

Resource Summary Report

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mtDB - Human Mitochondrial Genome Database

RRID:SCR_002945

Type: Tool

Proper Citation

mtDB - Human Mitochondrial Genome Database (RRID:SCR_002945)

Resource Information

URL: <http://www.genpat.uu.se/mtDB>

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Description: A database of human mitochondrial genomes containing mtDNA sequences, polymorphic sites, and the ability to search for specific variants. It contains 1865 complete sequences and 839 coding region sequences.

Abbreviations: mtDB

Synonyms: Human Mitochondrial Genome Database

Resource Type: data or information resource, database

Defining Citation: [PMID:16381973](#)

Keywords: human genome, mitochondrial dna, sequence, variant, population genetics, coding region, polymorphic site, population, mitochondrial sequence, mitochondrial polymorphism, FASEB list

Funding: Swedish Research Council

Availability: THIS RESOURCE IS NO LONGER IN SERVICE

Resource Name: mtDB - Human Mitochondrial Genome Database

Resource ID: SCR_002945

Alternate IDs: nif-0000-02994, OMICS_01642

Record Creation Time: 20220129T080216+0000

Record Last Update: 20260904T000106+0000

Ratings and Alerts

No rating or validation information has been found for mtDB - Human Mitochondrial Genome Database.

No alerts have been found for mtDB - Human Mitochondrial Genome Database.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 59 mentions in open access literature.

Listed below are recent publications. The full list is available at [kravitz2](#) .

Ding Y, et al. (2025) Dilated Cardiomyopathy May Be Associated With a Novel Mitochondrial tRNASer(AGY) Mutation. Human mutation, 2025, 7888334.

Yu X, et al. (2024) The Association Between Mitochondrial tRNAGlu Variants and Hearing Loss: A Case-Control Study. Pharmacogenomics and personalized medicine, 17, 77.

Yang L, et al. (2022) Late onset of type 2 diabetes is associated with mitochondrial tRNATrp A5514G and tRNASer(AGY) C12237T mutations. Journal of clinical laboratory analysis, 36(1), e24102.

Lin YH, et al. (2021) Impact of an MT-RNR1 Gene Polymorphism on Hepatocellular Carcinoma Progression and Clinical Characteristics. International journal of molecular sciences, 22(3).

Lin L, et al. (2021) Mutational Analysis of Mitochondrial tRNA Genes in 200 Patients with Type 2 Diabetes Mellitus. International journal of general medicine, 14, 5719.

Dawod PGA, et al. (2020) Whole Mitochondrial Genome Analysis in Serbian Cases of Leber's Hereditary Optic Neuropathy. Genes, 11(9).

Ding Y, et al. (2020) Screening for deafness-associated mitochondrial 12S rRNA mutations by using a multiplex allele-specific PCR method. Bioscience reports, 40(5).

Schlapakow E, et al. (2019) Distinct segregation of the pathogenic m.5667G>A mitochondrial tRNAAsn mutation in extraocular and skeletal muscle in chronic progressive external ophthalmoplegia. Neuromuscular disorders : NMD, 29(5), 358.

Poma A, et al. (2019) Analysis of ancient mtDNA from the medieval archeological site of Amiternum (L'Aquila), central Italy. Heliyon, 5(10), e02586.

Zhang Z, et al. (2019) Maternally inherited coronary heart disease is associated with a novel mitochondrial tRNA mutation. BMC cardiovascular disorders, 19(1), 293.

Zhu Y, et al. (2018) Mitochondrial DNA 7908-8816 region mutations in maternally inherited essential hypertensive subjects in China. BMC medical genomics, 11(1), 89.

Zhu Y, et al. (2016) A Mitochondrial DNA A8701G Mutation Associated with Maternally Inherited Hypertension and Dilated Cardiomyopathy in a Chinese Pedigree of a Consanguineous Marriage. Chinese medical journal, 129(3), 259.

Pallavi T, et al. (2016) Identical mitochondrial somatic mutations unique to chronic periodontitis and coronary artery disease. Journal of Indian Society of Periodontology, 20(1), 17.

Zhen X, et al. (2015) Increased Incidence of Mitochondrial Cytochrome C Oxidase 1 Gene Mutations in Patients with Primary Ovarian Insufficiency. PloS one, 10(7), e0132610.

McCann BJ, et al. (2015) A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscular disorders : NMD, 25(3), 262.

Wang Z, et al. (2015) Mitochondrial Variations in Non-Small Cell Lung Cancer (NSCLC) Survival. Cancer informatics, 14(Suppl 1), 1.

Fang H, et al. (2015) Exercise intolerance and developmental delay associated with a novel mitochondrial ND5 mutation. Scientific reports, 5, 10480.

Burrage LC, et al. (2014) Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia (MLASA) plus associated with a novel de novo mutation (m.8969G>A) in the mitochondrial encoded ATP6 gene. Molecular genetics and metabolism, 113(3), 207.

Liu Z, et al. (2014) The novel mitochondrial 16S rRNA 2336T>C mutation is associated with hypertrophic cardiomyopathy. Journal of medical genetics, 51(3), 176.

Liu Y, et al. (2014) Systematic analysis of the clinical and biochemical characteristics of maternally inherited hypertension in Chinese Han families associated with mitochondrial. BMC medical genomics, 7, 73.