



Homology and Disease curation at SGD: budding yeast as a model for eukaryotic biology



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The foundation for much of our understanding of basic cellular biology has been learned from the budding yeast *Saccharomyces cerevisiae*, and studies with yeast have provided powerful insights into human genetic diseases and the cellular pathways in which they are involved. This utility of yeast as a model for human disease arises from the biochemical unity that underlies all forms of life. Recent work with humanized yeast (in which yeast genes have been replaced with human orthologs) and humanized yeast proteins (in which key residues have been altered to match the human sequence) has demonstrated extensive conservation of ancestral functions through time and across taxa. In order to promote and support the ways in which yeast and yeast research can inform genetic medicine, the *Saccharomyces* Genome Database (SGD; www.yeastgenome.org) is providing comprehensive curation for human disease-related genes and their yeast homologs, including high quality manually curated information regarding functional complementation and conserved function. Curated information for yeast genes is displayed on Homology pages and Disease pages at SGD in ways that allow data mining and encourage innovation for researchers studying both yeast and other organisms. These efforts are part of our continuing mission to educate students, enable bench researchers, and facilitate scientific discovery. *SGD is supported by a grant from the NHGRI (U41 HG001315).*

Yeast Gene	Human Gene	Yeast Gene	Human Gene	Yeast Gene	Human Gene
ACO1	ACO2	GUF1	GUF1	RAD5	HLTF
ACT1	ACTA2	GUK1	GUK1	RAD50	RAD50
ACT1	ACTG1	HEM12	UROD	RAD51	RAD51
AFG3	AFG3L2	HEM15	FECH	RAD53	CHEK2
AFG3	SPG7	HEM25	SLC25A38	RAS1	HRAS
AGC1	SLC25A10	HEM3	HMB5	RAS1	KRAS
AGC1	SLC25A13	HEM4	UROS	RAS2	KRAS
AGX1	AGXT	HER2	QSOX1	RCH1	SLC10A7
AIM22	LIFT1	HFD1	ALDH3A2	RED1	SYCP2
API1	WDR1	HJL1	DNAA2	RER2	DHDDS
ALA1	AARS1	HNT1	HINT1	RET1	POLR3B
ALA1	AARS2	HRD1	SYVN1	RFT1	RFT1
ALG1	ALG1	HRP1	HNRNPDL	RFT1	SEC31A1
ALG12	ALG12	HRQ1	RECQL4	RHO1	RAC1
ALG3	ALG3	HRR25	CASK	RIO1	RIOK1
ALG5	ALG5	HRR25	CSNK1A1	RNH201	RNA5E2A
ALG6	ALG6	HRR25	CSNK1D	RNH202	RNA5E2B
ALG8	ALG8	HRT1	RBX1	RNH203	RNA5E2C
ALG9	ALG9	HSC82	HS90A01	RPB7	POLR2G
ALR1	MAGT1	HSH155	SF3B1	RPC19	POLR1D
ALR1	TUSC3	HSP31	PARK7	RPC40	POLR1C
AMD1	AMPD2	HSP32	PARK7	RPL10	RPL10
ARC19	ARPC4	HSP33	PARK7	RPL30	RPL30
ARD1	NAA10	HST1	HARS1	RPL33A	RPL35A
ARH1	FDXR	HYM1	CAB39	RPN5	PSMD12
ARV1	ARV1	HYM1	CAB39L	RP031	POLR3A
ATG1	ULK1	ICP55	XPINPEP3	RPT1	PSM2
ATG5	ATG5	ICT1	ABHD5	RPT3	SPAST
ATP12	ATPAF2	IDH1	IDH3B	RPT6	PSM5
ATP15	ATP5F1E	IDH2	IDH3B	RPT6	VCP
ATP6	MT-ATP6	ID1	ID1	RRP40	EXOS3
BCS1	BCS1L	IDP1	IDH1	RS1	RS1
BETS	TRAPP1	IDP2	IDH1	RS6	NEDD4
BP1	LYST	IFA38	HSD17B12	SAC5	LCF1
BRF1	BRF1	IFA38	HSD17B3	SAC6	PL53
BR2	SNRNP200	IK3	ELP1	SAH1	AHCY
CAB2	PPCS	IML1	DEPDC5	SAR1	SAR1B
CAB4	COASY	IRA1	BANF1	SC2	NIPBL
CAK1	CDK7	IRA1	NF1	SC01	SC01
CAK1	TKB1	IRA2	NF1	SC02	SC02
CAR2	OAT	ISM1	IARS2	SDH1	SDHA
CCA1	TRNT1	ISU1	ISCU	SDH2	SDHB
CC2	ATP7A	ISU2	ISCU	SDH4	SDHD
CC2	ATP7B	KA1	OSGPE	SDH5	SDHAF2
CC5	CC5	KIN28	ACVR1K	SDH6	SDHAF1
CC7	BBS10	KIN28	AKT2	SDH7	SDHAF3
CCD14	CDC14A	KIN28	BMPPR1B	SDO1	SBDS
CDC20	CDC20	KIN28	GRK4	SEC12	ELP2
CDC28	CDK1	KRS1	KARS1	SEC12	GNB1L
CDC28	CDK2	LAG1	CERS2	SEC12	IFT122
CDC34	EIF4E	LAS17	WAS	SEC14	SEC14L1
CDC34	UBE2B	LEU5	SLC25A16	SEC23	SEC23A
CDC45	CDC45	LIP2	LIP2	SEC23	SEC23B
CDC48	VCP	LIP2	LIP2	SEC4	RAB33B
CDH1	LIG1	LSM4	LSM4	SEC53	PM22
CDH1	FZR1	MAS1	PMPCB	SEC59	DOLK
CD51	CD52	MCD1	RAD21	SE1	B5C12
CHD1	CHD1	MCM4	MCM4	SEN1	SETX
CIN2	RP2	MCM5	MCM5	SEV1	ATL1
CIN8	KIF11	MDH1	MDH2	SGF73	ATXN7
CK1	CKA	MDM1	SNX14	SGS1	ABC4
CKS1	CKS1B	MDM35	TRIA1	SGS1	BLM
CLP1	CLP1	MEL1	ATM	SGS1	WRN
CMD1	CALM1	MEP2	RHCG	SGV1	MOK
CMD1	CALM2	MES1	MARS1	SHH4	SDHD
CMD1	CALM3	MGM1	OPA1	SIL1	SIL1
COA6	COA6	MIA40	CHCHD4	SIR2	SIRT1
COA2	COA2	MIP1	POLG	SIS1	DNAJB6
COA3	COA3	MLH1	MLH1	SK3	SK3
COA5	COA5	MLH3	MLH3	SLM3	TRMU
COA6	COA6	MNL1	EDEM1	SMP3	PIGZ
COA8	COQ8A	MOD5	TRIT1	SM3	SUMO1
COA8	COQ8B	MPC1	MPC1	SNF2	SMARCA4
COQ9	COQ9	MPE1	RBBP6	SNF4	PRKAG2
COX10	COX10	MPS1	GRK3	SN04	PARK7
COX12	COX6B1	MRE11	MRE11	SNU13	SNU13
CR1	SLC25A20	MRM2	MRM2	SOD1	SOD1
CR51	CARS1	MRPS12	MRPS12	SOD2	SOD2
CSE1	CSE1L	MSB3	USP6NL	SP014	PLD1
CSE4	CENPA	MSB4	USP6NL	SPT14	PIGA
CSE4	H2BW1	MSH2	MSH2	SPT15	TBP
CTR1	SLC31A1	MSH3	MSH3	SRB4	MED17
CUES	TOLLIP	MSH6	MSH6	SRV2	CAP1
CUP9	MEI51	MSR1	RARS2	SSC1	HSP9A
CUP9	PKN0X1	MSS4	PIPSK1A	SSL2	ERC3
CUP9	PKN0X2	MSS4	PIPSK1B	SSS1	SEC61G
CY1	CY1	MSW1	WARS2	STE24	ZMPSTE24
CY1	CYCS	MT01	MT01	STE6	CFR
CY3	HCCS	MYG1	MYG1	SUI2	EIF251
CY1M	PITRM1	MYO4	MYO5A	SUI3	EIF252
CYS4	CBS	MYO4	MYO5B	SYM1	MPV17
DBP2	DDX5	MZM1	LYRM7	TAF21	TAFAZIN
DDL1	DDHD1	NAM2	LARS2	TCAT7	TRAPP2L
DDL1	DDHD2	NAN1	COP1	TDP1	TDP1
DFG10	SRD5A3	NAN1	PAFAH1B1	TEF2	EEF1A2
DFR1	DHFR	NAT1	NAA15	TEL1	ATM
DIB1	PIGZ	NCR1	NPC1	THG1	THG1L
DIB1	TXNL4A	NDC80	NDC80	TH7	SLC19A2
DIC1	SLC25A10	ND1	MT-ND4	TH80	TPK1
DIS3	DIS3	NOB1	NOB1	THS1	TARS1
DNA2	BLM	NOP14	NOP14	TIF11	EIF1AX
DNA2	DNA2	NOP2	NOP2	TIF4631	EIF4G1
DNA2	WRN	NPC2	NPC2	TIM13	TIMM13
DNF2	ATP8B1	NPR2	NPR2	TIM44	TIMM44
DNM1	DNM1L	NPR3	NPR3	TIM8	TIMM8A
DOG2	PUDP	NUP145	NUP98	TOP2	TOP2A
DPM1	DPM1	NUS1	NUS1	TOP2	TOP2B
DUT1	DUT	OAR1	CBR4	TOR1	MTOR
DYN1	DYNC1H1	ORC1	ORC1	TP1	TP1
ECM16	DHX37	ORC4	ORC4	TRM10	TRMT10A
ECO1	ESCO2	ORC6	ORC6	TRM5	TRMT5
EFR3	EFR3A	ORM2	ORMD1	TRM7	FTSJ1
EMG1	EMG1	ORM2	ORMD3	TRM732	THADA
ENV9	RDH12	ORT1	SLC25A15	TRM82	WDR4
ERG1	SQLE	PAC10	VBP1	TR520	TRAPP2
ERG11	CYP19A1	PAF1	PAF1	TR520	ZNF547
ERG12	MVK	PAH1	LPIN1	TR523	TRAPP4
ERG26	NSDHL	PAH1	LPIN2	TR21	ELAC2
ERG27	HSD17B7	PAH1	LPIN3	TC10	KDSR
ERS1	CF12	PAM16	PAM16	TC13	TECR
ERS1	COG4	PCM1	PGM3	TT1	TT1
ERS1	CTNS	PDA1	PDHA1	TUB1	TUBA1A
ERS1	SLC16A1	PEP5	VPS11	TUB2	TUBB1
ERV1	GFR	PET9	SLC25A4	TUB2	TUBB2B
ES1	KATS	PEX1	PEX1	TUB2	TUBB6
FAA1	ACSBG1	PEX13	PEX13	TUB3	TUBA1A
FAA1	ACSL1	PEX6	PEX6	TUB3	TUBA1B
FAA1	ACSL3	PFK1	PFKM	TUB3	TUBA1C
FAA1	ACSL4	PFK2	PFKM	TUB4	TUBG1
FAA1	ACSL5	PFY1	PFN1	TY51	YARS1
FAA1	ACSL6	PGK1	PGK1	UBC9	UBE2I
FAA1	SLC27A2	PGK1	PGK2	UBP6	USP14
FAA1	SLC27A4	PHO8	ALPG	UBR1	UBR1
FAA4	ACSBG1	PHO8	ALPL	UF01	UF01
FAA4	ACSL1	PHO8	ALPP	UF04	HGNC:12306
FAA4	ACSL3	PHO85	CDK5	UG01	SLC25A46
FAA4	ACSL4	PHO92	YTHDF2	UGP1	UGP2
FAA4	ACSL5	PHS1	HACD2	ULP2	PML
FAA4	ACSL6	PIC2	SLC25A4	UR46	CMPK1
FAA4	SLC27A2	PIF1	PIF1	UTP4	UTP4
FAA4	SLC27A4	PI5	CDIP1	VAS1	VARS1
FAD1	FLAD1	PMC1	ATP2C1	VAS1	VARS2
FIG4	FIG4	PMR1	ATP2C1	VMA2	ATPGV1B1
FLX1	SLC25A32	PMT4	POMT1	VMA2	ATPGV1B2
FMN1	RFK	PNG1	NGLY1	VMA21	VMA21
FOL2	GCH1	POB3	SSRP1	VMA22	CCDC115
FOX2	HSD17B4	POL2	POLE	VMS1	ANKZF1
FPR2	FKBP2	POL3	POLD1	VPH1	ATPGV0A4
FUM1	FH	POL31	POLD2	VPH1	TCIRG1
FZ01	MFN2	POL32	POLD3	VPH2	TMEM199
GAL10	GALE	PPA2	PPA2	VPS13	VPS13A
GAL7	GALT	PRE5	PSMA1	VPS13	VPS13B
GAL7	UGP2	PRP16	DHX38	VPS13	VPS13C
GD11	EIF253	PRP31	PRPF31	VPS13	VPS13D
GD2	EIF2B4	PRP38	PRPF38B	VPS30	BECN1
GD6	EIF2B5	PRP8	PRPF8	VPS35	VPS35
GD7	EIF2B2	PR51	PRP51	VPS53	VPS53
GN3	EIF2B1	PR54	PRP51	WHI2	KCTD7
GD1	GD12	PSD1	PISD	YBLO36C	PLBPB
GDT1	TMEM165	PSD1	IPO5	YDC1	ACER3
GEF1	CLN5	PUP3	PSMB3	YDLO85C-A	SERF1A
GEF1	CLN7	PUS7	PUS7	YFH1	FTL
GIS2	CNBP	PUT2	ALDH4A1	YFH1	FXN
GLC3	GBE1	PXA1	ABCD1	YHC3	CLN3
GLC7	PPP1CA	PXA1	ABCD2	YME1	YME1L1
GLC7	PPP1CB	PXA2	ABCD1	YMR1	MTM1
GLE1	GLE1	PXA2	ABCD2	YMR1	MTMR2
GLR1	PYROXO1	PXR1	PINX1	YNG2	ING3
GNA1	GNPNA1	RAD1	ERC4	YOR1	CFTR
GNA1	SAT1	RAD14	XPA	YPC1	ACER3
GPA1	GNAS	RAD2	ERC5	YPK9	ATP13A2
GPI10	PIGB	RAD27	FEN1	YPT1	RAB1A
GR51	GARS1	RAD3	ERC2	YTA12	AFG3L2
GS2	GSS	RAD30	POLH	YTA12	SPG7
GTR2	RRAGC	RAD4	XPC	YTA7	ATA2D
				ZWF1	G6PD

Goal: promote ways in which yeast and yeast research can inform genetic medicine

Yeast and human homologs • Disease associations • Functional complementation

Does your favorite gene have a human disease homolog?

Disease associations

curated by SGD curators from the primary literature

MIP1 / YOR330C Homology

Standard Name: MIP1¹
Systematic Name: YOR330C
SGD ID: SGD:S00005857
Feature Type: ORF, Verified
Description: Mitochondrial DNA polymerase gamma; single subunit of mitochondrial DNA polymerase in yeast, in contrast to metazoan complex of catalytic and accessory subunits; polymorphic in yeast, petites occur more frequently in some lab strains; human ortholog POLG complements yeast mip1 mutant; mutations in human POLG associated with Alpers-Huttenlocher syndrome (AHS), progressive external ophthalmoplegia (PEO), parkinsonism, other mitochondrial diseases^{2,3,4,5,6,7,8,9}

Name Description: Mitochondrial DNA Polymerase¹
Comparative Info:

MIP1 / YOR330C Disease

Summary: Yeast MIP1 is homologous to human POLG, and has been used to study the pathogenesis of chronic progressive external ophthalmoplegia, Alpers syndrome, progeria, and peripheral nervous system disease

[Download All Annotations \(.txt\)](#)

Manually Curated 12 entries for 4 Disease Ontology terms

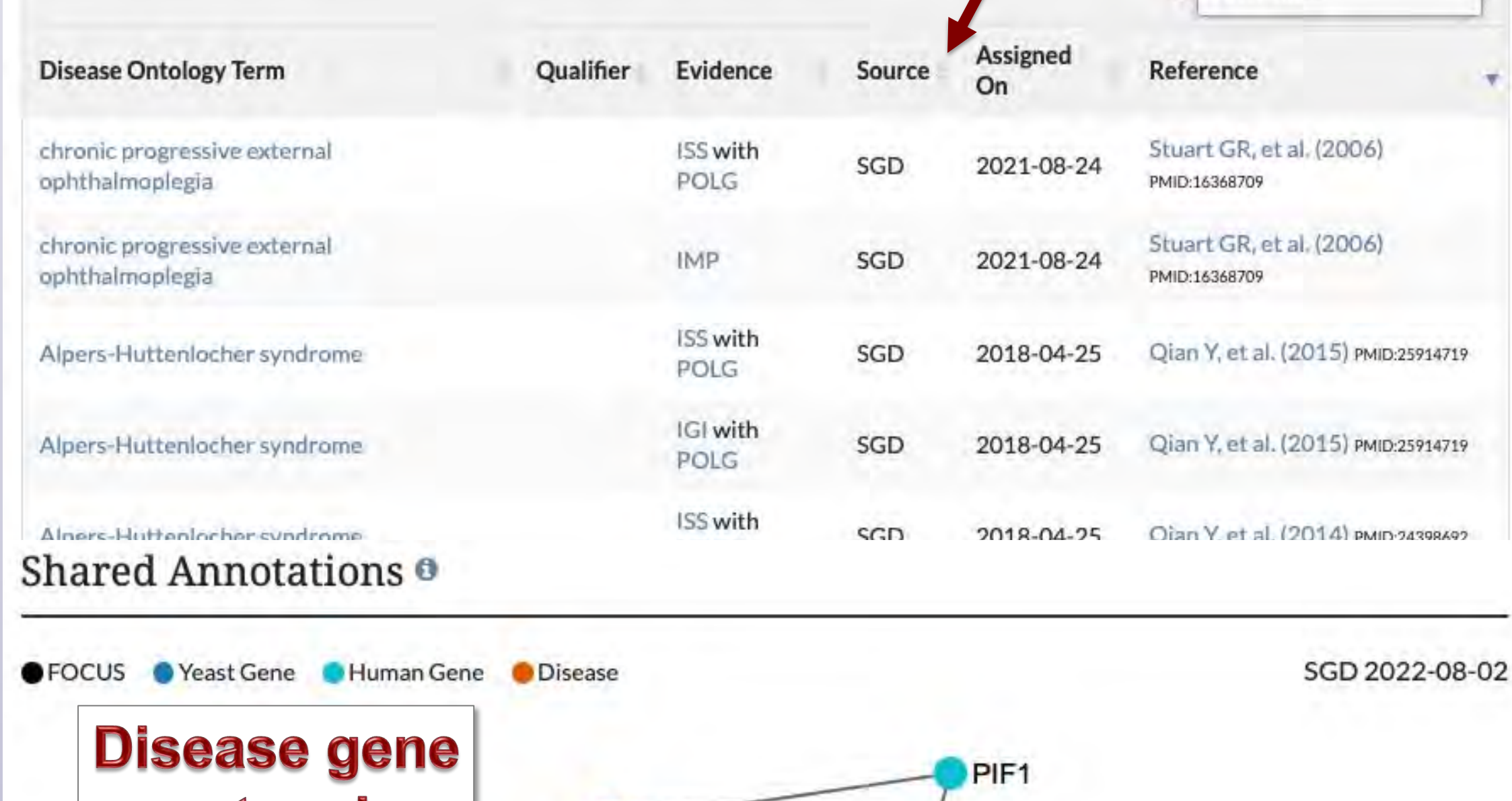
curated by SGD curators from the primary literature

Homologs

6 entries for 6 homologs

Species	Gene ID	Gene name	Source
<i>Caenorhabditis elegans</i>	WB:WBGene00013258	polg-1	Alliance
<i>Danio rerio</i>	ZFIN:ZDB-GENE-060303-1	polg	Alliance
<i>Drosophila melanogaster</i>	FB:EBgn0004406	PolG1	Alliance
<i>Homo sapiens</i>	HGNC:9179	POLG	Alliance
<i>Mus musculus</i>	MGI:1196389	Polg	Alliance
<i>Rattus norvegicus</i>	RGD:620057	Polg	Alliance

source: API @ Alliance of Genome Resources



Functional Complementation

2 entries

curated by SGD curators from the primary literature

Species	Gene ID	Strain background	Direction	Details	Source	Reference
<i>Homo sapiens</i>	HGNC:9179	S288C	other complements yeast	Human POLG complements the mitochondrial genome instability exhibited by the <i>S. cerevisiae</i> mip1 null mutant, but the Alpers disease-associated POLG-S305R mutant fails to complement; the Alpers disease-associated POLG-P1073L allele confers partial stability of the mitochondrial genome with elevated mutation frequency.	SGD	Qian Y, et al. (2015) PMID:25914719
<i>Homo sapiens</i>	HGNC:9179	S288C	other complements yeast		SGD	Qian Y, et al. (2014) PMID:24398692

YeastMine templates for complementation and disease

Search and retrieve *S. cerevisiae* data with YeastMine, populated by SGD and powered by InterMine.

Data Updated on: Jul-26-2022

Templates

Templates are predefined queries, each has a simple form and a description. Click on a template to run it, you can search for templates by category.

Filter: complement | Filter: -- all categories -- | Reset

Actions: Expect Unselected | 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 another species.
- Retrieve Functional Complementation for genes.
For gene(s), retrieve information about cross-species functional complementation between yeast and another species.

YeastMine templates for complementation and disease

Search and retrieve *S. cerevisiae* data with YeastMine, populated by SGD and powered by InterMine.

Data Updated on: Jul-26-2022

Templates

Templates are predefined queries, each has a simple form and a description. Click on a template to run it, you can search for templates by category.

Filter: disease | Filter: -- all categories -- | Reset

Actions: Expect Unselected | Options: Show descriptions | Show Tags

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

- DO Term Identifier -> Genes
Retrieve all Disease Ontology (DO) annotations for a specified gene. Wild card queries (such as "YAL") are supported.
- DO Term Identifier -> Genes
Retrieve all genes annotated to a particular Disease Ontology (DO) ID with a particular evidence code.
- Yeast gene -> OMIM human homolog(s) -> OMIM Disease Phenotype(s)
Retrieve human homolog(s) of yeast gene(s) and any of their associated OMIM disease phenotypes.