

## GEPHE SUMMARY

<p>Aquaporin (AQY2) (<a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase=<sup>^</sup>Aquaporin (AQY2)<sup>^</sup>#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001877</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Physiology (<a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category=<sup>^</sup>Physiology<sup>^</sup>#gephebase-summary-title)</p> <p>Growth rate (environment-dependent) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=&lt;sup&gt;^&lt;/sup&gt;Growth rate (environment-dependent)&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Growth rate (environment-dependent)<sup>^</sup>#gephebase-summary-title</a>)</p> <p>freeze-thaw<sup>^</sup>tolerant</p> <p>freeze-thaw<sup>^</sup>sensitive and higher growth rate in sugar-rich medium</p> <p>Taxon A</p> <p>Domesticated (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status=<sup>^</sup>Domesticated<sup>^</sup>#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p> <p>Taxon A Description</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p> <p>Taxon B Description</p>
<p>Saccharomyces cerevisiae (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=&lt;sup&gt;^&lt;/sup&gt;Saccharomyces cerevisiae&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Saccharomyces cerevisiae<sup>^</sup>#gephebase-summary-title</a>)</p> <p>baker's yeast</p> <p>Saccharomyces capensis; Saccharomyces italicus; Saccharomyces oviformis; Saccharomyces uvarum var. melibiosus; baker's yeast; S. cerevisiae; brewer's yeast; ATCC 18824; ATCC:18824; CBS 1171; CBS:1171; NRRL Y-12632; NRRL:Y:12632; Saccharomyces cerevisiae; Saccharomyce cerevisiae; Saccharomyes cerevisiae; Sccharomyces cerevisiae</p> <p>species</p> <p>cellular organisms; Eukaryota; Opisthokonta; Fungi; Dikarya; Ascomycota; saccharomyceta; Saccharomycotina; Saccharomycetes; Saccharomycetales; Saccharomycetaceae; Saccharomyces</p> <p>Saccharomyces () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4930">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4930</a>)</p> <p>4932 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4932">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4932</a>)</p> <p>Yes</p> <p>YPS163 collected from Pennsylvania oak trees</p>	<p>Saccharomyces cerevisiae (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=&lt;sup&gt;^&lt;/sup&gt;Saccharomyces cerevisiae&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Saccharomyces cerevisiae<sup>^</sup>#gephebase-summary-title</a>)</p> <p>baker's yeast</p> <p>Saccharomyces capensis; Saccharomyces italicus; Saccharomyces oviformis; Saccharomyces uvarum var. melibiosus; baker's yeast; S. cerevisiae; brewer's yeast; ATCC 18824; ATCC:18824; CBS 1171; CBS:1171; NRRL Y-12632; NRRL:Y:12632; Saccharomyces cerevisiae; Saccharomyce cerevisiae; Saccharomyes cerevisiae; Sccharomyces cerevisiae</p> <p>species</p> <p>cellular organisms; Eukaryota; Opisthokonta; Fungi; Dikarya; Ascomycota; saccharomyceta; Saccharomycotina; Saccharomycetes; Saccharomycetales; Saccharomycetaceae; Saccharomyces</p> <p>Saccharomyces () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4930">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4930</a>)</p> <p>4932 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4932">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4932</a>)</p> <p>Yes</p> <p>Malaysian strain</p>		

## GENOTYPIC CHANGE

<p>AQP1</p> <p>CO; CHIP28; AQP-CHIP</p> <p>9606.ENSP00000311165 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000311165">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000311165</a>)</p> <p>Belongs to the MIP/aquaporin (TC 1.A.8) family.</p> <p>GO:0042802 : identical protein binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0042802">https://www.ebi.ac.uk/QuickGO/term/GO:0042802</a>)</p> <p>GO:0015079 : potassium ion transmembrane transporter activity</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>P29972 (<a href="http://www.uniprot.org/uniprot/P29972">http://www.uniprot.org/uniprot/P29972</a>)</p> <p>()</p> <p>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
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(<https://www.ebi.ac.uk/QuickGO/term/GO:0015079>)  
GO:0015250 : water channel activity (<https://www.ebi.ac.uk/QuickGO/term/GO:0015250>)  
GO:0008519 : ammonium transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008519>)  
GO:0022857 : transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0022857>)  
GO:0035379 : carbon dioxide transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035379>)  
GO:0015168 : glycerol transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0015168>)  
GO:0005223 : intracellular cGMP-activated cation channel activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005223>)  
GO:0030184 : nitric oxide transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030184>)  
GO:0005267 : potassium channel activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005267>)  
GO:0005372 : water transmembrane transporter activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005372>)

#### GO - Biological Process

GO:0043066 : negative regulation of apoptotic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043066>)  
GO:0019934 : cGMP-mediated signaling  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019934>)  
GO:0006813 : potassium ion transport  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006813>)  
GO:0071260 : cellular response to mechanical stimulus  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071260>)  
GO:0042493 : response to drug (<https://www.ebi.ac.uk/QuickGO/term/GO:0042493>)  
GO:0071280 : cellular response to copper ion  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071280>)  
GO:0003091 : renal water homeostasis  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003091>)  
GO:0071456 : cellular response to hypoxia  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071456>)  
GO:0006833 : water transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0006833>)  
GO:0006972 : hyperosmotic response  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006972>)  
GO:0071300 : cellular response to retinoic acid  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071300>)  
GO:0042476 : odontogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0042476>)  
GO:0070301 : cellular response to hydrogen peroxide  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070301>)  
GO:0071732 : cellular response to nitric oxide  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071732>)  
GO:0015701 : bicarbonate transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015701>)  
GO:0030185 : nitric oxide transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0030185>)  
GO:0045766 : positive regulation of angiogenesis  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045766>)  
GO:0043154 : negative regulation of cysteine-type endopeptidase activity involved in  
apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0043154>)  
GO:0048146 : positive regulation of fibroblast proliferation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048146>)  
GO:0071320 : cellular response to cAMP  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071320>)  
GO:0015696 : ammonium transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015696>)  
GO:0071549 : cellular response to dexamethasone stimulus  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071549>)  
GO:0030950 : establishment or maintenance of actin cytoskeleton polarity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030950>)  
GO:0034644 : cellular response to UV  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0034644>)  
GO:0030157 : pancreatic juice secretion  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030157>)  
GO:0035378 : carbon dioxide transmembrane transport  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035378>)  
GO:0015670 : carbon dioxide transport  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0015670>)  
GO:0006884 : cell volume homeostasis  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006884>)  
GO:0019725 : cellular homeostasis (<https://www.ebi.ac.uk/QuickGO/term/GO:0019725>)  
GO:0071474 : cellular hyperosmotic response  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071474>)  
GO:0071241 : cellular response to inorganic substance  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071241>)  
GO:0071288 : cellular response to mercury ion  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071288>)  
GO:0071472 : cellular response to salt stress  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071472>)

GO:0033326 : cerebrospinal fluid secretion  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0033326)  
 GO:0015793 : glycerol transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015793)  
 GO:0021670 : lateral ventricle development  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0021670)  
 GO:0085018 : maintenance of symbiont-containing vacuole by host  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0085018)  
 GO:0050891 : multicellular organismal water homeostasis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0050891)  
 GO:0046878 : positive regulation of saliva secretion  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0046878)  
 GO:0003097 : renal water transport (https://www.ebi.ac.uk/QuickGO/term/GO:0003097)  
 GO:0035377 : transepithelial water transport  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0035377)

GO - Cellular Component

GO:0016021 : integral component of membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)  
 GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)  
 GO:0016324 : apical plasma membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016324)  
 GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)  
 GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)  
 GO:0005887 : integral component of plasma membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0005887)  
 GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)  
 GO:0016323 : basolateral plasma membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016323)  
 GO:0031965 : nuclear membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0031965)  
 GO:0045177 : apical part of cell (https://www.ebi.ac.uk/QuickGO/term/GO:0045177)  
 GO:0009925 : basal plasma membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0009925)  
 GO:0005903 : brush border (https://www.ebi.ac.uk/QuickGO/term/GO:0005903)  
 GO:0042383 : sarcolemma (https://www.ebi.ac.uk/QuickGO/term/GO:0042383)  
 GO:0031526 : brush border membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0031526)  
 GO:0020003 : symbiont-containing vacuole  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0020003)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title)

Aberration Type

Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title)

Deletion Size

1-9 bp

Molecular Details of the Mutation

aquaporin yeast 2 gene on chromosome 11 - G528 deletion which creates a frameshift the AQY2 gene - tested in assays - GenBank accession numbers for AQY1 and AQY2: GQ848552-74 and GQ870433-54

Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene^#gephebase-summary-title)

Main Reference

Incipient balancing selection through adaptive loss of aquaporins in natural *Saccharomyces cerevisiae* populations. (2010) (https://pubmed.ncbi.nlm.nih.gov/20369021)

Authors

Will JL; Kim HS; Clarke J; Painter JC; Fay JC; Gasch AP

Abstract

A major goal in evolutionary biology is to understand how adaptive evolution has influenced natural variation, but identifying loci subject to positive selection has been a challenge. Here we present the adaptive loss of a pair of paralogous genes in specific *Saccharomyces cerevisiae* subpopulations. We mapped natural variation in freeze-thaw tolerance to two water transporters, AQY1 and AQY2, previously implicated in freeze-thaw survival. However, whereas freeze-thaw-tolerant strains harbor functional aquaporin genes, the set of sensitive strains lost aquaporin function at least 6 independent times. Several genomic signatures at AQY1 and/or AQY2 reveal low variation surrounding these loci within strains of the same haplotype, but high variation between strain groups. This is consistent with recent adaptive loss of aquaporins in subgroups of strains, leading to incipient balancing selection. We show that, although aquaporins are critical for surviving freeze-thaw stress, loss of both genes provides a major fitness advantage on high-sugar substrates common to many strains' natural niche. Strikingly, strains with non-functional alleles have also lost the ancestral requirement for aquaporins during spore formation. Thus, the antagonistic effect of aquaporin function-providing an advantage in freeze-thaw tolerance but a fitness defect for growth in high-sugar environments-contributes to the maintenance of both functional and nonfunctional alleles in *S. cerevisiae*. This work also shows that gene loss through multiple missense and nonsense mutations, hallmarks of pseudogenization presumed to emerge after loss of constraint, can arise through positive selection.

Additional References

RELATED GEPHE

Related Genes

6 (Allantoin permease DAL4, Allantoinase DAL1, Proline specific permease PUT4, Aquaporin (AQY1), BMH1, IRA2) (https://www.gephebase.org/search-criteria?/or+Taxon ID=~4932^/and+Trait=Growth rate/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

2 (https://www.gephebase.org/search-criteria?/or+Gene Gephbase=Aquaporin (AQY2)^/and+Taxon ID=~4932^/or+Gene Gephbase=Aquaporin (AQY2)^/and+Taxon ID=~4932^#gephebase-summary-title)

## EXTERNAL LINKS

## COMMENTS

The paralogs AQY1 and AQY2 may have arisen in the whole-genome duplication (WGD) event in the *Saccharomyces* lineage since all post-WGD species all have two aquaporins whereas most pre-WGD species have a single ortholog (Dana Wohlbach and A.P.G., unpublished).