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).*

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?

MIP1 / YOR330C Homology

Standard Name:	MIP1 [1]
Systematic Name:	YOR330C
SGD ID:	SGD:S000005857
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 Polymerase [1]
Comparative Info:	

Yeast and model organism homologs

source: API @ Alliance of Genome Resources

Homologs — 6 entries for 6 homologs

Species	Gene ID	Gene name	Source
Caenorhabditis elegans	WB:WBGene00013258	polg-1	Alliance
Danio rerio	ZFIN:ZDB-GENE-060303-1	polg	Alliance
Drosophila melanogaster	FB:FBgn0004406	PolG1	Alliance
Homo sapiens	HGNC:9179	POLG	Alliance
Mus musculus	MGI:1196389	Polg	Alliance
Rattus norvegicus	RGD:620057	Polg	Alliance

Functional Complementation — 2 entries

Functional complementation

curated by SGD curators from the primary literature

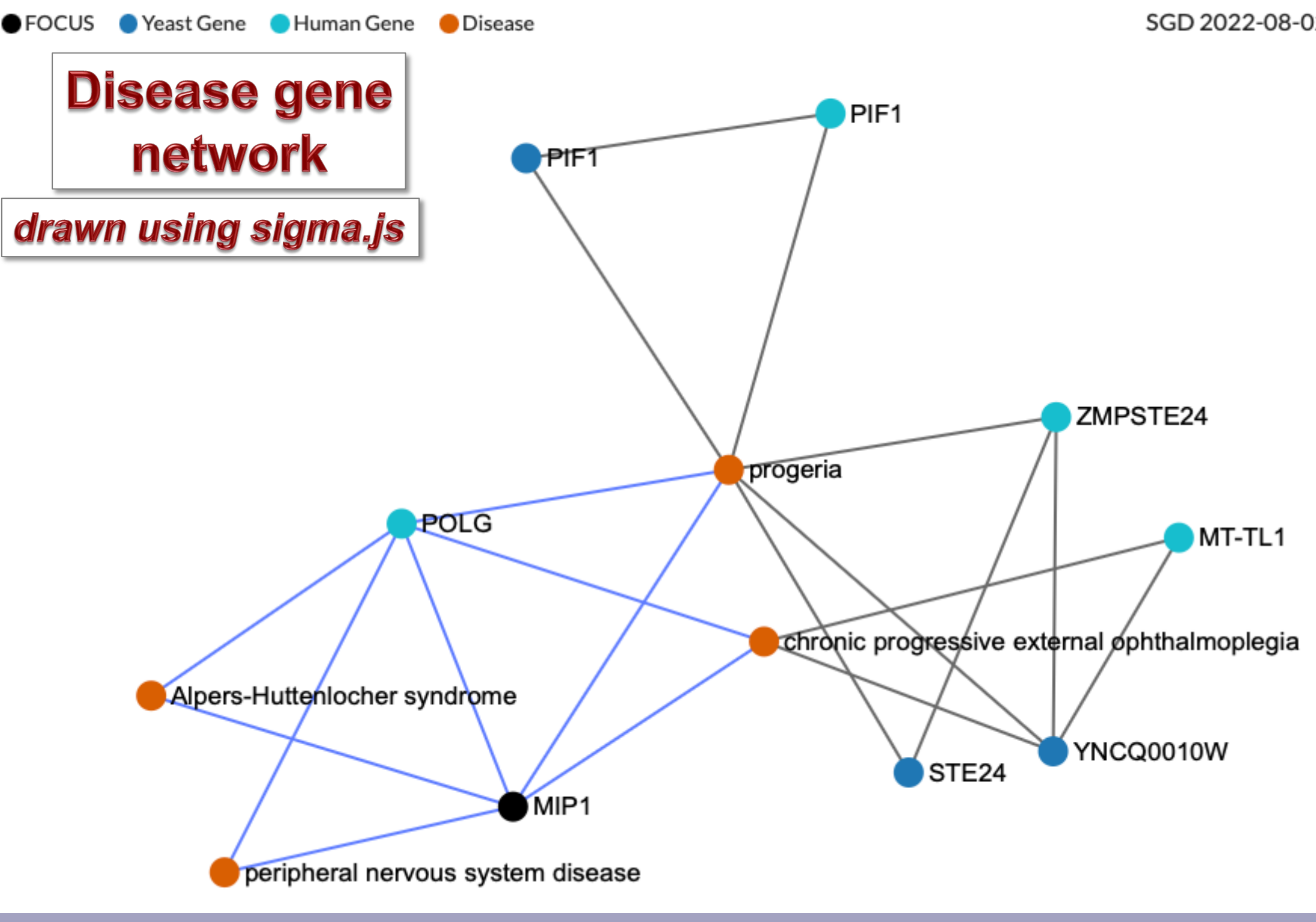| Species	Gene ID	Strain background	Direction	Details	Source	Reference
Homo sapiens	HGNC:9179	S288C	other complements yeast	Human POLG complements the mitochondrial genome instability exhibited by the S. cerevisiae mip1 null mutant, but the Alpers disease-associated POLG-S305R mutant fails to complement; 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
Homo sapiens	HGNC:9179	S288C	other complements yeast		SGD	Qian Y, et al. (2014) PMID:24398692

MIP1 / YOR330C Disease

Summary: Yeast MIP1 is homologous to human POLG, and has been used to ... ophthalmoplegia, Alpers syndrome, progeria, and peripheral nervous system disease

⬇ Download All Annotations (.txt)

Manually Curated — 12 entries for 4 Disease Ontology terms

Disease associations

curated by SGD curators from the primary literature

Disease Ontology Term	Qualifier	Evidence	Source	Assigned On	Reference
chronic progressive external ophthalmoplegia		ISS with POLG	SGD	2021-08-24	Stuart GR, et al. (2006) PMID:16368709
chronic progressive external ophthalmoplegia		IMP	SGD	2021-08-24	Stuart GR, et al. (2006) PMID:16368709
Alpers-Huttenlocher syndrome		ISS with POLG	SGD	2018-04-25	Qian Y, et al. (2015) PMID:25914719
Alpers-Huttenlocher syndrome		IGI with POLG	SGD	2018-04-25	Qian Y, et al. (2015) PMID:25914719
Alpers-Huttenlocher syndrome		ISS with	SGD	2018-04-25	Qian Y, et al. (2014) PMID:24398692

Shared Annotations

Disease gene network

drawn using sigma.js

FOCUS · Yeast Gene · Human Gene · Disease

SGD 2022-08-02

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

Data Updated on: Jul-26-2022

YeastMine templates for complementation and disease

Home | Templates | Lists | QueryBuilder | Tools | Regions | Data Sources | API

Templates (Filter: complement)
Templates are predefined queries, each has a simple form and a description...

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

Templates (Filter: disease)
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.

- Gene --> DO Terms
  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 and any of their associated OMIM disease phenotypes.

Contact us! sgd-helpdesk@lists.stanford.edu • www.yeastgenome.org/submitData @yeastgenome

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	SLC25A40	HMBS	HMBS	RAS1	KRAS
AGC1	SLC25A13	HEM4	UROS	RAS2	KRAS
AGX1	AGXT	HER2	HER2	RCH1	SLC10A7
AIP1	WDR1	HLJ1	DNAJA2	RER2	SYCP2
ALA1	AARS1	HINT1	HINT1	RET1	POLR3B
ALA1	AARS2	HRD1	SYVN1	RFT1	RFT1
ALG1	ALG1	HRD1	HNRNPDL	RFT1	SEC61A1
ALG12	ALG12	HRQ1	RECQL4	RHO1	RAC1
ALG3	ALG3	HRR25	CASK	RIO1	RIOK1
ALG5	ALG5	HRR25	CSNK1A1	RNH201	RNASEH2A
ALG6	ALG6	HRR25	CSNK1D	RNH202	RNASEH2B
ALG8	ALG8	HRT1	RBX1	RNH203	RNASEH2C
ALG9	ALG9	HSCB2	HSP90AB1	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	HYM1	CAB39	RPL35A	RPL35A
ARH1	FDXR	HYM1	CAB39L	RPO31	POLR3A
ARV1	ARV1	HYM1	CAB39	RPT1	PSMC2
ATG1	ULK1	ICP55	XPNPEP3	RPT1	PSMC2
ATG5	ATG5	ICT1	ABHD5	RPT3	PSMC4
ATP12	ATPAF2	IDH1	IDH3B	RPT6	PSMC5
ATP15	ATP5F1E	IDH2	IDH3B	RRP40	EXOSC3
ATP6	MT-ATP6	IDI1	IDI1	RRS1	RRS1
BCS1	BCS1L	IDP1	IDH1	RRS1	NEDD4
BET5	TRAPPC1	IDP2	IDH2	SAC6	LCP1
BPH1	LYST	IFA38	HSD17B12	SAC6	PLS3
BRF1	BRF1	IFA38	HSD17B3	SAH1	AHCY
BRR2	SNRNP200	IKI3	ELP1	SAH1	AHCY
CAB2	PPCS	IML1	DEPDC5	SAR1	SAR1B
CAB4	COASY	IRA1	BANF1	SCC2	NIPBL
CAK1	CDK7	IRA1	NF1	SCO1	SCO1
CAK1	TBK1	IRA2	NF1	SCO2	SCO2
CAR2	OAT	ISM1	AARS2	SDH2	SDHB
CCA1	TRNT1	ISU1	ISCU	SDH4	SDHD
CCC2	ATP7A	ISU2	ISCU	SDH4	SDHB
CCC2	ATP7B	KAE1	OSGEP	SDH5	SDHAF2
CCS1	CCS	KIN28	ACVR1C	SDH6	SDHAF3
CCT5	BBS10	KIN28	AKT2	SDH7	SDHAF3
CDC14	CDC14A	KIN28	BMPR1B	SDO1	SBDS
CDC20	CDC20	KIN28	GRK4	SEC12	ELP2
CDC28	CDK1	KRS1	KARS1	SEC12	GBF1
CDC28	CDK2	LAG1	CERS2	SEC12	IFT122
CDC33	EIF4E	LAS17	WAS	SEC14	SEC14L1
CDC34	UBE2B	LEU5	SLC25A16	SEC23	SEC23A
CDC45	CDC45	LIP2	LIP2	SEC23	SEC23B
CDC48	VCP	LIG1	DLD	SEC4	RAB8A
CDC9	LIG1	LSM4	LSM4	SEC53	PMM2
CDH1	FZR1	MAS1	PMPCB	SEC59	DOLK
CDS1	CDS2	MCD1	RAD21	SEI1	BSCL2
CHD1	CHD1	MCM4	MCM4	SEN1	SETX
CIN2	RP2	MCM5	MCM5	SEY1	ATL1
CIN8	KIF11	MDH1	MDH2	SGF73	ATXN7
CKI1	CHKA	MDH1	SNX14	SGS1	ABCG4
CKS1	CKS1B	MDM35	TRIAP1	SGS1	BLM
CLP1	CLP1	MEC1	ATM	SGS1	WRN
CMD1	CALM1	MEF2	RHCG	SGV1	MOK
CMD1	CALM2	MES1	MARS1	SHH4	SDHD
CMD1	CALM3	MGM1	OPA1	SIL1	SIL1
COA6	COA6	MIA40	CHCHD4	SIR2	SIRT1
COQ2	COQ2	MIP1	POLG	SIS1	DNAJB6
COQ4	COQ4	MLH1	MLH1	SKI3	SKIC3
COQ5	COQ5	MLH3	MLH3	SLM3	TRMU
COQ6	COQ6	MNL1	EDEM1	SMP3	PIGZ
COQ8	COQ8A	MOD5	TRIT1	SMT3	SUMO1
COQ8	COQ8B	MPC1	MPC1	SNF2	SMARCA4
COQ9	COQ9	MPE1	RBBP6	SNF4	PRKAG2
COX10	COX10	MPS1	GRK3	SNO4	PARK7
COX12	COX6B1	MRE11	MRE11	SNU13	SNU13
CRS1	SLC25A20	MRM2	MRM2	SOD1	SOD1
CSE1	CSE1L	MRPS12	MRPS12	SOD2	SOD2
CSE4	CENPA	MSB3	USP6NL	SPO14	PLD1
CSE4	CENPA	MSB4	USP6NL	SPT14	PIGA
CTR1	SLC31A1	MSH2	MSH2	SPT15	TBP
CUE5	TOLLIP	MSH6	MSH6	SRB4	MED17
CUP9	PKNOX1	MSS4	PIP5K1A	SRV2	CAP1
CUP9	PKNOX2	MSS4	PIP5K1B	SSL2	ERCC3
CYC1	CYC1	MSW1	WARS2	SSL2	SEC61G
CYC1	CYCS	MTO1	MTO1	STE6	CFTR
CYC3	HCCS	MYG1	MYG1	SUI2	EIF2S1
CYM1	PITRM1	MYO5	MYO5A	SUI3	EIF2S2
CYS4	CBS	MYO5	MYO5B	SYM1	MPV17
DBP2	DDX5	MZM1	LYRM7	TAZ1	TAFAZZIN
DDL1	DDHD1	NAM2	LARS2	TCA17	TRAPPC2L
DDL1	DDHD2	NAN1	COP1	TDP1	TDP1
DFG10	SRD5A3	NAN1	PAFAH1B1	TEF2	EEF1A2
DFR1	DHFR	NAT1	NAA15	TEL1	ATM
DIB1	PIGZ	NCP1	NCP1	THG1	THG1L
DIB1	TXNL4A	NDC80	NDC80	THI7	SLC19A2
DIC1	SLC25A10	NDI1	MT-ND4	THI80	TPK1
DIS3	DIS3	NOB1	NOB1	TRS1	TARS1
DNA2	BLM	NOP14	NOP14	TIF11	EIF1AX
DNA2	DNA2	NOP2	NOP2	TIF4631	EIF4G1
DNA2	WRN	NPC2	NPC2	TIM13	TIMM13
DNM1	DNM1L	NPR2	NPRL2	TIM44	TIMM44
DOG2	PUDP	NPR3	NPRL3	TIM8	TIMM8A
DPM1	DPM1	NUP145	NUP98	TOP2	TOP2A
DUT1	DUT	NUS1	NUS1	TOP2	TOP2B
DYN1	DYNC1H1	OAR1	CBR4	TOR1	MTOR
ECM16	DHX37	ORC1	ORC1	TPI1	TPI1
ECO1	ESCO2	ORC4	ORC4	TRM10	TRMT10A
EFR3	EFR3A	ORC6	ORC6	TRM5	TRMT5
EMG1	EMG1	ORM2	ORMDL1	TRM7	FTSJ1
EMG1	EMG1	ORM2	ORMDL3	TRM732	THADA
ENV9	RDH12	ORT1	SLC25A15	TRM82	WDR4
ERG1	SQLE	PAC10	VBP1	TRS20	TRAPPC2
ERG11	CYP19A1	PAF1	PAF1	TRS20	ZNF547
ERG12	MVK	PAH1	LPIN1	TRS23	TRAPPC4
ERG26	NSDHL	PAH1	LPIN2	TRZ1	ELAC2
ERG27	HSD17B7	PAH1	LPIN3	TSC10	KDSR
ERS1	CFL2	PAM16	PAM16	TSC13	TECR
ERS1	COG4	PCM1	PGM3	TTI1	TTI1
ERS1	CTNS	PDA1	PDHA1	TUB1	TUBA1A
ERS1	PCM1	PEP5	VPS11	TUB2	TUBB1
ERS1	SLC16A1	PET112	GATB	TUB2	TUBB2B
ERS1	SLC16A7	PET9	SLC25A4	TUB2	TUBB3
ERV1	GFER	PEX9	PEX1	TUB3	TUBA1A
ESA1	KAT5	PEX1	PEX1	TUB3	TUBA1B
FAA1	ACSBG1	PEX13	PEX13	TUB3	TUBA1C
FAA1	ACSL1	PEX6	PEX6	TUB4	TUBG1
FAA1	ACSL3	PFK1	PFKM	TUB3	TUBB
FAA1	ACSL4	PFK2	PFKM	TUB4	TUBG1
FAA1	ACSL5	PFY1	PFN1	TYS1	YARS1
FAA1	ACSL6	PGI1	GPI	UBC9	UBE2I
FAA1	SLC27A2	PGK1	PGK1	UBP6	USP14
FAA1	SLC27A4	PGK1	PGK2	UBR1	UBR1
FAA4	ACSBG1	PHO8	ALPG	UFD1	UFD1
FAA4	ACSL1	PHO8	ALPL	UFD4	HGNC:12306
FAA4	ACSL3	PHO85	CDK5	UGO1	SLC25A46
FAA4	PHO85	YTHDF2	UGP1	UGP2
FAA4	ACSL5	PHS1	HACD2	ULP2	PML
FAA4	ACSL6	PIC2	SLC25A4	URA6	CMPK1
FAA4	SLC27A2	PIF1	PIF1	UTP4	UTP4
FAA4	SLC27A4	PMC1	ATP2C1	VAS1	VARS2
FIG4	FIG4	PMR1	ATP2C1	VMA21	VMA21
FLX1	SLC25A32	POB3	SSRP1	VMA22	CCDC115
FMN1	RFK	PNG1	NGLY1	VMS1	ANKZF1
FOL2	GCH1	POB3	SSRP1	VPH1	ATP6V0A1
FOX2	HSD17B4	POL2	POLE	VMS1	ANKZF1
FPR2	FKBP2	POL3	POLD1	VPH1	ATP6V0A4
FUM1	FH	POL31	POLD2	VPH2	TMEM199
FZO1	MFN2	POL32	POLD3	VPS13	VPS13A
GAL10	GALE	PPA2	PPA2	VPS13	VPS13B
GAL7	GALT	PRE5	PSMA1	VPS13	VPS13C
GAL7	UGP2	PRP16	DHX38	VPS13	VPS13D
GCD11	EIF2S3	PRP31	PRPF31	VPS30	BECN1
GCD2	EIF2B4	PRP38	PRPF38B	VPS35	VPS35
GCD6	EIF2B5	PRS1	PRPS1	VPS53	VPS53
GCD7	EIF2B2	PRS5	PRPS1	WHI2	KCTD7
GCN3	EIF2B1	PSD1	PISD	YBL036C	PLPBP
GDI1	GDI2	PSE1	IPO5	YDC1	ACER3
GDT1	TMEM165	PUP3	PSMB3	YDL085C-A	SERF1A
GEF1	CLCN5	PUP3	PSMB3	YFH1	FTL
GEF1	CLCN7	PUS7	PUS7	YHC3	CLN3
GIS2	CNBP	PUT2	ALDH4A1	YHC3	YHC3
GLC3	GBE1	PXA1	ABCD1	YME1L	YME1L1
GLC7	PPP1CA	PXA2	ABCD2	YMR1	MTM1
GLC7	PPP1CC	PXA2	ABCD2	YMR1	MTMR2
GLE1	GLE1	PXR1	PINX1	YNG2	ING3
GLR1	PYROXD1	PXR1	PINX1	YOR1	CFTR
GNA1	GNPNAT1	RAD14	XPA	YPC1	ACER3
GPA1	GNAS	RAD23	ERCC5	YPK9	ATP13A2
GPI10	PIGB	RAD27	FEN1	YPT1	RAB1A
GRS1	GARS1	RAD3	ERCC2	YTA12	AFG3L2
GSH2	GSS	RAD30	POLH	YTA7	ATAD2
GSH2	RRAGC	RAD30	POLH	ZWF1	G6PD