Resource Summary Report

Generated by T1D on Sep 5, 2026

LeafCutter

RRID:SCR_017639
Type: Tool

Proper Citation

LeafCutter (RRID:SCR_017639)

Resource Information

URL: https://github.com/davidaknowles/leafcutter/

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Description: Software tool for identifying and quantifying RNA splicing variation. Used to study sample and population variation in intron splicing. Identifies variable intron splicing events from short read RNA-seq data and finds alternative splicing events of high complexity. Used for detecting differential splicing between sample groups, and for mapping splicing quantitative trait loci (sQTLs).

Resource Type: data analysis software, data analytics software, data processing software, software application, software resource

Defining Citation: PMID:29229983, DOI:10.1038/s41588-017-0004-9

Keywords: Identify, quantitate, RNA, splicing, variation, intron, short, read, RNAseq, data, mapping, trait, loci, sQTL

Funding: CEHG Fellowship ;
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NHGRI HG007036;
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NHGRI HG009431;
NIMH R01 MH107666

Availability: Free, Available for download, Freely available

Resource Name: LeafCutter

Resource ID: SCR_017639

License: Apache License 2.0

Record Creation Time: 20220129T080336+0000

Record Last Update: 20260903T235824+0000

Ratings and Alerts

No rating or validation information has been found for LeafCutter.

No alerts have been found for LeafCutter.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 32 mentions in open access literature.

Listed below are recent publications. The full list is available at T1D .

Takahashi K, et al. (2026) Diagnostic potential of cryptic exon-derived peptides in serum extracellular vesicles for sporadic amyotrophic lateral sclerosis. Inflammation and regeneration, 46(1).

Brownstein CA, et al. (2026) Multimodal Sequencing and Reanalysis Approaches to End the Diagnostic Odyssey of Individuals with Suspected Rare Monogenic Diseases. Genes, 17(6).

Luo M, et al. (2025) The interplay between epigenomic and transcriptomic variation during ecotype divergence in stickleback. BMC biology, 23(1), 70.

Wang H, et al. (2025) Adipocyte-specific Steap4 deficiency reduced thermogenesis and energy expenditure in mice. iScience, 28(2), 111903.

Scotton WJ, et al. (2025) Identification of common variants influencing risk of the three-repeat tauopathy Pick's disease: a genome wide association study. medRxiv : the preprint server for health sciences.

Segarra-Casas A, et al. (2025) Translating Muscle RNAseq Into the Clinic for the Diagnosis of Muscle Diseases. Annals of clinical and translational neurology, 12(7), 1465.

Lall D, et al. (2025) An organ-chip model of sporadic ALS using iPSC-derived spinal cord motor neurons and an integrated blood-brain-like barrier. Cell stem cell, 32(7), 1139.

Hodonsky CJ, et al. (2024) Multi-ancestry genetic analysis of gene regulation in coronary arteries prioritizes disease risk loci. Cell genomics, 4(1), 100465.

Mapel XM, et al. (2024) Molecular quantitative trait loci in reproductive tissues impact male fertility in cattle. Nature communications, 15(1), 674.

Solovyeva EM, et al. (2024) Integrative Proteogenomics for Differential Expression and Splicing Variation in a DM1 Mouse Model. Molecular & cellular proteomics : MCP, 23(1),

100683.

Hazell Pickering S, et al. (2024) Alternative isoform expression of key thermogenic genes in human beige adipocytes. Frontiers in endocrinology, 15, 1395750.

Ueda MT, et al. (2024) Functional and dynamic profiling of transcript isoforms reveals essential roles of alternative splicing in interferon response. Cell genomics, 4(10), 100654.

Mehta P, et al. (2024) Reduced protein-coding transcript diversity in severe dengue emphasises the role of alternative splicing. Life science alliance, 7(8).

Saevarsdottir S, et al. (2024) Start codon variant in LAG3 is associated with decreased LAG-3 expression and increased risk of autoimmune thyroid disease. Nature communications, 15(1), 5748.

Kerimov N, et al. (2023) Systematic visualisation of molecular QTLs reveals variant mechanisms at GWAS loci. bioRxiv : the preprint server for biology.

Kerimov N, et al. (2023) eQTL Catalogue 2023: New datasets, X chromosome QTLs, and improved detection and visualisation of transcript-level QTLs. PLoS genetics, 19(9), e1010932.

Real R, et al. (2023) Association between the LRP1B and APOE loci and the development of Parkinson's disease dementia. Brain : a journal of neurology, 146(5), 1873.

Bendik J, et al. (2023) OutSplice: A Novel Tool for the Identification of Tumor-Specific Alternative Splicing Events. BioMedInformatics, 3(4), 853.

Workman MJ, et al. (2023) Large-scale differentiation of iPSC-derived motor neurons from ALS and control subjects. Neuron, 111(8), 1191.

Kumar R, et al. (2022) Oligonucleotide correction of an intronic TIMMDC1 variant in cells of patients with severe neurodegenerative disorder. NPJ genomic medicine, 7(1), 9.