

# Resource Summary Report

Generated by [RRID](#) on Apr 9, 2025

## MToolBox

RRID:SCR\_012112

Type: Tool

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### Proper Citation

MToolBox (RRID:SCR\_012112)

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### Resource Information

**URL:** <http://sourceforge.net/projects/mtoolbox/>

**Proper Citation:** MToolBox (RRID:SCR\_012112)

**Description:** Software for a highly automated bioinformatics pipeline to reconstruct and analyze human mitochondrial DNA from high throughput sequencing data.

**Resource Type:** software resource

**Defining Citation:** [PMID:25028726](#)

**Keywords:** standalone software, bio.tools

**Funding:**

**Availability:** GNU General Public License

**Resource Name:** MToolBox

**Resource ID:** SCR\_012112

**Alternate IDs:** OMICS\_05466, biotools:mtoolbox

**Alternate URLs:** <https://bio.tools/mtoolbox>

**Record Creation Time:** 20220129T080308+0000

**Record Last Update:** 20250214T183206+0000

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### Ratings and Alerts

No rating or validation information has been found for MToolBox.

No alerts have been found for MToolBox.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 48 mentions in open access literature.

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

Wang Z, et al. (2024) VarCards2: an integrated genetic and clinical database for ACMG-AMP variant-interpretation guidelines in the human whole genome. *Nucleic acids research*, 52(D1), D1478.

Donato L, et al. (2024) The genomic mosaic of mitochondrial dysfunction: Decoding nuclear and mitochondrial epigenetic contributions to maternally inherited diabetes and deafness pathogenesis. *Heliyon*, 10(14), e34756.

Slapnik B, et al. (2024) The quality and detection limits of mitochondrial heteroplasmy by long read nanopore sequencing. *Scientific reports*, 14(1), 26778.

Pettenuzzo I, et al. (2024) COQ7 defect causes prenatal onset of mitochondrial CoQ10 deficiency with cardiomyopathy and gastrointestinal obstruction. *European journal of human genetics : EJHG*, 32(8), 938.

Baltar F, et al. (2024) Two compound heterozygous variants in the CLN8 gene are responsible for neuronal cereidolipofuscinoses disorder in a child: a case report. *Frontiers in pediatrics*, 12, 1379254.

Sun X, et al. (2024) Association analysis of mitochondrial DNA heteroplasmic variants: methods and application. *medRxiv : the preprint server for health sciences*.

Stenton SL, et al. (2024) Mitochondrial DNA variant detection in over 6,500 rare disease families by the systematic analysis of exome and genome sequencing data resolves undiagnosed cases. *medRxiv : the preprint server for health sciences*.

Raggio V, et al. (2023) Computational and mitochondrial functional studies of novel compound heterozygous variants in SPATA5 gene support a causal link with epileptogenic encephalopathy. *Human genomics*, 17(1), 14.

Mahar NS, et al. (2023) A systematic comparison of human mitochondrial genome assembly tools. *BMC bioinformatics*, 24(1), 341.

Vats S, et al. (2023) Characterization of the Mitochondrial Genetic Landscape in Abdominal Aortic Aneurysm. *Journal of the American Heart Association*, 12(8), e029248.

Li Y, et al. (2023) Mitochondrial heteroplasmic shifts reveal a positive selection of breast cancer. *Journal of translational medicine*, 21(1), 696.

Tsybrovskyy O, et al. (2022) Papillary thyroid carcinoma tall cell variant shares accumulation of mitochondria, mitochondrial DNA mutations, and loss of oxidative phosphorylation complex I integrity with oncocytic tumors. *The journal of pathology. Clinical research*, 8(2), 155.

Nie Y, et al. (2022) Heteroplasmic mitochondrial DNA mutations in frontotemporal lobar degeneration. *Acta neuropathologica*, 143(6), 687.

Calabrese C, et al. (2022) Heteroplasmic mitochondrial DNA variants in cardiovascular diseases. *PLoS genetics*, 18(4), e1010068.

Hiz Kurul S, et al. (2022) High diagnostic rate of trio exome sequencing in consanguineous families with neurogenetic diseases. *Brain : a journal of neurology*, 145(4), 1507.

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).

Ye Z, et al. (2022) Genetic Diversity, Heteroplasmy, and Recombination in Mitochondrial Genomes of *Daphnia pulex*, *Daphnia pulicaria*, and *Daphnia obtusa*. *Molecular biology and evolution*, 39(4).

Wei W, et al. (2022) Nuclear-embedded mitochondrial DNA sequences in 66,083 human genomes. *Nature*, 611(7934), 105.

Wei W, et al. (2021) Cell reprogramming shapes the mitochondrial DNA landscape. *Nature communications*, 12(1), 5241.

Bicci I, et al. (2021) Single-molecule mitochondrial DNA sequencing shows no evidence of CpG methylation in human cells and tissues. *Nucleic acids research*, 49(22), 12757.