



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



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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

Filter: complementation Filter: Homology Reset

Actions: Export selected Options: Show descriptions Show Tags

You are not logged in. Log in to mark items as favourites.

☐ Gene -> Functional Complementation  
For a given yeast gene(s), retrieve information about cross-species functional complementation between yeast and another species.

☐ Human Gene -> Functional Complementation  
For a given human gene(s), enter gene name or Entrez GeneID and retrieve information about cross-species functional complementation between yeast and humans.

## Tabbed page disease content

Summary

CCC2 / YDR270W Disease

Summary: Yeast CCC2 is homologous to human ATP7A and ATP7B, and has been used to study spinal muscular atrophy, Menkes disease, and Wilson disease

Download All Annotations (.txt)

Manually Curated 18 entries for 3 Disease Ontology terms

Disease Ontology Term	Qualifier	Evidence	Source	Assigned On	Reference
Wilson disease	ISS with ATP7B	SGD	2018-04-25	Hsi G, et al. (2004)	PMID:14962673
Wilson disease	IGI with ATP7B	SGD	2018-04-25	Hsi G, et al. (2004)	PMID:14962673
Wilson disease	ISS with ATP7B	SGD	2018-04-25	Hung IH, et al. (1997)	PMID:9261163
Wilson disease	IGI with ATP7B	SGD	2018-04-25	Hung IH, et al. (1997)	PMID:9261163
spinal muscular atrophy	ISS with ATP7A	SGD	2018-04-25	Kennerston ML, et al. (2010)	PMID:20170900
spinal muscular atrophy	IGI with ATP7A	SGD	2018-04-25	Kennerston ML, et al. (2010)	PMID:20170900
Menkes disease	ISS with ATP7A	SGD	2018-04-25	Mercer JF, et al. (2003)	PMID:12572677
Menkes disease	IGI with ATP7A	SGD	2018-04-25	Mercer JF, et al. (2003)	PMID:12572677

FOCUS Yeast Gene Human Gene Disease

SGD 2019-07-29

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.

Maximum Number of Nodes: 50 150

Download Back to Top

## Disease Ontology Term page

Term details

Disease Ontology Term: Wilson disease

DO ID: D01893

Description: None

Synonyms: Cerebral pseudosclerosis, hepatolenticular degeneration, Westphal pseudosclerosis, Westphal-Strumpell syndrome, Wilson's disease

View DO Annotations for yeast and other model organisms at the Alliance of Genome Resources

Ontology Diagram

Reset

Annotations

Manually Curated 8 entries for 1 gene

Gene	Disease Ontology Term	Qualifier	Evidence	Source	Assigned On	Reference
CCC2	Wilson disease	ISS with ATP7B	SGD	2018-04-25	Hung IH, et al. (1997)	PMID:9261163
CCC2	Wilson disease	IGI with ATP7B	SGD	2018-04-25	Hung IH, et al. (1997)	PMID:9261163
CCC2	Wilson disease	ISS with ATP7B	SGD	2018-04-25	Hsi G, et al. (2004)	PMID:14962673
CCC2	Wilson disease	ISS with ATP7B	SGD	2018-04-25	Portmann R and Solioz M (2005)	PMID:1603006

Showing 1 to 8 of 8 entries 10 records per page

Download (.txt) Analyze

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.

atrophy

Show all results ...

skin atrophy Disease

gyrate atrophy Disease

dentatorubral-pallidoluysian atrophy Disease

CCC2 / YDR270W Disease

Summary: Yeast CCC2 is homologous to human ATP7A and ATP7B, and has been used to study spinal muscular atrophy, Menkes disease, and Wilson disease

Manually Curated

- Menkes disease (IMP, ISS)
- spinal muscular atrophy (IMP, ISS)
- Wilson disease (IMP, ISS)

Disease Details



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



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