

Resource Summary Report

Generated by [Kravitz](#) on Sep 10, 2026

HmtVar

RRID:SCR_017288

Type: Tool

Proper Citation

HmtVar (RRID:SCR_017288)

Resource Information

URL: <https://www.hmtvar.uniba.it>

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Description: Manually curated database offering variability and pathogenicity information about mtDNA variants. Human mitochondrial variants data of healthy and diseased subjects. Data and text mining pipeline to annotate human mitochondrial variants with functional and clinical information.

Resource Type: data or information resource, database, service resource

Defining Citation: [PMID:30371888](#), [PMID:31821723](#)

Keywords: manually, curated, data, variability, mitochondria, pathogenicity, mtDNA, variant, human, bio.tools

Funding: DHOMOS Worldwide Cancer Research ;
DISCO TRIP ;
Italian Ministry of Health ;
Rosa Maria Massari fellowship from the Italian Association for Cancer Research

Availability: Free, Freely available

Resource Name: HmtVar

Resource ID: SCR_017288

Alternate IDs: biotools:HmtVar

Alternate URLs: <https://bio.tools/HmtVar>

Record Creation Time: 20220129T080334+0000

Record Last Update: 20260905T133214+0000

Ratings and Alerts

No rating or validation information has been found for HmtVar.

No alerts have been found for HmtVar.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 12 mentions in open access literature.

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

Schneider K, et al. (2022) Differential modulation of cancer-related genes by mitochondrial DNA haplogroups and the STING DNA sensing system. *FASEB bioAdvances*, 4(10), 675.

Li M, et al. (2022) Contribution of mitochondrial gene variants in diabetes and diabetic kidney disease. *Frontiers in endocrinology*, 13, 953631.

Miglietta S, et al. (2022) MicroRNA and Metabolic Profiling of a Primary Ovarian Neuroendocrine Carcinoma Pulmonary-Type Reveals a High Degree of Similarity with Small Cell Lung Cancer. *Non-coding RNA*, 8(5).

Saha SK, et al. (2021) Evaluation of D-loop hypervariable region I variations, haplogroups and copy number of mitochondrial DNA in Bangladeshi population with type 2 diabetes. *Heliyon*, 7(7), e07573.

Girolimetti G, et al. (2021) Mitochondrial DNA analysis efficiently contributes to the identification of metastatic contralateral breast cancers. *Journal of cancer research and clinical oncology*, 147(2), 507.

Atilano SR, et al. (2021) Low frequency mitochondrial DNA heteroplasmy SNPs in blood, retina, and [RPE+choroid] of age-related macular degeneration subjects. *PloS one*, 16(1), e0246114.

Labory J, et al. (2020) Multi-Omics Approaches to Improve Mitochondrial Disease Diagnosis: Challenges, Advances, and Perspectives. *Frontiers in molecular biosciences*, 7, 590842.

Vitale O, et al. (2020) A data and text mining pipeline to annotate human mitochondrial variants with functional and clinical information. *Molecular genetics & genomic medicine*, 8(2), e1085.

Yuan H, et al. (2020) Profiling of mitochondrial genomes in SCA3/MJD patients from mainland China. *Gene*, 738, 144487.

Abedi S, et al. (2020) Differential effects of cisplatin on cybrid cells with varying mitochondrial DNA haplogroups. PeerJ, 8, e9908.

Patel TH, et al. (2019) European mtDNA Variants Are Associated With Differential Responses to Cisplatin, an Anticancer Drug: Implications for Drug Resistance and Side Effects. Frontiers in oncology, 9, 640.

Bris C, et al. (2018) Bioinformatics Tools and Databases to Assess the Pathogenicity of Mitochondrial DNA Variants in the Field of Next Generation Sequencing. Frontiers in genetics, 9, 632.