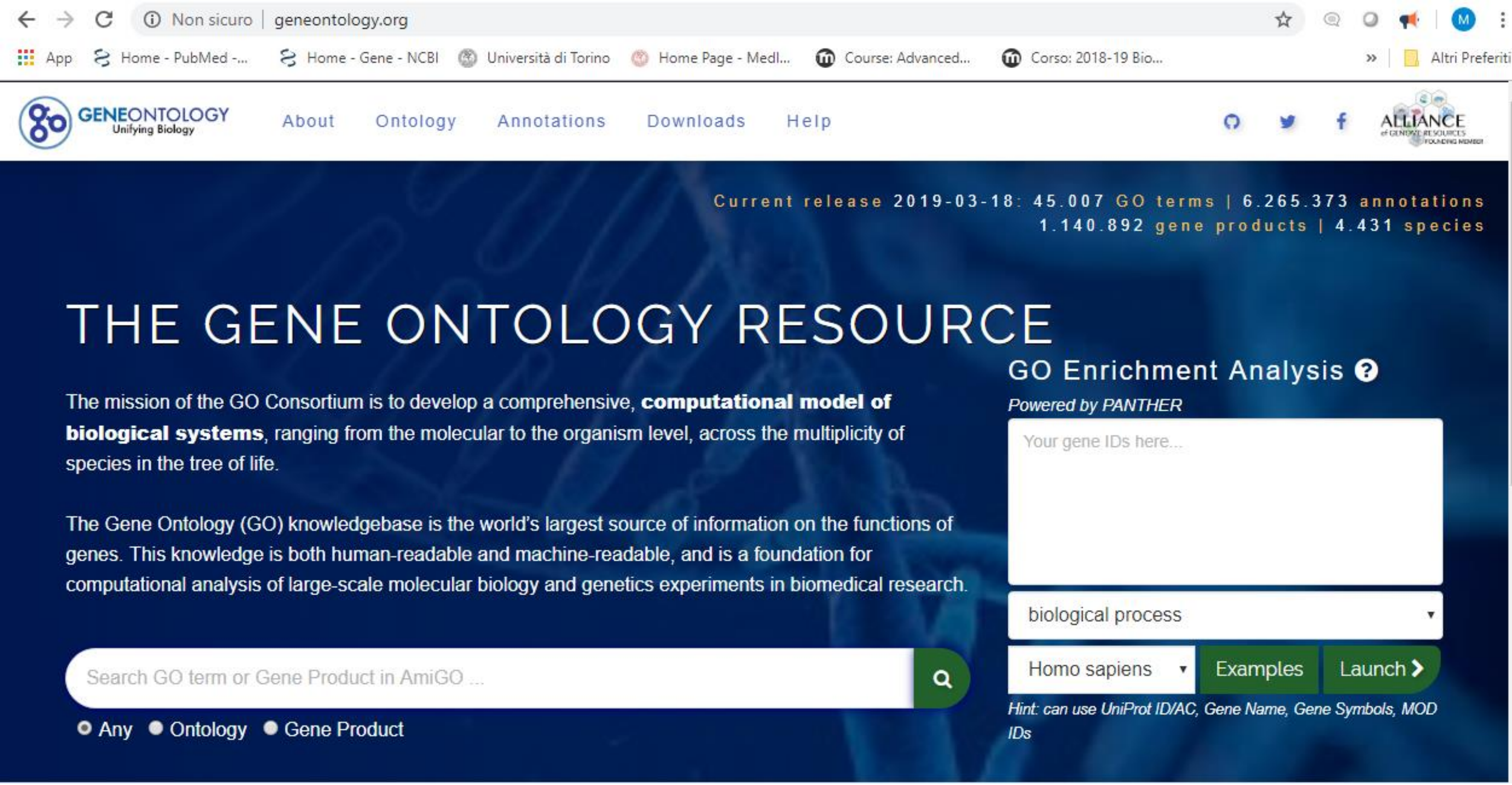
Ontologies are established and organized by the Gene Ontology Consortium

Project: **Gene Ontology**

Definition of controlled terms to describe genes

Gene Ontology

http://www.geneontology.org/



Current release 2019-03-18: 45.007 GO terms | 6.265.373 annotations
1.140.892 gene products | 4.431 species

THE GENE ONTOLOGY RESOURCE

The mission of the GO Consortium is to develop a comprehensive, **computational model of biological systems**, ranging from the molecular to the organism level, across the multiplicity of species in the tree of life.

The Gene Ontology (GO) knowledgebase is the world's largest source of information on the functions of genes. This knowledge is both human-readable and machine-readable, and is a foundation for computational analysis of large-scale molecular biology and genetics experiments in biomedical research.

Search GO term or Gene Product in AmiGO ...

● Any ○ Ontology ○ Gene Product

GO Enrichment Analysis

Powered by PANTHER

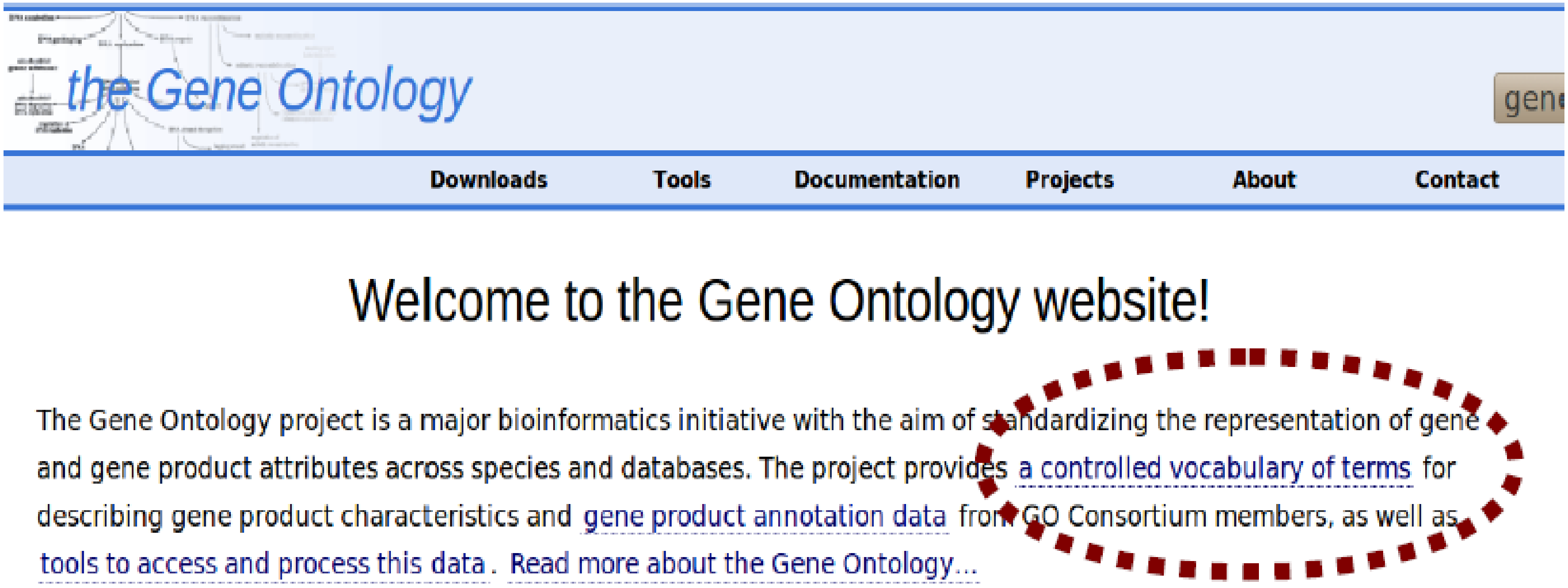
Your gene IDs here...

biological process

Homo sapiens

Hint: can use UniProt ID/AC, Gene Name, Gene Symbols, MOD IDs

Aim:



Welcome to the Gene Ontology website!

The Gene Ontology project is a major bioinformatics initiative with the aim of standardizing the representation of gene and gene product attributes across species and databases. The project provides a controlled vocabulary of terms for describing gene product characteristics and gene product annotation data from GO Consortium members, as well as tools to access and process this data. Read more about the Gene Ontology...

Aim:

Biologists tend to use different names for the same concept:

eg: ***translation*** vs ***protein synthesis***

The Gene Ontology (GO) project is a collaborative effort to address the need for consistent descriptions of gene products in different databases.

→ a unified (universal) vocabulary for biology

The GO project has developed three structured controlled vocabularies (**ontologies**) that describe gene products and defined logical relationships between them.

Ontology: vocabulary

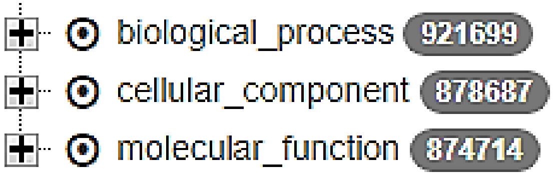
The GO consortium has defined three different structured vocabularies (ontologies):

- *Molecular Function:* elemental activity or task
  DNA binding, catalysis of a reaction

- *Biological Process:* broad objective or goal
  mitosis, signal transduction, metabolism

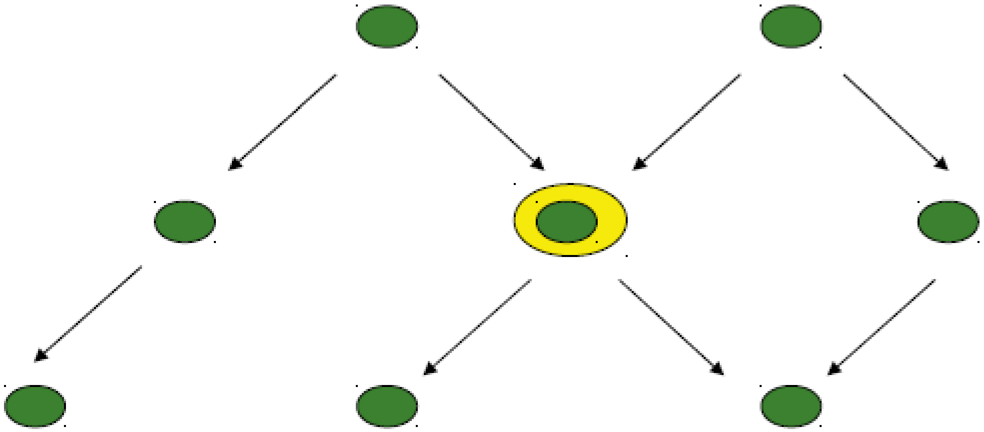
- *Cellular Component:* location or complex
  nucleus, ribosome

Today, March 29, 2019



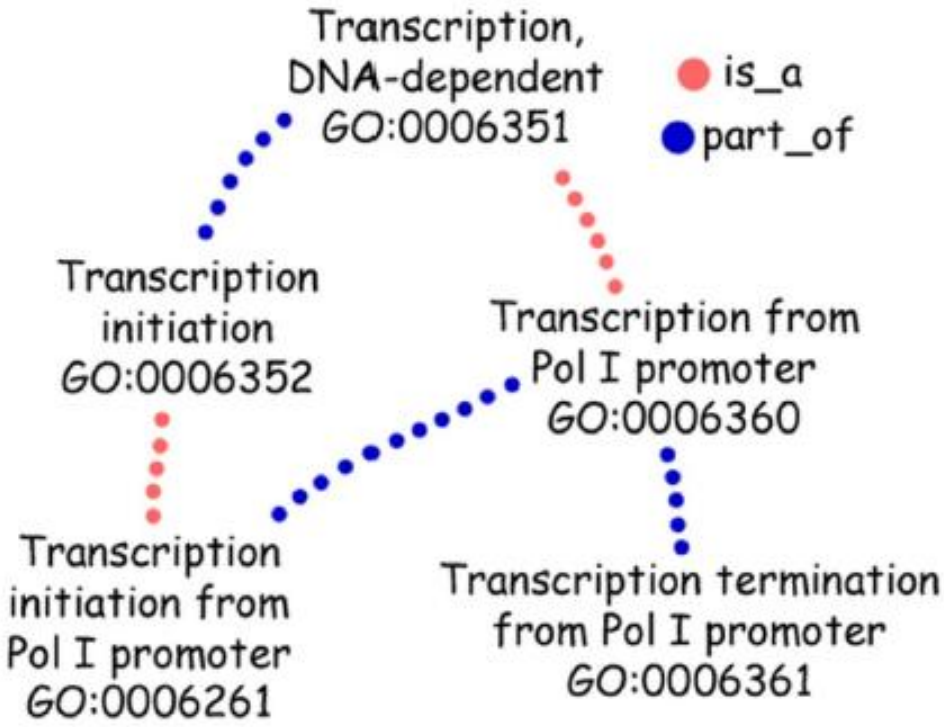
biological_process 921699
cellular_component 878687
molecular_function 874714

The ontologies are structured as directed acyclic graphs, which are similar to hierarchies but differ in that a child, or more specialized, term can have many parents, or less specialized, terms.



The vocabulary: relationships

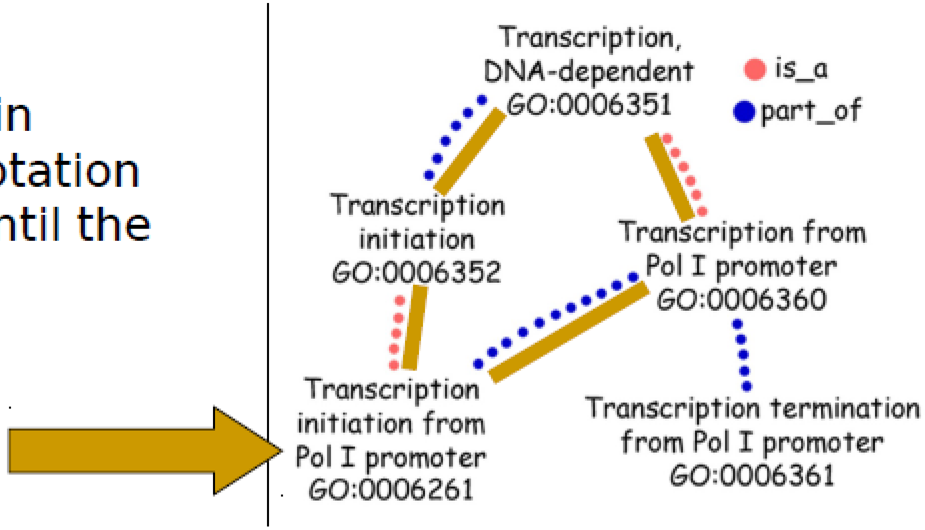
The terms in an ontology are linked by different types of relationships, like *is_a* and *part_of*.



The vocabulary: true path rule

If the child term describes a gene product, then all its parent terms must also apply to that gene product.

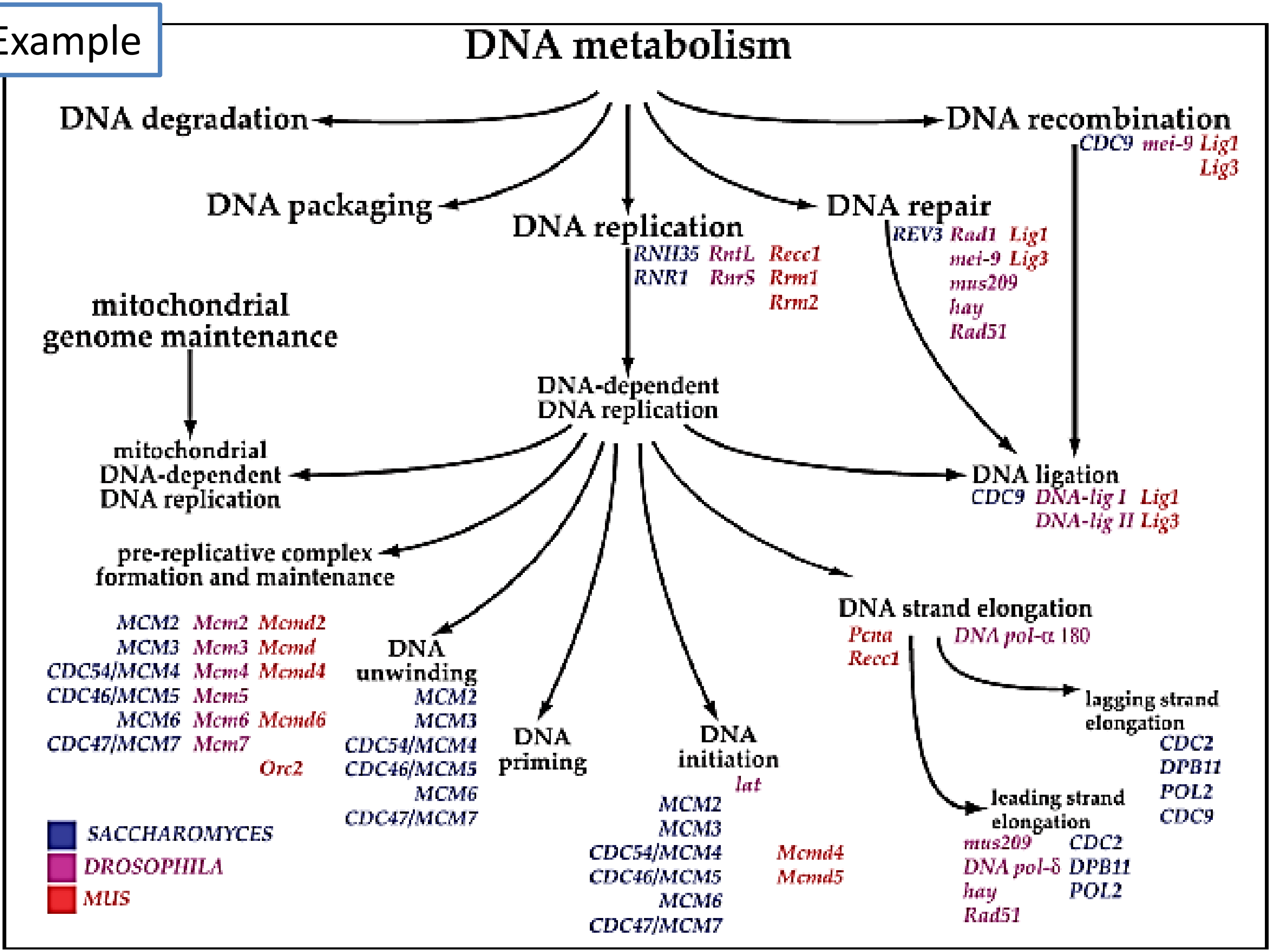
Annotation to a certain GO term implies annotation to all the ancestors until the root GO term.



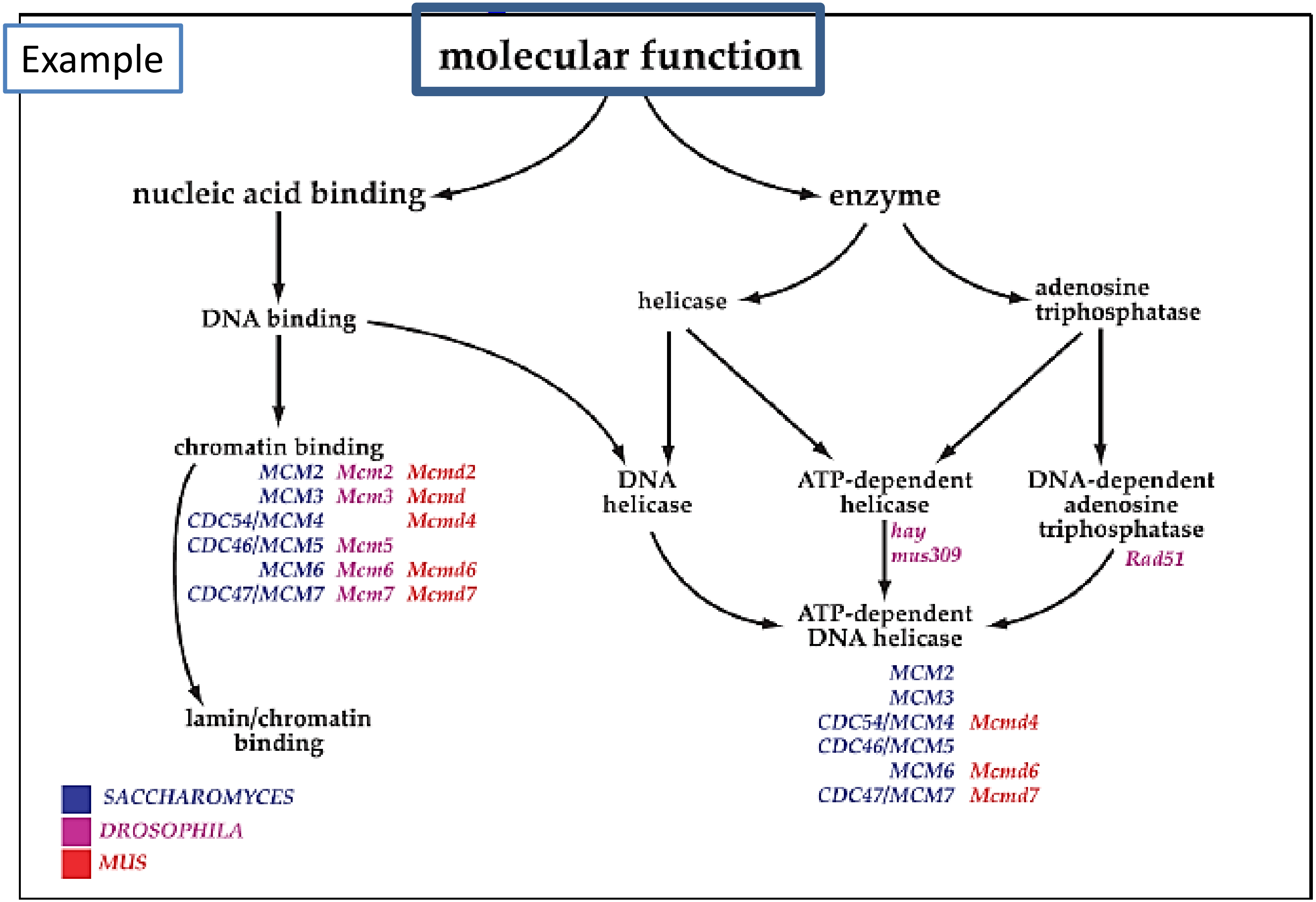
The Vocabulary

Biological processes

Example



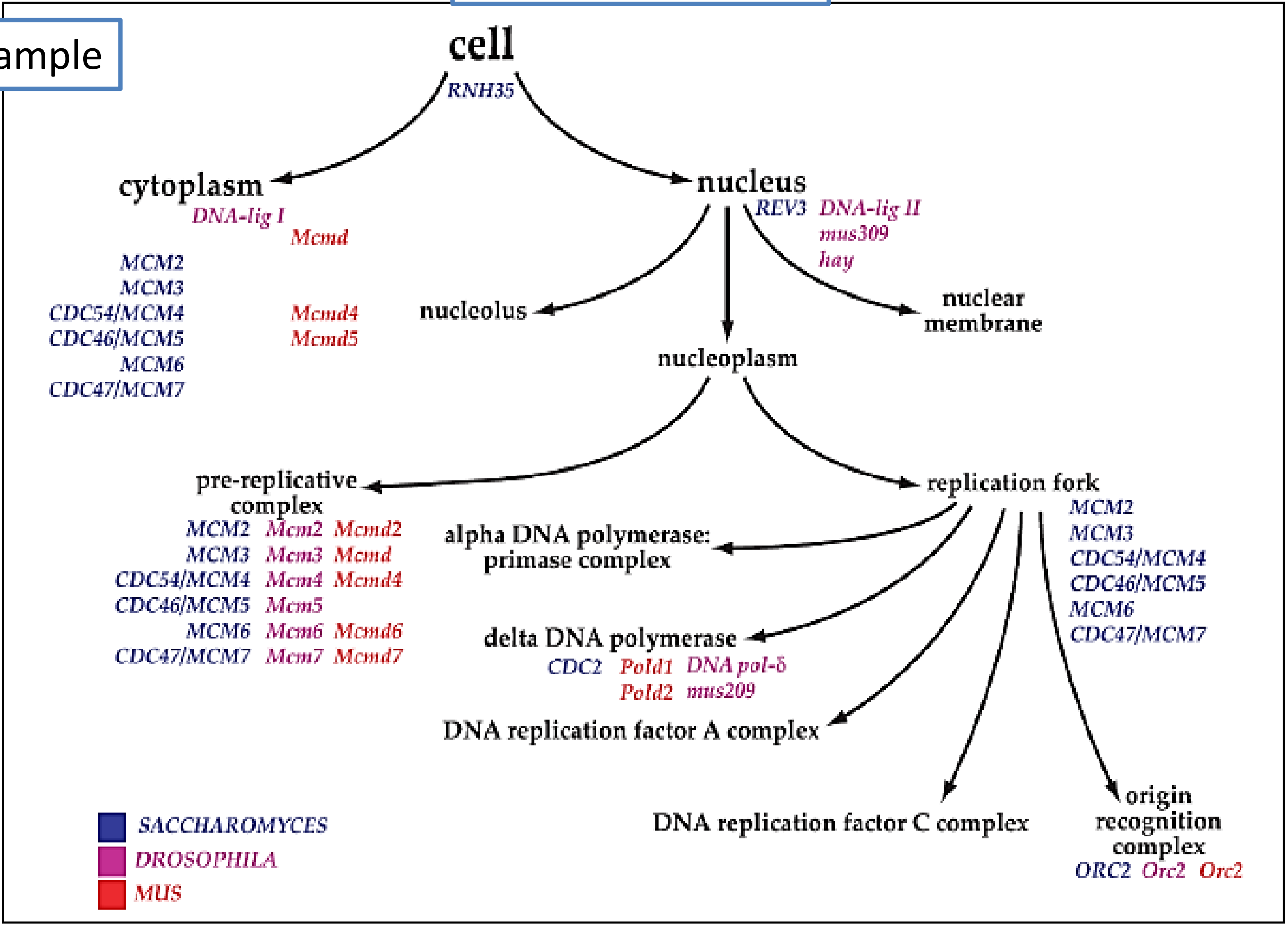
Example


molecular function
nucleic acid binding
enzyme
DNA binding
helicase
adenosine triphosphatase
chromatin binding
MCM2 Mcm2 Mcmd2
MCM3 Mcm3 Mcmd
CDC54/MCM4 Mcmd4
CDC46/MCM5 Mcm5
MCM6 Mcm6 Mcmd6
CDC47/MCM7 Mcm7 Mcmd7
DNA helicase
ATP-dependent helicase
hay
mus309
DNA-dependent adenosine triphosphatase
Rad51
ATP-dependent DNA helicase
MCM2
MCM3
CDC54/MCM4 Mcmd4
CDC46/MCM5
MCM6 Mcmd6
CDC47/MCM7 Mcmd7
lamin/chromatin binding
SACCHAROMYCES
DROSOPHILA
MUS

PAUSE

Cell component

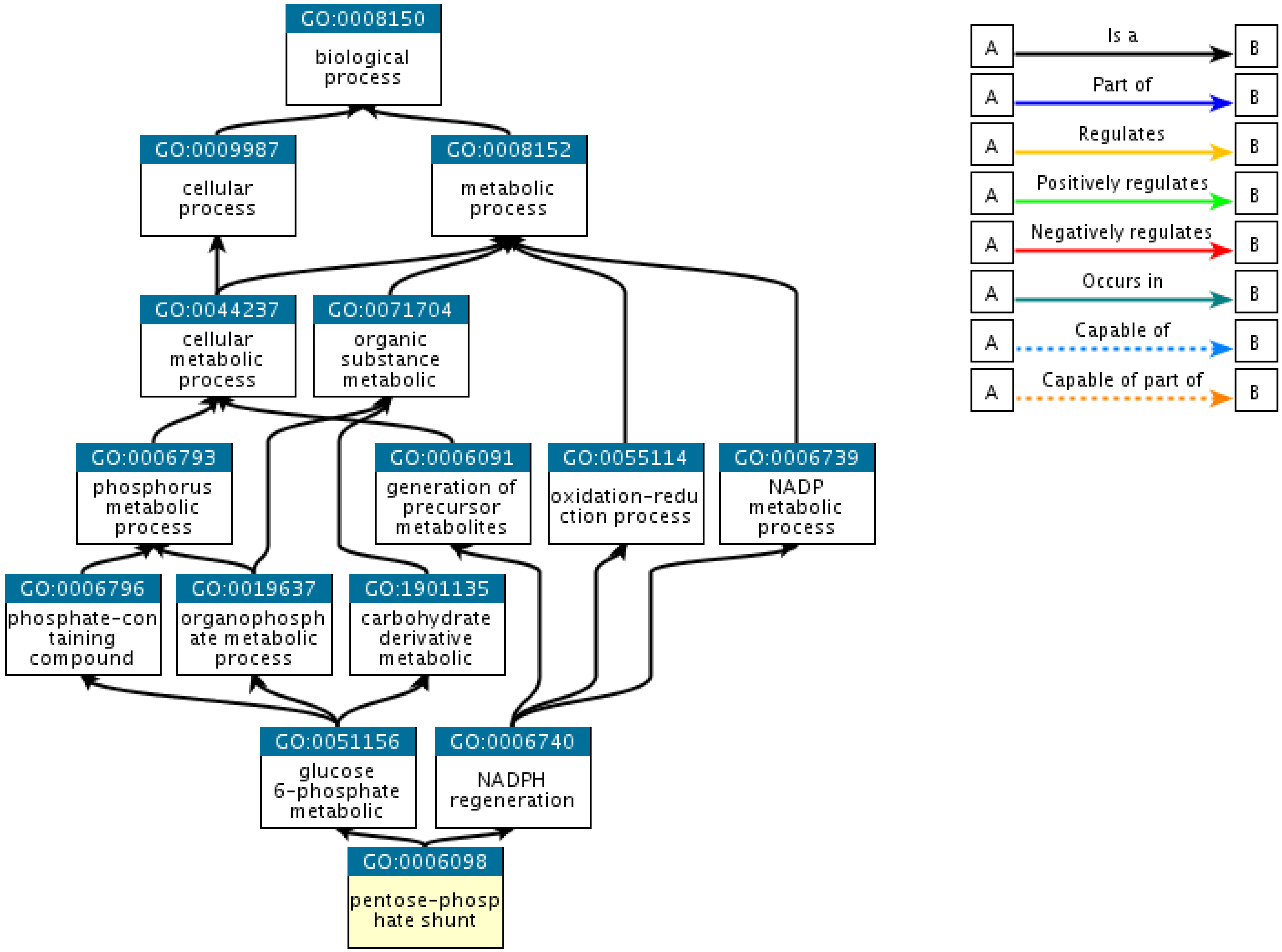
Example



Ancestor Chart

Ancestor chart for GO:0006098 Chart options

search term «pentose phosphate shunt»



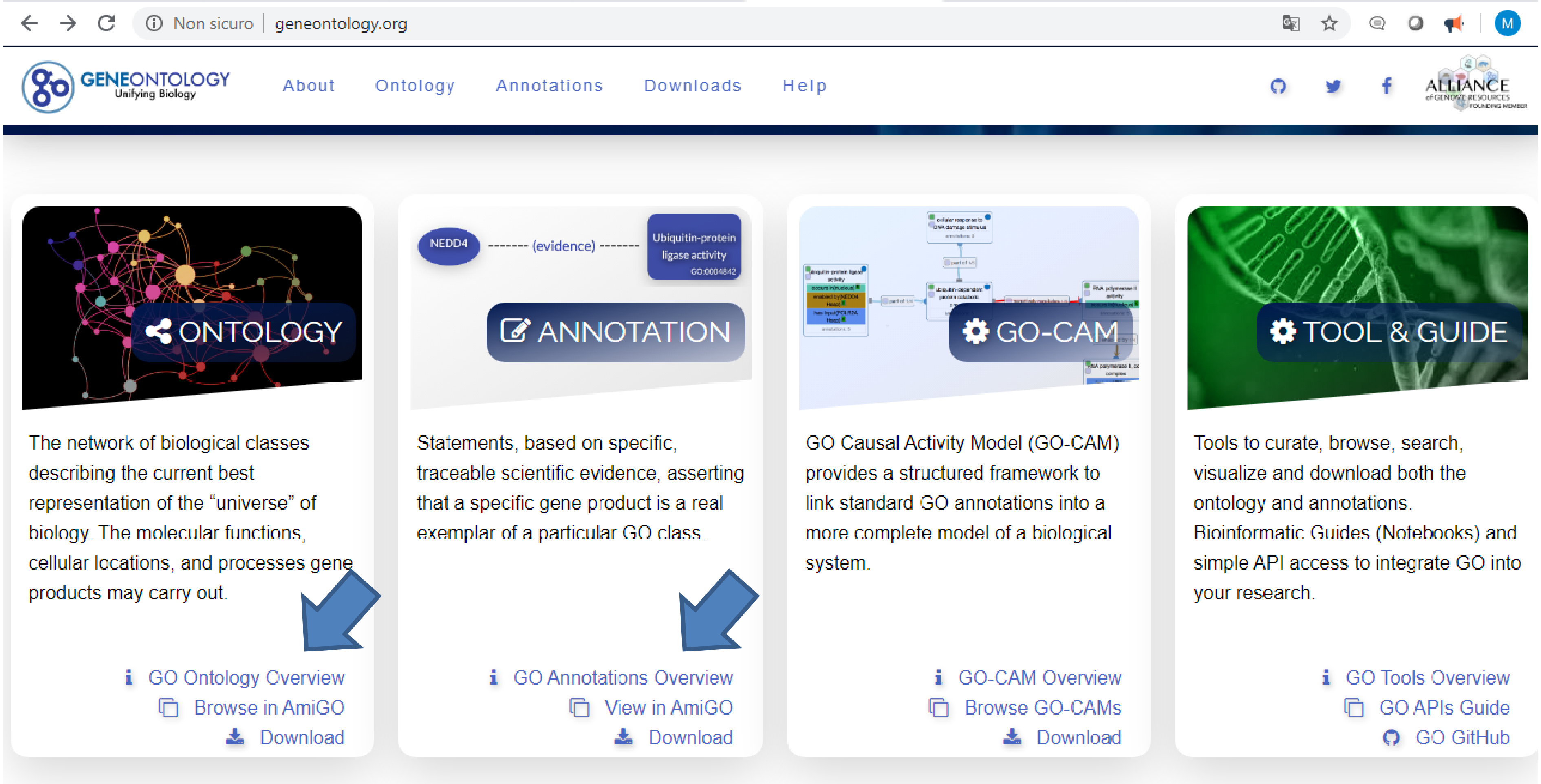
QuickGO - https://www.ebi.ac.uk/QuickGO

GO does not include:

- Processes, functions or components that are unique to mutants or diseases: e.g. **oncogenesis**
- Attributes of sequence such as intron/exon parameters
- Protein domains or structural features or interactions
- Evolution and expression information
- Anatomical or histological features

Go back now to the first page of the Gene Ontology website:

http://geneontology.org/



ONTOLOGY

The network of biological classes describing the current best representation of the “universe” of biology. The molecular functions, cellular locations, and processes gene products may carry out.

GO Ontology Overview
Browse in AmiGO
Download

ANNOTATION

Statements, based on specific, traceable scientific evidence, asserting that a specific gene product is a real exemplar of a particular GO class.

GO Annotations Overview
View in AmiGO
Download

GO-CAM

GO Causal Activity Model (GO-CAM) provides a structured framework to link standard GO annotations into a more complete model of a biological system.

GO-CAM Overview
Browse GO-CAMs
Download

TOOL & GUIDE

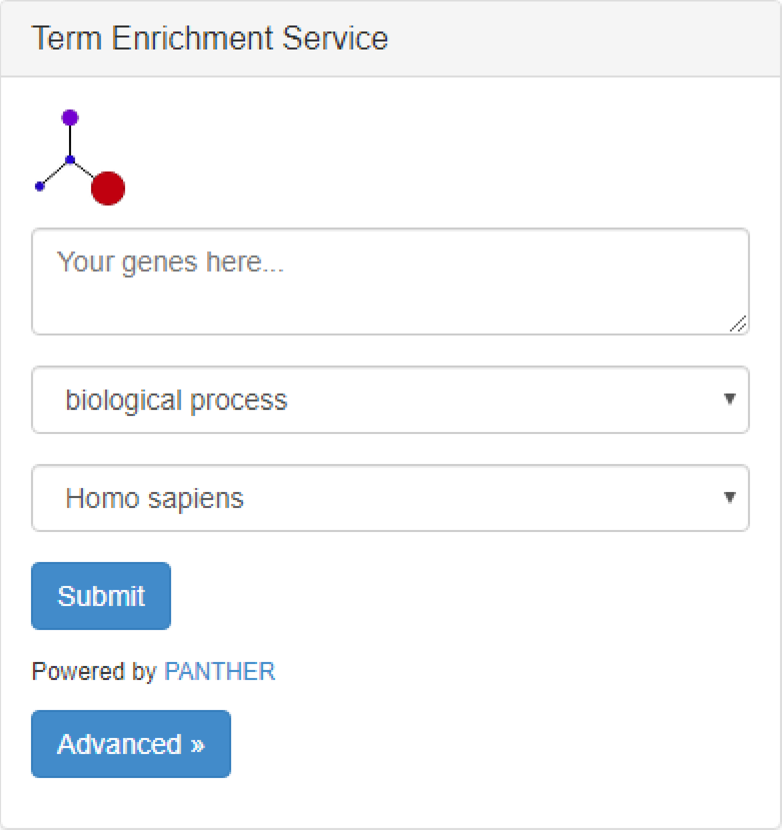
Tools to curate, browse, search, visualize and download both the ontology and annotations. Bioinformatic Guides (Notebooks) and simple API access to integrate GO into your research.

GO Tools Overview
GO APIs Guide
GO GitHub

A «tool» to examine single genes and gene groups
(as, for example, when you have a «gene expression profile» or a signature)

AMIGO

http://amigo.geneontology.org/amigo



Term Enrichment Service

Powered by PANTHER

Also see Statistics http://amigo.geneontology.org/amigo/base_statistics

Examples: choose a gene name

Go to see it in:

NCBI Gene
ENSEMBL
UniProt
PDB
Gene Ontology

OMIM - https://www.ncbi.nlm.nih.gov/omim

Medicine

NIH – National Institutes of health
https://www.nih.gov/

OMIM -
https://www.ncbi.nlm.nih.gov/omim

OMM site: https://omim.org/