## SGD and SGD MIP1 MIP1 the Alliance of Genome Resources Saccharomyces cerevisiae Species Saccharomyces cerevisiae SGD:S000005857 ♂ of GENOME RESOURCES Stacia R. Engel, Edith D. Wong, Robert S. Nash, Felix Gondwe, Summary MItochondrial DNA Polymerase Kevin A. MacPherson, Patrick Ng, Suzi Aleksander, YOR330C Synonyms Sequence Feature Viewer Biotype protein coding gene Stuart Miyasato, J. Michael Cherry, and The SGD Project Function - GO Annotations Automated Description Exhibits 3'-5' exonuclease activity and DNA-directed DNA polymerase activity. Involved in mitochondrial DNA catabolic process and mitochondrial DNA replication. Localizes to the mitochondrion. Used to study Alpers Department of Genetics, Stanford University, Stanford, CA 94305, USA Orthology syndrome; chronic progressive external ophthalmoplegia; peripheral nervous system disease; and progeria. Human ortholog(s) of this gene implicated in several diseases, including brain disease (multiple); mitochondrial DNA depletion syndrome (multiple); mitochondrial myopathy (multiple); neurodegenerative disease (multiple); Phenotypes The yeast research community has long enjoyed the support provided by the Saccharomyces and ovarian disease (multiple). Orthologous to human POLG (DNA polymerase gamma, catalytic subunit). Disease Associations SGD Description Mitochondrial DNA polymerase gamma; single subunit of mitochondrial DNA polymerase in yeast, in contrast Genome Database (SGD), and has flourished because of its existence, making great 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 breakthroughs and technological advances, and contributing countless key insights to the associated with Alpers-Huttenlocher syndrome (AHS), progressive external ophthalmoplegia (PEO), parkinsonism, other mitochondrial diseases fields of genetics and genomics over the past decades. SGD has recently joined forces with five CBI Gene:854508 2 Genomic Resources other model organism databases (MODs) - WormBase, FlyBase, ZFIN, RGD, and MGI - plus the Molecular Interactions UniProtKB:P15801 2 Additional Information Literature 2 Gene Ontology Consortium (GOC) to form the Alliance of Genome Resources (the Alliance; alliancegenome.org). The Alliance website integrates expertly-curated information on model Sequence Feature Viewer organisms and the functioning of cellular systems, and enables unified access to comparative chrXV:939621...943385 2 (3.76 kb) **Curated summaries** genomics and genetics data, facilitating cross-species analyses. The site is undergoing rapid R64-2-1 development as we work to harmonize various datatypes across the various organisms. about genes, Explore your favorite genes in the Alliance to find information regarding orthology sets, gene their functions, expression, gene function, mutant phenotypes, alleles, disease associations and more! disease associations The Alliance is supported by NIH NHGRI U24HG002223-19S1, NIH NHGRI U41HG001315 (SGD), NIH NHGRI P41HG002659 (ZFIN), NIH **Function - GO Annotations** NHGRI U24HG002223 (WormBase), MRC-UK MR/L001020/1 (WormBase), NIH NHGRI U41HG000739 (FlyBase), NIH NHLBI HL64541 (RGD), NIH NHGRI HG000330 (MGD), and NIH NHGRI U41HG002273 (GOC, which also provides funding to WB, MGD, SGD). Goal: develop and maintain sustainable genome information resources that facilitate the use of diverse model organisms to understand the genetic and genomic bases of human biology, health, and disease Yeast, human, and model organism orthologs • Alleles and phenotype variants • Disease associations • Expression Yeast, fly, worm, Orthology ? rat, mouse, and Phenotype Sequence Protein Gene Ontology Disease Interactions Regulation Expression Literature Summary human homologs MIP1 / YOR330C Overview MIP1/ YOR330C Species MIP1<sup>1</sup> Standard Name: Locus POLG Homo sapiens Systematic Name: YOR330C Overview SGD ID: SGD:S000005857 Mus musculus Polg Sequence Feature Type: ORF, Verified Protein Polg Rattus norvegicus 7 of 9 Description: Mitochondrial DNA polymerase gamma; single subunit of mitochondrial DNA polymerase in yeast, in Gene contrast to metazoan complex of catalytic and accessory subunits; polymorphic in yeast, petites occur Danio rerio polg Ontology more frequently in some lab strains; human ortholog POLG complements yeast mip1 mutant; mutations Drosophila melanogaster in human POLG associated with Alpers-Huttenlocher syndrome (AHS), progressive external Phenotype ophthalmoplegia (PEO), parkinsonism, other mitochondrial diseases 23456789 Disease Caenorhabditis elegans Name Description: MItochondrial DNA Polymerase 1 Interaction Integrated model organism details available at the Alliance of Genome Resources website Phenotypes Comparative Info: Regulation 36 phenotypes based on 36 annotations Expression Sequence • Sequence Details Jump to Alliance gene Literature Phenotype Term T Genetic Entity T Genetic Entity Type 🔻 References T page from SGD History ♣ Download (.fsa) abnormal chemical PMID:27693354 27 View in: JBrowse compound accumulation References abnormal mitochondrial PMID:19751518 2, gene MIP1 Location: Chromosome XV 939621..943385 Resources PMID:16368709 @ genome maintenance PMID:23383298 @ abnormal vacuolar gene morphology gene PMID:12432101 @ abnormal vegetative growth absent mitochondrial PMID:25170845 2, PMID:20883824 27 **Phenotypes** genome maintenance 940000 944000 32000 934000 936000 938000 Disease Associations Gene Ontology • Gene Ontology Details • Compare to ortholog genes Mitochondrial DNA polymerase and 3'-5' exonuclease involved in mitochondrial DNA replication and Summary: Stringency: O Stringent Moderate No filter mitochondrial DNA catabolism during nitrogen starvation POLG (Hsa) X Polg (Mmu) X Polg (Rno) X polg (Dre) X tam (Dme) X polg-1 (Cel) X View computational annotations Disease associations Molecular Function Manually Curated: • 3'-5' exonuclease activity (IDA) across species Phenotype • Phenotype Details • Non-essential gene; null mutants grow slowly, have abnormal vacuolar morphology, accumulate Summary: glycogen, cannot respire or use various nitrogen sources, are sensitive to oxidative stress and MIP1 desiccation, and have shortened lifespan and small cell size; missense mutants that correspond to ♠ POLG human Alpers disease mutations show increased mitochondrial DNA point mutations; heterozygous diploid nulls are haploinsufficient Polg polg **Classical Genetics** 🐞 tam dominant negative: • mitochondrial genome maintenance: abnormal polg-1 Disease associations Disease • Expression Yeast MIP1 is homologous to human POLG, and has been used to study chronic progressive external Summary: **Primary Sources** Serial Patterns of Expression Levels Locator (SPELL) ☑ ophthalmoplegia, Alpers syndrome, progeria, and peripheral nervous system disease SGD ♂ Other Sources GEO ☑ Manually Curated Alpers syndrome (ISS, IGI) chronic progressive external ophthalmoplegia (ISS, IGI) Compare to ortholog genes

Expression Details 2

Expression •

peripheral nervo

progeria (ISS, IGI)

SGD

**Expression data** 

Stringency: Stringent Moderate No filter

**Expression annotations** 

across species

Polg (Mmu) x Polg (Rno) x polg (Dre) x tam (Dme) x