

Resource Summary Report

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MITOMAP - A human mitochondrial genome database

RRID:SCR_002996

Type: Tool

Proper Citation

MITOMAP - A human mitochondrial genome database (RRID:SCR_002996)

Resource Information

URL: <http://www.mitomap.org/>

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Description: Database of polymorphisms and mutations of the human mitochondrial DNA. It reports published and unpublished data on human mitochondrial DNA variation. All data is curated by hand. If you would like to submit published articles to be included in mitomap, please send them the citation and a pdf.

Abbreviations: MITOMAP

Resource Type: data or information resource, database

Defining Citation: [PMID:17178747](#), [PMID:15608272](#), [PMID:9399813](#), [PMID:9016535](#), [PMID:8594574](#)

Keywords: gene, genome, diabetes, disease, disease-association, high resolution screening, human, inversion, metabolism, mitochondrial dna, mutation, phenotype, polymorphism, polypeptide assignment, pseudogene, restriction site, rna, sequence, trna, unpublished, variation, mitochondria, dna, insertion, deletion, FASEB list

Funding: NIH ;
Muscular Dystrophy Foundation ;
Ellison Foundation ;
Diputacion General de Aragon Grupos consolidados B33 ;
NIGMS GM46915;
NINDS NS21328;
NHLBI HL30164;
NIA AG10130;
NIA AG13154;

NINDS NS213L8;
NHLBI HL64017;
NIH Biomedical Informatics Training Grant T15 LM007443;
NSF EIA-0321390;
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Ciber Enfermedades raras CB06/07/0043

Availability: Except where otherwise noted, Creative Commons Attribution License, The community can contribute to this resource

Resource Name: MITOMAP - A human mitochondrial genome database

Resource ID: SCR_002996

Alternate IDs: nif-0000-00511, OMICS_01641

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Record Creation Time: 20220129T080216+0000

Record Last Update: 20260729T044035+0000

Ratings and Alerts

No rating or validation information has been found for MITOMAP - A human mitochondrial genome database.

No alerts have been found for MITOMAP - A human mitochondrial genome database.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 368 mentions in open access literature.

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

Mareckova K, et al. (2025) Mitochondrial DNA variants and their impact on epigenetic and biological aging in young adulthood. Translational psychiatry, 15(1), 16.

Imasawa T, et al. (2025) Comprehensive review of mitochondrial nephropathy-a renal phenotype in mitochondrial disease: causative genes, clinical and pathological features, diagnosis, prognosis, and treatment. Clinical and experimental nephrology, 29(1), 39.

Cabrera-Alarcon JL, et al. (2025) Shaping current European mitochondrial haplogroup frequency in response to infection: the case of SARS-CoV-2 severity. Communications biology, 8(1), 33.

- Amor H, et al. (2025) Effects of marijuana and tobacco on male fertility and their relationship to genetic variation of mitochondrial cytochrome C oxidase genes. *Scientific reports*, 15(1), 7547.
- Ratnaike T, et al. (2025) Mitochondrial DNA disease discovery through evaluation of genotype and phenotype data: The Solve-RD experience. *American journal of human genetics*, 112(6), 1376.
- Rosdi SNM, et al. (2025) Chiari malformation type 1 with combined nuclear PAX1 and DKK1 genes and mitochondrial D-loop variants: a case report. *Croatian medical journal*, 66(1), 56.
- Liu Z, et al. (2025) Novel pathogenic mtDNA variants in Chinese children with neurological mitochondrial disorders. *Annals of clinical and translational neurology*, 12(3), 586.
- Tang J, et al. (2025) Mitochondrial base editing: from principle, optimization to application. *Cell & bioscience*, 15(1), 9.
- Rao X, et al. (2025) Mitochondrial tRNAGlu 14687A>G May Be A Novel Mutation for Type 2 Diabetes Mellitus. *Journal of clinical laboratory analysis*, 39(13), e70056.
- Lyu L, et al. (2025) Advances in gene therapy for mitochondrial genetic disorders: current status and clinical implementation challenges. *Journal of translational medicine*, 23(1), 1415.
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- Luo Z, et al. (2025) Molecular Characterization of Two Hypertension Pedigrees Carrying Mitochondrial tRNAGln 4386T>C Mutation. *Human heredity*, 90(1), 1.
- Liu H, et al. (2025) CUX1 variant and 9p deletion: expanding the spectrum and resolving variable GDD/ID in a family. *BMC medical genomics*, 18(1), 134.
- Walker VE, et al. (2025) Mutational analysis of the mitochondrial tRNA genes and flanking regions in lymphocytes from long-term pediatric cancer survivors given doxorubicin chemotherapy for acute lymphoblastic leukemia. *Cardio-oncology (London, England)*, 11(1), 99.
- Boso D, et al. (2024) Pathogenic mitochondrial DNA variants are associated with response to anti-VEGF therapy in ovarian cancer PDX models. *Journal of experimental & clinical cancer research : CR*, 43(1), 325.

Lv L, et al. (2024) A Comprehensive Prognostic Model for Colon Adenocarcinoma Depending on Nuclear-Mitochondrial-Related Genes. *Technology in cancer research & treatment*, 23, 15330338241258570.

Yang HK, et al. (2024) Mitochondrial DNA mutations in Korean patients with Leber's hereditary optic neuropathy. *Scientific reports*, 14(1), 5702.

Lu JL, et al. (2024) Taurine hypomodification underlies mitochondrial tRNA^{Trp}-related genetic diseases. *Nucleic acids research*, 52(21), 13351.