Resource Summary Report

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Exonic Splicing Enhancer Finder

RRID:SCR_002835
Type: Tool

Proper Citation

Exonic Splicing Enhancer Finder (RRID:SCR_002835)

Resource Information

URL: http://rulai.cshl.edu/tools/ESE

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Description: A web-based analysis service for identifying exonic splicing enhancers in eukaryotic genes. ESEfinder accept sequences in the FASTA format. A typical mammalian gene is composed of several relatively short exons that are interrupted by much longer introns. To generate correct mature mRNAs, the exons must be identified and joined together precisely and efficiently, in a process that requires the coordinated action of five small nuclear (sn)RNAs (U1, U2, U4, U5 and U6) and more than 60 polypeptides. The inaccurate recognition of exon/intron boundaries or the failure to remove an intron generates aberrant mRNAs that are either unstable or code for defective or deleterious protein isoforms. Exonic enhancers are thought to serve as binding sites for specific serine/arginine-rich (SR) proteins, a family of structurally related and highly conserved splicing factors characterized by one or two RNA-recognition motifs (RRM) and by a distinctive C-terminal domain highly enriched in RS dipeptides (the RS domain). The RRMs mediate sequence-specific binding to the RNA, and so determine substrate specificity, whereas the RS domain appears to be involved mainly in protein-protein interactions. SR proteins bound to ESEs can promote exon definition by directly recruiting the splicing machinery through their RS domain and/or by antagonizing the action of nearby silencer elements. Sponsors: ESEfinder is supported by the Cold Spring Harbor Laboratory.

Synonyms: ESEfinder

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

Keywords: element, enhancer, eukaryotic, exon, exonic, gene, analysis, arginine, boundary, c-terminal, dipeptide, intron, isoform, mammalian, mrna, nuclear, polypeptide, protein, recognition, rna, serine, service, snrna, splice

Availability: Free, Freely available

Resource Name: Exonic Splicing Enhancer Finder

Resource ID: SCR_002835

Alternate IDs: nif-0000-25204

Record Creation Time: 20220129T080215+0000

Record Last Update: 20260821T042559+0000

Ratings and Alerts

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

No alerts have been found for Exonic Splicing Enhancer Finder.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 66 mentions in open access literature.

Listed below are recent publications. The full list is available at nidm-terms .

Xu N, et al. (2024) Genetic insights into the 'sandwich fusion' subtype of Klippel-Feil syndrome: novel FGFR2 mutations identified by 21 cases of whole-exome sequencing. Orphanet journal of rare diseases, 19(1), 141.

Zhen Z, et al. (2024) Novel mutation in XPNPEP3 in a patient with heart failure without nephronophthisis-like nephropathy (NPHPL1): case report and literature review. BMC pediatrics, 24(1), 632.

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.

Lee J, et al. (2022) Noncanonical Splice Site and Deep Intronic FRMD7 Variants Activate Cryptic Exons in X-linked Infantile Nystagmus. Translational vision science & technology, 11(6), 25.

Pio MG, et al. (2021) A novel mutation in intron 11 donor splice site, responsible of a rare genotype in thyroglobulin gene by altering the pre-mRNA splincing process. Cell expression and bioinformatic analysis. Molecular and cellular endocrinology, 522, 111124.

Wang SY, et al. (2021) A synonymous mutation in IGF-1 impacts the transcription and translation process of gene expression. Molecular therapy. Nucleic acids, 26, 1446.

Nassisi M, et al. (2019) Prevalence of ABCA4 Deep-Intronic Variants and Related Phenotype in An Unsolved "One-Hit" Cohort with Stargardt Disease. International journal of molecular sciences, 20(20).

Ni J, et al. (2019) Genotype and phenotype analysis of a cohort of patients with congenital hyperinsulinism based on DOPA-PET CT scanning. European journal of pediatrics, 178(8), 1161.

Kim JH, et al. (2019) SON haploinsufficiency causes impaired pre-mRNA splicing of CAKUT genes and heterogeneous renal??phenotypes. Kidney international, 95(6), 1494.

Jdila MB, et al. (2019) A novel C-terminal truncated mutation in hCDKL5 protein causing a severe West syndrome: Comparison with previous truncated mutations and genotype/phenotype correlation. International journal of developmental neuroscience : the official journal of the International Society for Developmental Neuroscience, 72, 22.

Neuh??user K, et al. (2019) Assessment of the role of translationally controlled tumor protein 1 (TPT1/TCTP) in breast cancer susceptibility and ATM signaling. Clinical and translational radiation oncology, 15, 99.

Esmaeili R, et al. (2018) Unique CD44 intronic SNP is associated with tumor grade in breast cancer: a case control study and in silico analysis. Cancer cell international, 18, 28.

Kim JS, et al. (2016) Cardiovascular autonomic dysfunctions in elderly patients with essential tremor: comparison with healthy controls. Neurological sciences : official journal of the Italian Neurological Society and of the Italian Society of Clinical Neurophysiology, 37(5), 711.

Kim JH, et al. (2016) De Novo Mutations in SON Disrupt RNA Splicing of Genes Essential for Brain Development and Metabolism, Causing an Intellectual-Disability Syndrome. American journal of human genetics, 99(3), 711.

Di Maggio S, et al. (2016) The Mouse-Specific Splice Variant mRAGE_v4 Encodes a Membrane-Bound RAGE That Is Resistant to Shedding and Does Not Contribute to the Production of Soluble RAGE. PloS one, 11(9), e0153832.

Belforte FS, et al. (2016) Compound heterozygous DUOX2 gene mutations (c.2335-1G>C/c.3264_3267delCAGC) associated with congenital hypothyroidism. Characterization of complex cryptic splice sites by minigene analysis. Molecular and cellular endocrinology, 419, 172.

Citterio CE, et al. (2015) Novel compound heterozygous Thyroglobulin mutations c.745+1G>A/c.7036+2T>A associated with congenital goiter and hypothyroidism in a Vietnamese family. Identification of a new cryptic 5' splice site in the exon 6. Molecular and cellular endocrinology, 404, 102.

Li XW, et al. (2015) Epigenetic regulation of CDH1 exon 8 alternative splicing in gastric cancer. BMC cancer, 15, 954.

Fan ZC, et al. (2015) Uncovering the molecular pathogenesis of congenital hyperinsulinism by panel gene sequencing in 32 Chinese patients. Molecular genetics & genomic medicine, 3(6), 526.

Burrage LC, et al. (2015) De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American journal of human genetics, 97(6), 904.