

# Resource Summary Report

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## MAJIQ

RRID:SCR\_016706

Type: Tool

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### Proper Citation

MAJIQ (RRID:SCR\_016706)

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### Resource Information

**URL:** <https://majiq.biociphers.org/>

**Proper Citation:** MAJIQ (RRID:SCR\_016706)

**Description:** Software package to detect and quantify local splicing variations (LSV) from RNA-Seq data. Used to automatically detect and downweight outliers in RNA-Seq datasets with replicates for differential splicing (SD) analysis.

**Abbreviations:** MAJIQ,

**Synonyms:** MAJIQ, MAJIQ/Viola, Modeling Alternative Junction Inclusion Quantification

**Resource Type:** data analysis software, data processing software, software application, software resource, software toolkit

**Defining Citation:** [PMID:29236961](#)

**Keywords:** detect, quantify, local, splicing, variation, RNA-Seq, data, replicate, differential, analysis

**Funding:** NIA R01 AG046544;  
Penn Institute for Biomedical Informatics Pilot

**Availability:** Free, Available for download, Freely available, Tutorial available

**Resource Name:** MAJIQ

**Resource ID:** SCR\_016706

**Record Creation Time:** 20220129T080332+0000

**Record Last Update:** 20260919T195327+0000

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### Ratings and Alerts

No rating or validation information has been found for MAJIQ.

No alerts have been found for MAJIQ.

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## Data and Source Information

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

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## Usage and Citation Metrics

We found 33 mentions in open access literature.

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

Aicher JK, et al. (2025) MAJIQ-CLIN: A novel tool for the identification of Mendelian disease-causing variants from RNA-Seq data. medRxiv : the preprint server for health sciences.

Weinreb A, et al. (2025) Alternative splicing across the C. elegans nervous system. Nature communications, 16(1), 4508.

Todorow V, et al. (2025) Comparative Analysis of Splicing Alterations in Three Muscular Dystrophies. Biomedicines, 13(3).

Alessandrini F, et al. (2025) TDP-43 dysfunction compromises UPF1-dependent mRNA metabolism in ALS. Neuron.

Hung C, et al. (2025) Mislocalization of nucleic acids is a convergent and targetable mechanism in Alzheimer's disease and frontotemporal dementia. Cell reports, 44(7), 115867.

Quesnel-Vallières M, et al. (2024) Trametinib Sensitivity is Defined by a Myeloid Differentiation Profile in Acute Myeloid Leukemia. Drugs in R&D, 24(3), 489.

Fonseca R, et al. (2024) Insights into the functional role of tomato TM6 as a transcriptional regulator of flower development. Horticulture research, 11(3), uhae019.

Torres-Diz M, et al. (2024) An Alternatively Spliced Gain-of-Function NT5C2 Isoform Contributes to Chemoresistance in Acute Lymphoblastic Leukemia. Cancer research, 84(20), 3327.

Schätzl T, et al. (2024) Meta-analysis towards FSHD reveals misregulation of neuromuscular junction, nuclear envelope, and spliceosome. Communications biology, 7(1), 640.

Ziff OJ, et al. (2023) Integrated transcriptome landscape of ALS identifies genome instability linked to TDP-43 pathology. Nature communications, 14(1), 2176.

Vaquero-Garcia J, et al. (2023) RNA splicing analysis using heterogeneous and large RNA-seq datasets. Nature communications, 14(1), 1230.

Martinelli M, et al. (2023) The poly(C)-binding protein Pcbp2 is essential for CD4+ T cell activation and proliferation. *iScience*, 26(1), 105860.

Han SW, et al. (2023) Contrasting and Combining Transcriptome Complexity Captured by Short and Long RNA Sequencing Reads. *bioRxiv* : the preprint server for biology.

Ziff OJ, et al. (2023) Nucleocytoplasmic mRNA redistribution accompanies RNA binding protein mislocalization in ALS motor neurons and is restored by VCP ATPase inhibition. *Neuron*, 111(19), 3011.

Postel MD, et al. (2022) Transcriptome analysis provides critical answers to the "variants of uncertain significance" conundrum. *Human mutation*, 43(11), 1590.

Deschamps-Francoeur G, et al. (2022) The snoGloBe interaction predictor reveals a broad spectrum of C/D snoRNA RNA targets. *Nucleic acids research*, 50(11), 6067.

Karakulak T, et al. (2021) Probing Isoform Switching Events in Various Cancer Types: Lessons From Pan-Cancer Studies. *Frontiers in molecular biosciences*, 8, 726902.

Xu SJ, et al. (2021) Chromatin-mediated alternative splicing regulates cocaine-reward behavior. *Neuron*, 109(18), 2943.

Dent CI, et al. (2021) Quantifying splice-site usage: a simple yet powerful approach to analyze splicing. *NAR genomics and bioinformatics*, 3(2), lqab041.

Lester E, et al. (2021) Tau aggregates are RNA-protein assemblies that mislocalize multiple nuclear speckle components. *Neuron*, 109(10), 1675.