Resource Summary Report

Generated by ASWG on Aug 3, 2026

MToolBox

RRID:SCR_012112
Type: Tool

Proper Citation

MToolBox (RRID:SCR_012112)

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

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: 20260801T190433+0000

Ratings and Alerts

No rating or validation information has been found for MToolBox.

No alerts have been found for MToolBox.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 56 mentions in open access literature.

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

Gao F, et al. (2025) Mitochondrial DNA disorders in neuromuscular diseases in diverse populations. Annals of clinical and translational neurology, 12(8), 1680.

Stenton SL, et al. (2025) Mitochondrial DNA variant detection in over 6,500 rare disease families by the systematic analysis of exome and genome sequencing data resolves undiagnosed cases. HGG advances, 6(3), 100441.

Kremer LS, et al. (2025) The bottleneck for maternal transmission of mtDNA is linked to purifying selection by autophagy. Science advances, 11(46), eaea4660.

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.

Yamaguchi TN, et al. (2025) The Germline and Somatic Origins of Prostate Cancer Heterogeneity. Cancer discovery, 15(5), 988.

Laurie S, et al. (2025) Genomic reanalysis of a pan-European rare-disease resource yields new diagnoses. Nature medicine, 31(2), 478.

Nie Y, et al. (2025) Origin and cell type specificity of mitochondrial DNA mutations in C9ORF72 ALS-FTLD human brain organoids. Science advances, 11(10), eadr0690.

Lai M, et al. (2025) Epigenome-wide association study of nuclear DNA methylation in relation to mitochondrial heteroplasmy. Nature communications, 16(1), 10962.

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.

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

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.

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.