GEPHE SUMMARY

Gephebase Gene

Aquaporin (AQY1) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Aquaporin (AQY1)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001872

Main curator

Courtier

PHENOTYPIC CHANGE

Trait Category

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait

Growth rate (environment-dependent) (https://www.gephebase.org/search-criteria?/and+Trait=^Growth rate (environment-dependent)^#gephebase-summary-title)

Trait State in Taxon A

freeze-thawâ€“tolerant

Trait State in Taxon B

freeze-thawâ€“sensitive and higher growth rate in sugar-rich medium

Ancestral State

Taxon A

Taxonomic Status

Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A	Taxon B
Latin Name	Latin Name
Saccharomyces cerevisiae (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Saccharomyces cerevisiae^#gephebase-summary-title)	Saccharomyces cerevisiae (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Saccharomyces cerevisiae^#gephebase-summary-title)
Common Name	Common Name
baker's yeast	baker's yeast
Synonyms	Synonyms
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; Saccaromyces cerevisiae; Saccharomyce cerevisiae; Saccharomyes cerevisiae; Sccharomyces cerevisiae	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; Saccaromyces cerevisiae; Saccharomyce cerevisiae; Saccharomyes cerevisiae; Sccharomyces cerevisiae
Rank	Rank
species	species
Lineage	Lineage
cellular organisms; Eukaryota; Opisthokonta; Fungi; Dikarya; Ascomycota; saccharomyceta; Saccharomycotina; Saccharomycetes; Saccharomycetales; Saccharomycetaceae; Saccharomyces	cellular organisms; Eukaryota; Opisthokonta; Fungi; Dikarya; Ascomycota; saccharomyceta; Saccharomycotina; Saccharomycetes; Saccharomycetales; Saccharomycetaceae; Saccharomyces
Parent	Parent
Saccharomyces () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4930)	Saccharomyces () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4930)
NCBI Taxonomy ID	NCBI Taxonomy ID
4932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4932)	4932 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 4932)
is Taxon A an Infraspecies?	is Taxon B an Infraspecies?
Yes	Yes
Taxon A Description	Taxon B Description
YPS163 - collected from Pennsylvania oak trees	laboratory strain S288c

GENOTYPIC CHANGE

Generic Gene Name

AQP1

Synonyms

CO; CHIP28; AQP-CHIP

String

9606.ENSP00000311165 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000311165)

Sequence Similarities

Belongs to the MIP/aquaporin (TC 1.A.8) family.

GO - Molecular Function

GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)

GO:0015079 : potassium ion transmembrane transporter activity

UniProtKB Homo sapiens

P29972 (http://www.uniprot.org/uniprot/P29972)

GenebankID or UniProtKB

()

(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)

Mutation #1

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

aquaporin yeast 1 gene on chromosome 12 - V121M and P255T - both mutations have an effect on water transport - tested in Xenopus oocytes - GenBank accession numbers for AQY1 and AQY2: GQ848552-74 and GQ870433-54

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	ATG	-
Amino-acid	Val	Met	121

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

Aquaporins in Saccharomyces. Genetic and functional distinctions between laboratory and wild-type strains. (1998) (https://pubmed.ncbi.nlm.nih.gov/9765289)

Mutation #2

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

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

aquaporin yeast 1 gene on chromosome 12 - V121M and P255T - both mutations have an effect on water transport - tested in Xenopus oocytes - GenBank accession numbers for AQY1 and AQY2: GQ848552-74 and GQ870433-54

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Pro	Thr	255

Main Reference

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

Authors

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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

Aquaporins in Saccharomyces. Genetic and functional distinctions between laboratory and wild-type strains. (1998) (https://pubmed.ncbi.nlm.nih.gov/9765289)

RELATED GEPHE

Related Genes

6 (Allantoin permease DAL4, Allantoinase DAL1, Proline specific permease PUT4, Aquaporin (AQY2), 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 Gephebase=^Aquaporin (AQY1)^/and+Taxon ID=^4932^/or+Gene Gephebase=^Aquaporin (AQY1)^/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). @SeveralMutationsWithEffect