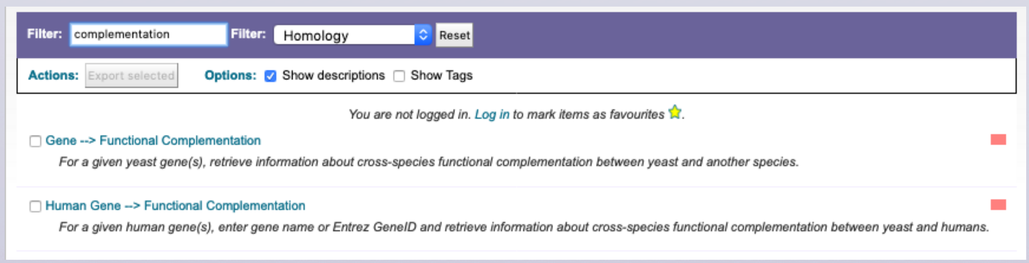
Yeast-Human Cross-Species Complementation and Associations with Disease-related Genes

Rob Nash, Stacia R. Engel, Kevin A. MacPherson, Kalpana Karra, Gail A. Binkley, Travis K. Sheppard, Edith D. Wong, J. Michael Cherry and the SGD Project
Department of Genetics, School of Medicine, Stanford University, Palo Alto, CA

The Saccharomyces Genome Database (SGD; http://www.yeastgenome.org) is a comprehensive resource for curated, molecular and genetic information on the genes and proteins of S. cerevisiae. Model organism genetics holds great promise for advancing our understanding of human gene function and involvement in disease. Elucidating the biology of yeast genes has in many cases provided valuable insight into the function of their homologous human counterparts. With the goal of making connections between yeast genes, their human homologs and associated diseases, we have undertaken a project to collect and display this information at SGD.

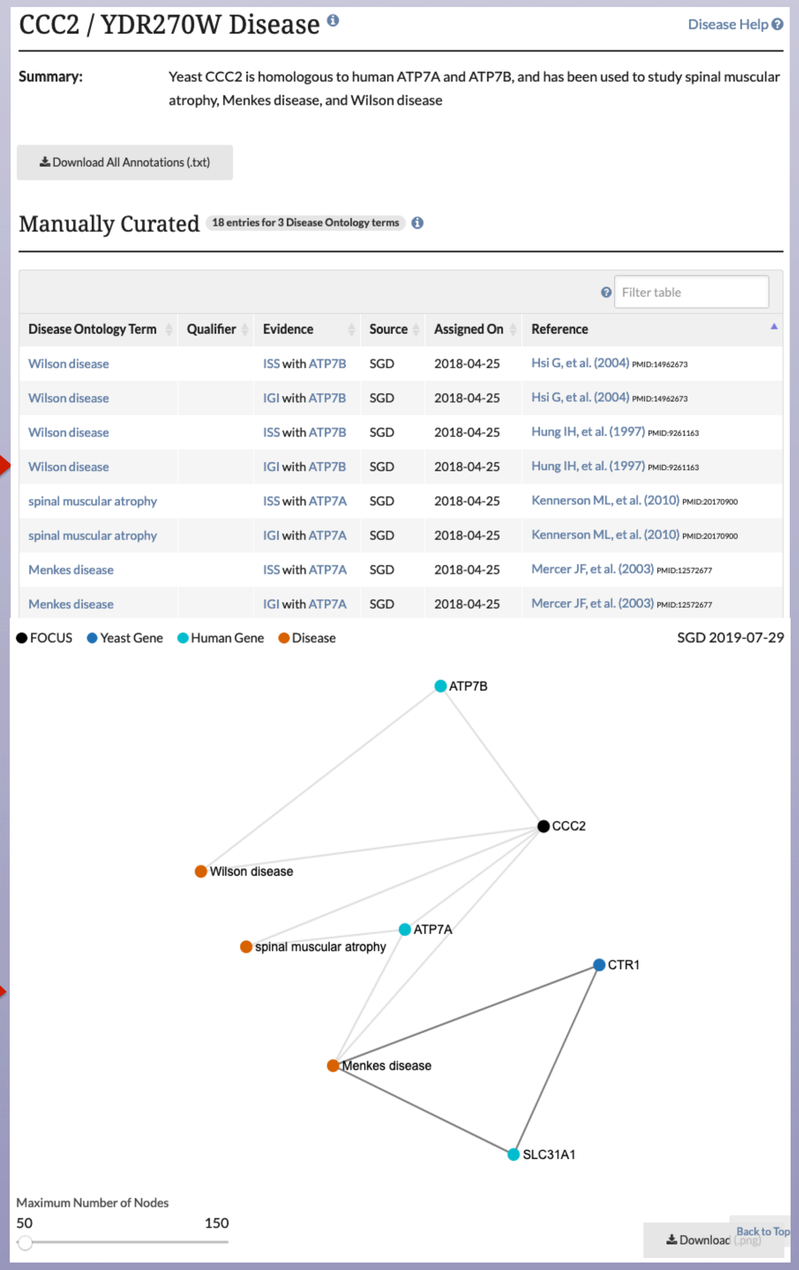
At the start of this project, yeast-human cross-species functional complementation results were collected from the literature and stored in the YeastMine data warehouse where the data can be accessed using preformed template queries. Relevant information was also added to the respective Locus Summary Page descriptions. These functional complementation relationships including some where genes also share homology and the corresponding relationship types have been stored in the database and will soon be displayed on SGD pages. A subset of these human homologs have been determined to be disease associated. For this subset, the corresponding disease ontology (DO) terms were identified and associated with both the human gene and the corresponding yeast homolog, along with supporting information. Diseases associated with human genes that have a computationally determined yeast homolog will soon be included in this set. Disease pages have been designed that include the following pieces of information: disease name, ID and definition from DO, yeast systematic and ORF names, human HGNC-approved gene names (https://www.genenames.org), annotation type (manual vs HTP), evidence code, reference, source and relevant links. A disease summary that has been generated at SGD is included on relevant Locus Summary pages with a link to the browsable Disease page. It is our hope that making this information available to our users will facilitate studies aimed at understanding the biological functions of these genes and the role these genes play in the pathology of disease. Funded by NIH NHGRI [5U41HG001315-18] and NIH NHGRI [U41HG02223-17S1].

YeastMine templates to access all cross-species complementation data



Tabbed page disease content

Summary



Association details:

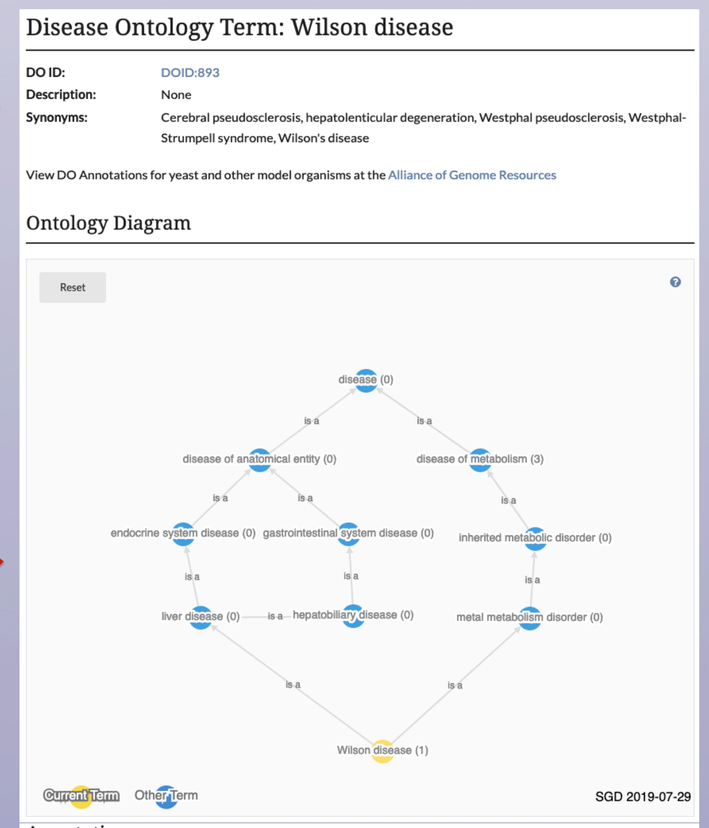
- DO term
- Evidence
- Source
- Assigned on date
- Reference

Network Diagram

- Connects genes to orthologs to the DO terms they share.
- Move the slider to adjust the network and change the number of nodes to display.

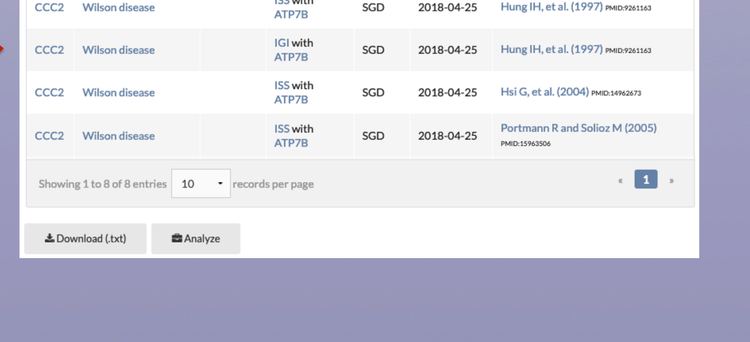
Disease Ontology Term page

Term details



Interactive ontology diagram with relationships.

DO term annotation table.

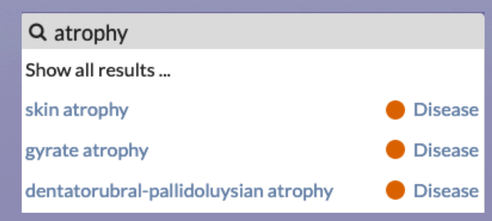


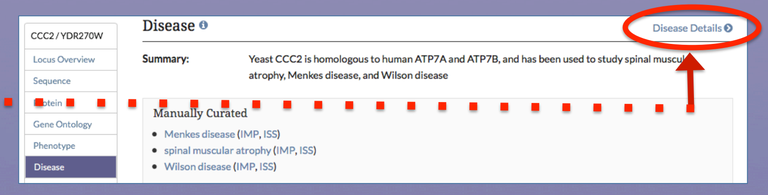
How to access disease associations

1) Searching with DO terms that match a text query, such as “atrophy”, using the search box (top right of SGD webpages).

OR

2) Access the Disease Details link in the summary section on the Locus Summary page or the disease tab.





@yeastgenome
sgd-helpdesk@lists.stanford.edu
https://www.facebook.com/yeastgenome/
https://www.youtube.com/SaccharomycesGenomeData

ALLIANCE of GENOME RESOURCES FOUNDING MEMBER

Stanford MEDICINE