Resource Summary Report

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Human Splicing Finder

RRID:SCR_005181
Type: Tool

Proper Citation

Human Splicing Finder (RRID:SCR_005181)

Resource Information

URL: http://www.umd.be/HSF3/

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Description: Software tool to help study pre-mRNA splicing and to better understand intronic and exonic mutations leading to splicing defects. To calculate the consensus values of potential splice sites and search for branch points, new algorithms were developed. Furthermore, they have integrated all available matrices to identify exonic and intronic motifs, as well as new matrices to identify hnRNP A1, Tra2-? and 9G8.

Abbreviations: HSF

Resource Type: service resource, production service resource, analysis service resource, data analysis service

Defining Citation: PMID:19339519

Keywords: splicing, mutation, splicing signal, sequence, transcript, nucleotide, exon, bio.tools

Availability: Acknowledgement requested

Resource Name: Human Splicing Finder

Resource ID: SCR_005181

Alternate IDs: biotools:human_splicing_finder, OMICS_00176

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

Old URLs: http://www.umd.be/HSF/

Record Creation Time: 20220129T080228+0000

Record Last Update: 20260731T042710+0000

Ratings and Alerts

No rating or validation information has been found for Human Splicing Finder.

No alerts have been found for Human Splicing Finder.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 963 mentions in open access literature.

Listed below are recent publications. The full list is available at kravitz2 .

Yalcouyé A, et al. (2025) Whole-exome sequencing reveals known and candidate genes for hearing impairment in Mali. HGG advances, 6(1), 100391.

D'Abrusco F, et al. (2025) Pathogenic cryptic variants detectable through exome data reanalysis significantly increase the diagnostic yield in Joubert syndrome. European journal of human genetics : EJHG, 33(1), 72.

Qiao F, et al. (2025) A Novel EBP c.452A>G Mutation Identified in a Girl with Conradi-Hünermann-Happle Syndrome Presenting with Hydronephrosis. The application of clinical genetics, 18, 63.

Shao Y, et al. (2025) Identifying six single nucleotide variants in the COL17A1 gene that alter RNA splicing: database analysis and minigene assays. Scientific reports, 15(1), 11387.

Tian A, et al. (2025) Early diagnosed Zaki syndrome: identification of two novel WLS variants and a literature review. Italian journal of pediatrics, 51(1), 209.

Hendricks RM, et al. (2025) Genome-first determination of the prevalence and penetrance of eight germline myeloid malignancy predisposition genes: a study of two population-based cohorts. Leukemia, 39(2), 400.

Zheng Y, et al. (2025) Clinical and genetic landscape of optic atrophy in 826 families: insights from 50 nuclear genes. Brain : a journal of neurology, 148(5), 1604.

Chen D, et al. (2025) Effects of a Novel COL4A3 Homozygous/Heterozygous Splicing Mutation on the Mild Phenotype in a Family With Autosomal Recessive Alport Syndrome and a Literature Review. Molecular genetics & genomic medicine, 13(2), e70053.

Li X, et al. (2025) G6PC3 promotes genome maintenance and is a candidate mammary tumor suppressor. JCI insight, 10(11).

Zhang H, et al. (2025) Genetic spectrum of congenital cataract with optional ocular and multisystem abnormalities. BMC medical genomics, 18(1), 189.

Rashid A, et al. (2025) Exome sequencing identifies a homozygous splice site variant in RP1 as the underlying cause of autosomal recessive retinitis pigmentosa in a Pakistani family. Annals of medicine, 57(1), 2470953.

Turgut GT, et al. (2025) Aarskog Syndrome: Deep Phenotyping and Genomic Landscape of a New Cohort Including Adult Patients. Clinical genetics, 108(3), 334.

Díaz-Belloso R, et al. (2025) HMGCR genetic variability in Parkinson's disease in a Spanish cohort: associations with lipid metabolism and early onset. Journal of neurology, 272(10), 671.

Zhang K, et al. (2025) Whole Exome Sequencing Identifies Novel Splicing Variants in the PTPRQ Gene and Their Mechanisms in Autosomal Recessive Non-Syndromic Hearing Loss. Journal of otology, 20(3), 204.

Hong Z, et al. (2025) Unveiling the Pathogenic Role of Novel CPLANE1 Compound Heterozygous Variants in Joubert Syndrome: Insights Into mRNA Stability and NMD Pathway. Journal of cellular and molecular medicine, 29(5), e70484.

Aspromonte MC, et al. (2025) Genetic variants and phenotypic data curated for the CAGI6 intellectual disability panel challenge. Human genetics, 144(2-3), 309.

Zhang B, et al. (2025) Identified five variants in CFTR gene that alter RNA splicing by minigene assay. Frontiers in genetics, 16, 1543623.

Chin JJ, et al. (2025) Investigation of GSDME results in the identification of the first pathogenic synonymous variants and genotype-phenotype correlations. Human genetics, 144(11-12), 1127.

Bu H, et al. (2025) Clinical and genetic characteristics of diseases caused by CFTR gene mutations in 15 Chinese children: a retrospective analysis. BMC pediatrics, 25(1), 701.

Bonetti E, et al. (2025) RENOVO-NF1 accurately predicts NF1 missense variant pathogenicity. Human genomics, 19(1), 106.