



Associating Yeast Genes with Human Disease-related Genes at SGD



Barbara Dunn, 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
Stanford University, School of Medicine, Department of Genetics, Stanford, 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. 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 have also been 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-derived gene names, annotation type (manual vs HTP), evidence code, reference, source and relevant links. A disease summary has been generated at SGD and will be 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].

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.

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

Date Last Reviewed: 2006-10-12

Disease Ontology Term	Qualifier	Evidence	Source	Assigned On	Reference
Wilson disease	IMP	SGD	2018-04-25	Hsi G, et al. (2004) PMID:14962673	
Wilson disease	ISS with ATP7B	SGD	2018-04-25	Hsi G, et al. (2004) PMID:14962673	
Wilson disease	IMP	SGD	2018-04-25	Hung IH, et al. (1997) PMID:9261163	
Wilson disease	ISS with ATP7B	SGD	2018-04-25	Hung IH, et al. (1997) PMID:9261163	
spinal muscular atrophy	IMP	SGD	2018-04-25	Kennerson ML, et al. (2010) PMID:20170900	
spinal muscular atrophy	ISS with ATP7A	SGD	2018-04-25	Kennerson ML, et al. (2010) PMID:20170900	
Menkes disease	IMP	SGD	2018-04-25	Mercer JF, et al. (2003) PMID:12572677	
Menkes disease	ISS with ATP7A	SGD	2018-04-25	Mercer JF, et al. (2003) PMID:12572677	
Wilson disease	IMP	SGD	2018-04-25	Papur OS, et al. (2015) PMID:26004889	
Wilson disease	ISS with ATP7B	SGD	2018-04-25	Papur OS, et al. (2015) PMID:26004889	

Showing 1 to 10 of 18 entries 10 records per page

Maximum Number of Nodes 50 150

Download (png)

Disease Ontology Term page

Term details

Interactive ontology diagram with relationships.

DO term annotation table.

Disease Ontology Term: Menkes disease

DO ID: DOID:1838

Description: None

Synonyms: COPPER TRANSPORT DISEASE, Menkes kinky-hair syndrome, steely hair syndrome

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

Ontology Diagram

Annotations

Manually Curated 10 entries for 2 genes

Gene	Disease Ontology Term	Qualifier	Evidence	Source	Assigned On	Reference
CCC2	Menkes disease	IMP	SGD	2018-04-25	Payne AS and Gitlin JD (1998) PMID:9452509	
CCC2	Menkes disease	ISS with ATP7A	SGD	2018-04-25	Payne AS and Gitlin JD (1998) PMID:9452509	
CCC2	Menkes disease	IMP	SGD	2018-04-25	Voskoboinik I, et al. (2001) PMID:11373292	
CCC2	Menkes disease	ISS with ATP7A	SGD	2018-04-25	Voskoboinik I, et al. (2001) PMID:11373292	
CCC2	Menkes disease	IMP	SGD	2018-04-25	Mercer JF, et al. (2003) PMID:12572677	
CCC2	Menkes disease	ISS with ATP7A	SGD	2018-04-25	Mercer JF, et al. (2003) PMID:12572677	
CCC2	Menkes disease	IMP	SGD	2018-04-25	Tang J, et al. (2008) PMID:18752978	
CCC2	Menkes disease	ISS with ATP7A	SGD	2018-04-25	Tang J, et al. (2008) PMID:18752978	
CTR1	Menkes disease	IMP	SGD	2018-04-25	Lee J, et al. (2000) PMID:10974539	
CTR1	Menkes disease	ISS with SLC31A1	SGD	2018-04-25	Lee J, et al. (2000) PMID:10974539	

Showing 1 to 10 of 10 entries 10 records per page

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.

Q atrophy

Show all results ...

- skin atrophy
- dentatorubral-pallidoluysian atrophy
- muscular atrophy
- optic atrophy

CCC2 / YDR270W

Locus Overview

Sequence

Protein

Gene Ontology

Phenotype

Disease

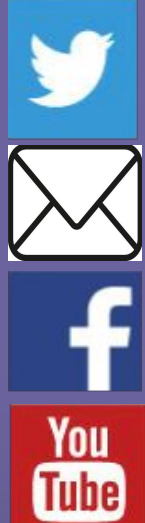
Disease

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



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



ALLIANCE
of GENOME RESOURCES
FOUNDING MEMBER



Stanford
MEDICINE