Resource Summary Report

Generated by dkNET on Sep 3, 2026

Splice AI

RRID:SCR_024532
Type: Tool

Proper Citation

Splice AI (RRID:SCR_024532)

Resource Information

URL: https://pypi.org/project/spliceai/

Proper Citation: Splice AI (RRID:SCR_024532)

Description: Software package annotates genetic variants with their predicted effect on splicing. Used to identify splice variants.

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

Keywords: annotate genetic variants, predicted effect on splicing, identify splice variants,

Availability: Free, Available for download, Freely available

Resource Name: Splice AI

Resource ID: SCR_024532

Alternate URLs: https://github.com/Illumina/SpliceAI

License: GNU GPL v3

Record Creation Time: 20231003T050246+0000

Record Last Update: 20260829T182835+0000

Ratings and Alerts

No rating or validation information has been found for Splice AI.

No alerts have been found for Splice AI.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 44 mentions in open access literature.

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

Yang Y, et al. (2026) Loss-of-function variants in MARK2 cause neurodevelopmental disorder. HGG advances, 7(3), 100600.

Boonen RACM, et al. (2026) Site-saturation functional screens identify PALB2 missense variants associated with increased breast cancer risk. Nature communications, 17(1), 775.

Pantea CL, et al. (2026) Whole Genome Sequencing as First Diagnostic Approach for Inborn Errors of Immunity in Adults: Diagnostic Yield and Clinical Correlations. International journal of molecular sciences, 27(8).

Li K, et al. (2026) Mapping the Non-Canonical Splicing Variants: Decrypting the Hidden Genetic Architecture of Idiopathic Male Infertility. Advanced science (Weinheim, Baden-Wurttemberg, Germany), 13(9), e15512.

Shi T, et al. (2026) Functional reassessment of extended splice region variants in MYO7A with hearing loss and Usher syndrome. The Journal of pathology, 269(2), 222.

Lyu Y, et al. (2026) Identification and functional analysis of a novel TRAPPC2 intronic variant in a four-generation Chinese pedigree with SEDT. Frontiers in genetics, 17, 1763609.

Shohdy KS, et al. (2025) Genetic testing for BRCA1/2 variants in Northern African women with ovarian and breast cancers: a multicentre study of an under-represented ancestry. ESMO open, 10(8), 105510.

Wang L, et al. (2025) Whole-exome sequencing study of opioid dependence offers novel insights into the contributions of exome variants. Translational psychiatry, 15(1), 380.

Alioua N, et al. (2025) Intronic branchpoint-to-acceptor variants underlying inborn errors of immunity. Journal of human immunity, 1(3), e20250041.

Chao KH, et al. (2025) OpenSpliceAI provides an efficient modular implementation of SpliceAI enabling easy retraining across nonhuman species. eLife, 14.

Dominguez-Alonso S, et al. (2025) Alternative splicing analysis in a Spanish ASD (Autism Spectrum Disorders) cohort: in silico prediction and characterization. Scientific reports, 15(1), 10730.

Kamizela AE, et al. (2025) Timing and trajectory of BCR::ABL1-driven chronic myeloid leukaemia. Nature, 640(8060), 982.

Fiorica PN, et al. (2025) Germline Pathogenic DROSHA Variants Are Linked to Pineoblastoma and Wilms Tumor Predisposition. Clinical cancer research : an official

journal of the American Association for Cancer Research, 31(8), 1491.

Shimizu N, et al. (2025) Novel FBN1 intron variant causes isolated ectopia lentis via in-frame exon skipping. Journal of human genetics, 70(4), 199.

Nejati P, et al. (2025) A case study of a novel homozygous EDAR splice site variant in hypohidrotic ectodermal dysplasia with tooth agenesis: molecular dynamics insights. BMC medical genomics, 19(1), 19.

Canson DM, et al. (2025) The SeqSplice multiplexed minigene splicing assay for characterization and quantitation of variant-induced BRCA1 and BRCA2 splice isoforms. Genome research, 35(9), 2104.

Li K, et al. (2024) Prioritizing de novo potential non-canonical splicing variants in neurodevelopmental disorders. EBioMedicine, 99, 104928.

Han SH, et al. (2024) Integrative Analysis of Germline Rare Variants in Clear and Non-clear Cell Renal Cell Carcinoma. European urology open science, 62, 107.

Lin Q, et al. (2024) A genetic investigation in five Chinese families with keratoconus. PeerJ, 12, e18037.

Liao X, et al. (2024) A Case of a Patient With MYH2-Associated Myopathy Presenting With a Chief Complaint of Hand Tremor. Tremor and other hyperkinetic movements (New York, N.Y.), 14, 50.