

# 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:** analysis service resource, data analysis service, production service resource, service resource

**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](https://bio.tools/human_splicing_finder)

**Old URLs:** <http://www.umd.be/HSF/>

**Record Creation Time:** 20220129T080228+0000

**Record Last Update:** 20260912T200136+0000

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## Ratings and Alerts

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

No alerts have been found for Human Splicing Finder.

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## Data and Source Information

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

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## Usage and Citation Metrics

We found 1009 mentions in open access literature.

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

Akkus N, et al. (2026) Whole Exome Sequencing in Patients With Developmental Delay/Intellectual Disability (DD/ID), Epilepsy and the First Turkish Patient Diagnosed With BCL11A-Related Intellectual Disability. *Molecular genetics & genomic medicine*, 14(1), e70180.

Liu J, et al. (2026) Whole-exome sequencing increases variant detection compared to karyotyping and CMA in an unselected FGR cohort. *Scientific reports*, 16(1).

Zanotti S, et al. (2026) Expanding the genetic landscape of Dusty Core Disease: new RYR1 variants in Italian patients. *European journal of human genetics : EJHG*, 34(5), 609.

Zhang H, et al. (2026) Molecular mechanisms and therapeutic strategies for the recurrent F9 (c.520 + 13 A > G) variant in hemophilia B. *Human genomics*, 20(1), 33.

Ten Hoor MAC, et al. (2026) Molecular characterization of Cdh12-SCON conditional knockout mice reveals unexpected splicing changes. *Transgenic research*, 35(1).

Aloi C, et al. (2026) Deciphering Silence: Functional Studies of GCK Synonymous and Nonsense Variants and Their Importance in Understanding Diabetes. *Genes*, 17(2).

Othman AA, et al. (2026) Genotypic and phenotypic features of 23 Egyptian patients with tuberous sclerosis complex. *BMC pediatrics*, 26(1).

Li JN, et al. (2026) Clinical features and genetic analysis of androgen receptor gene variants in 30 prepubertal patients with androgen insensitivity syndrome. *Asian journal of andrology*, 28(3), 335.

Serpieri V, et al. (2026) Bi-allelic variants in FSD1L cause a neurodevelopmental disorder overlapping with L1 syndrome. *American journal of human genetics*, 113(3), 600.

Can ND, et al. (2026) Variant reclassification in cancer susceptibility genes and an updated variant spectrum of Turkish breast and colorectal cancer patients. *Human genomics*, 20(1), 39.

- Li Y, et al. (2026) Multilevel Genetic and Functional Assessment of an ARR3 Frameshift Variant in Early-Onset High Myopia. *International journal of genomics*, 2026, 8894003.
- Zhao D, et al. (2026) Stepwise genetic testing strategy identified pathogenic variants in 10 Chinese duchenne muscular dystrophy patients. *Frontiers in genetics*, 17, 1805459.
- Su J, et al. (2026) Identification of a new frameshift homozygous variant of PEX3 gene in a preterm infant with profound global developmental delay and bilateral ptosis: a case report and updated literature review. *BMC pediatrics*, 26(1), 81.
- Durmaz CD, et al. (2026) TMC6/8-associated epidermodysplasia verruciformis: germline variants and a complex structural alteration in a skin cancer predisposition syndrome. *European journal of human genetics : EJHG*, 34(3), 429.
- Lim JH, et al. (2026) Personalized pharmacokinetic-pharmacodynamic guided therapy via an induced pluripotent stem cell-derived multi-organoid platform in NF1-mutant breast cancer. *Signal transduction and targeted therapy*, 11(1).
- Koparir A, et al. (2026) Chromatinopathies: clinically overlapping disorders, revealing novel variants and their DNA methylation signatures. *Clinical epigenetics*, 18(1).
- Alafghani R, et al. (2026) Clinical, genetic and bioinformatic analysis of Saudi families with Joubert syndrome and related disorders. *Human genomics*, 20(1).
- Pantea CL, et al. (2026) Phenotypic Variability Associated with Jagunal Homolog 1 (JAGN1) Deficiency Caused by the c.63G>T Variant. *International journal of molecular sciences*, 27(4).
- Cammarota F, et al. (2026) Molecular Basis of Adenomatous Gastrointestinal Polyposis Syndromes: Role of Pathogenic and Benign Variants in Disease Onset. *Biomedicines*, 14(2).
- Liu S, et al. (2026) Genetic Landscape and Clinical Characterization of FRMD7-Related Infantile Nystagmus Based on Large In-House Datasets and Literature Review. *Investigative ophthalmology & visual science*, 67(1), 30.