## **Resource Summary Report**

Generated by <u>dkNET</u> on May 20, 2025

# ESEfinder 3.0

RRID:SCR\_007088 Type: Tool

**Proper Citation** 

ESEfinder 3.0 (RRID:SCR\_007088)

#### **Resource Information**

URL: http://rulai.cshl.edu/cgi-bin/tools/ESE3/esefinder.cgi?process=home

Proper Citation: ESEfinder 3.0 (RRID:SCR\_007088)

**Description:** A web-based resource that facilitates rapid analysis of exon sequences to identify putative exonic splicing enhancers (ESEs) responsive to the human SR proteins SF2/ASF, SC35, SRp40 and SRp55, and to predict whether exonic mutations disrupt such elements.

Abbreviations: ESEfinder

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

Defining Citation: PMID:12824367

Keywords: exonic splicing enhancer, sr protein, bio.tools

Funding: NIGMS GM42699; NCI CA88351; NHGRI HG01696

**Availability:** Free for non-profit use, Non-commercial, Acknowledgement requested, Commercial use with license

Resource Name: ESEfinder 3.0

Resource ID: SCR\_007088

Alternate IDs: biotools:esefinder, nif-0000-30496

Alternate URLs: http://rulai.cshl.edu/tools/ESE2/, https://bio.tools/esefinder

Old URLs: http://exon.cshl.edu/ESE/

**Record Creation Time:** 20220129T080239+0000

Record Last Update: 20250519T204717+0000

### **Ratings and Alerts**

No rating or validation information has been found for ESEfinder 3.0.

No alerts have been found for ESEfinder 3.0.

#### Data and Source Information

Source: <u>SciCrunch Registry</u>

#### **Usage and Citation Metrics**

We found 204 mentions in open access literature.

Listed below are recent publications. The full list is available at <u>dkNET</u>.

Legebeke J, et al. (2025) Uplift of genetic diagnosis of rare respiratory disease using airway epithelium transcriptome analysis. Human molecular genetics, 34(2), 148.

Munteanu CV, et al. (2024) In silico splicing analysis of the PMS2 gene: exploring alternative molecular mechanisms in PMS2-associated Lynch syndrome. BMC genomic data, 25(1), 100.

Zhang Y, et al. (2024) Novel Germline KIT Variants in Families With Severe Piebaldism: Case Series and Literature Review. Journal of clinical laboratory analysis, 38(11-12), e25073.

Pan D, et al. (2024) Splicing factor hnRNPA1 regulates alternative splicing of LOXL2 to enhance the production of LOXL2?13. The Journal of biological chemistry, 300(7), 107414.

Santos-Rebouças CB, et al. (2024) Immune response stability to the SARS-CoV-2 mRNA vaccine booster is influenced by differential splicing of HLA genes. Scientific reports, 14(1), 8982.

Donelson CJH, et al. (2024) Functional evaluation of rare variants in complement factor I using a minigene assay. Frontiers in immunology, 15, 1446081.

Koparir A, et al. (2024) Zebrafish as a model to investigate a biallelic gain-of-function variant in MSGN1, associated with a novel skeletal dysplasia syndrome. Human genomics, 18(1), 23.

Spangsberg Petersen US, et al. (2024) Regulating PCCA gene expression by modulation of pseudoexon splicing patterns to rescue enzyme activity in propionic acidemia. Molecular therapy. Nucleic acids, 35(1), 102101.

García-Bohórquez B, et al. (2024) Exploring non-coding variants and evaluation of antisense oligonucleotides for splicing redirection in Usher syndrome. Molecular therapy. Nucleic acids, 35(4), 102374.

Liu L, et al. (2023) HBV Enhances Sorafenib Resistance in Hepatocellular Carcinoma by Reducing Ferroptosis via SRSF2-Mediated Abnormal PCLAF Splicing. International journal of molecular sciences, 24(4).

Jin B, et al. (2023) MEN1 is a regulator of alternative splicing and prevents R-loop-induced genome instability through suppression of RNA polymerase II elongation. Nucleic acids research, 51(15), 7951.

Choi SY, et al. (2023) Prediction of medication-related osteonecrosis of the jaws using machine learning methods from estrogen receptor 1 polymorphisms and clinical information. Frontiers in medicine, 10, 1140620.

Seyama R, et al. (2023) A missense variant at the RAC1-PAK1 binding site of RAC1 inactivates downstream signaling in VACTERL association. Scientific reports, 13(1), 9789.

Gerber S, et al. (2023) Autosomal recessive pathogenic MSTO1 variants in hereditary optic atrophy. EMBO molecular medicine, 15(8), e16090.

Zhong J, et al. (2023) A novel splicing variant of VCAN identified in a Chinese family initially diagnosed with familial exudative vitreoretinopathy. Molecular genetics & genomic medicine, 11(2), e2083.

Seidizadeh O, et al. (2023) Population-based prevalence and mutational landscape of von Willebrand disease using large-scale genetic databases. NPJ genomic medicine, 8(1), 31.

Elworthy S, et al. (2023) Activated PI3K delta syndrome 1 mutations cause neutrophilia in zebrafish larvae. Disease models & mechanisms, 16(3).

Vona B, et al. (2023) Unraveling haplotype errors in the DFNA33 locus. Frontiers in genetics, 14, 1214736.

Thomassen M, et al. (2022) Clinical, splicing, and functional analysis to classify BRCA2 exon 3 variants: Application of a points-based ACMG/AMP approach. Human mutation, 43(12),

#### 1921.

Holcomb DD, et al. (2022) Protocol to identify host-viral protein interactions between coagulation-related proteins and their genetic variants with SARS-CoV-2 proteins. STAR protocols, 3(3), 101648.