Resource Summary Report

Generated by RRID on Apr 10, 2025

SpliceDisease

RRID:SCR_006130
Type: Tool

Proper Citation

SpliceDisease (RRID:SCR_006130)

Resource Information

URL: http://202.38.126.151:8080/SDisease/

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Description: Curated database of experimentally supported data of RNA Splicing mutation and disease. The RNA Splicing mutations include cis-acting mutations that disrupt splicing and trans-acting mutations that affecting RNA-dependent functions that cause disease. Information such as EntrezGeneID, gene genomic sequence, mutation (nucleotide substitutions, deletions and insertions), mutation location within the gene, organism, detailed description of the splicing mutation and references are also given. Users are able to submit new entries to the database. This database integrating RNA splicing and disease associations would be helpful for understanding not only the RNA splicing but also its contribution to disease. In SpliceDisease database, they manually curated 2337 splicing mutation disease entries involving 303 genes and 370 diseases, which have been supported experimentally in 898 publications. The SpliceDisease database provides information including the change of the nucleotide in the sequence, the location of the mutation on the gene, the reference PubMed ID and detailed description for the relationship among gene mutations, splicing defects and diseases. They standardized the names of the diseases and genes and provided links for these genes to NCBI and UCSC genome browser for further annotation and genomic sequences. For the location of the mutation, they give direct links of the entry to the respective position/region in the genome browser.

Abbreviations: SpliceDisease

Synonyms: Splice Disease, SpliceDisease Database Site, Splice Disease Database, SpliceDisease Database

Resource Type: storage service resource, data or information resource, service resource, database, data repository

Defining Citation: PMID:22139928

Keywords: rna splicing, mutation, disease, gene, genomic sequence, nucleotide substitution, deletion, insertion, mutation location, splicing mutation, nucleotide, disease association, bio.tools

Funding: National Natural Science Foundation of China 81001481

Availability: The community can contribute to this resource

Resource Name: SpliceDisease

Resource ID: SCR_006130

Alternate IDs: biotools:splicedisease_db, nlx_151614

Alternate URLs: https://bio.tools/splicedisease_db

Record Creation Time: 20220129T080234+0000

Record Last Update: 20250410T065417+0000

Ratings and Alerts

No rating or validation information has been found for SpliceDisease.

No alerts have been found for SpliceDisease.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 2 mentions in open access literature.

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

Li P, et al. (2016) Fusing literature and full network data improves disease similarity computation. BMC bioinformatics, 17(1), 326.

Jian X, et al. (2014) In silico prediction of splice-altering single nucleotide variants in the human genome. Nucleic acids research, 42(22), 13534.