Resource Summary Report

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Human DNA Polymerase Gamma Mutation Database

RRID:SCR_004722
Type: Tool

Proper Citation

Human DNA Polymerase Gamma Mutation Database (RRID:SCR_004722)

Resource Information

URL: http://tools.niehs.nih.gov/polg/

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Description: Database that lists all known mutations in the coding region of the POLG gene and describes the associated disease. Human DNA polymerase is composed of two subunits, a 140 kDa catalytic subunit encoded by the POLG on chromosome 15q25, and a 55kDa accessory subunit encoded by the POLG2 gene on chromosome 17q23-24. A number of mutations have been mapped to the gene for the catalytic subunit of DNA polymerase, POLG, and found to be associated with mitochondrial diseases. The nucleotide changes are numbered from the initiation Methionine codon and are based on the cDNA (accession U60325.1) and gene sequence (accession AF497906.1).

Abbreviations: Human DNA Polymerase Gamma Mutation Database

Resource Type: database, data or information resource

Keywords: mutation, polg, gene, dna polymerase, FASEB list

Related Condition: Mitochondrial disease

Availability: Free

Resource Name: Human DNA Polymerase Gamma Mutation Database

Resource ID: SCR_004722

Alternate IDs: OMICS_01639, nlx_71693

Record Creation Time: 20220129T080226+0000

Record Last Update: 20260725T191135+0000

Ratings and Alerts

No rating or validation information has been found for Human DNA Polymerase Gamma Mutation Database.

No alerts have been found for Human DNA Polymerase Gamma Mutation Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 38 mentions in open access literature.

Listed below are recent publications. The full list is available at SPARC SAWG .

Gilea AI, et al. (2021) Saccharomyces cerevisiae as a Tool for Studying Mutations in Nuclear Genes Involved in Diseases Caused by Mitochondrial DNA Instability. Genes, 12(12).

Gustafson MA, et al. (2020) Consequences of compromised mitochondrial genome integrity. DNA repair, 93, 102916.

Zatopek KM, et al. (2019) RADAR-seq: A RAre DAmage and Repair sequencing method for detecting DNA damage on a genome-wide scale. DNA repair, 80, 36.

Son JM, et al. (2019) Mitochondria: multifaceted regulators of aging. BMB reports, 52(1), 13.

Ross JM, et al. (2019) Voluntary exercise normalizes the proteomic landscape in muscle and brain and improves the phenotype of progeroid mice. Aging cell, 18(6), e13029.

Rohatensky MG, et al. (2018) Assessing the performance of a Loop Mediated Isothermal Amplification (LAMP) assay for the detection and subtyping of high-risk suptypes of Human Papilloma Virus (HPV) for Oropharyngeal Squamous Cell Carcinoma (OPSCC) without DNA purification. BMC cancer, 18(1), 166.

Nikkanen J, et al. (2018) A complex genomic locus drives mtDNA replicase POLG expression to its disease-related nervous system regions. EMBO molecular medicine, 10(1), 13.

Viscomi C, et al. (2017) MtDNA-maintenance defects: syndromes and genes. Journal of inherited metabolic disease, 40(4), 587.

Nurminen A, et al. (2017) Pathogenicity in POLG syndromes: DNA polymerase gamma pathogenicity prediction server and database. BBA clinical, 7, 147.

Béreau M, et al. (2016) The wide POLG-related spectrum: An integrated view. Journal of the neurological sciences, 368, 70.

Rajakulendran S, et al. (2016) A Clinical, Neuropathological and Genetic Study of Homozygous A467T POLG-Related Mitochondrial Disease. PloS one, 11(1), e0145500.

Rahn JJ, et al. (2015) Zebrafish lacking functional DNA polymerase gamma survive to juvenile stage, despite rapid and sustained mitochondrial DNA depletion, altered energetics and growth. Nucleic acids research, 43(21), 10338.

Lasserre JP, et al. (2015) Yeast as a system for modeling mitochondrial disease mechanisms and discovering therapies. Disease models & mechanisms, 8(6), 509.

Falk MJ, et al. (2015) Mitochondrial Disease Sequence Data Resource (MSeqDR): a global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Molecular genetics and metabolism, 114(3), 388.

Lodi T, et al. (2015) DNA polymerase ? and disease: what we have learned from yeast. Frontiers in genetics, 6, 106.

Kaliszewska M, et al. (2015) Yeast model analysis of novel polymerase gamma variants found in patients with autosomal recessive mitochondrial disease. Human genetics, 134(9), 951.

Qian Y, et al. (2015) Alpers disease mutations in human DNA polymerase gamma cause catalytic defects in mitochondrial DNA replication by distinct mechanisms. Frontiers in genetics, 6, 135.

Szczepanowska K, et al. (2015) Different faces of mitochondrial DNA mutators. Biochimica et biophysica acta, 1847(11), 1362.

Baruffini E, et al. (2015) Polymorphisms in DNA polymerase ? affect the mtDNA stability and the NRTI-induced mitochondrial toxicity in Saccharomyces cerevisiae. Mitochondrion, 20, 52.

Copeland WC, et al. (2014) Mitochondrial genome maintenance in health and disease. DNA repair, 19, 190.