Resource Summary Report

Generated by NIF on Apr 18, 2025

RESCUE-ESE

RRID:SCR_008496
Type: Tool

Proper Citation

RESCUE-ESE (RRID:SCR_008496)

Resource Information

URL: http://hollywood.mit.edu/burgelab/rescue-ese/

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Description: Specific short oligonucleotide sequences that enhance pre-mRNA splicing when present in exons, termed exonic splicing enhancers (ESEs), play important roles in constitutive and alternative splicing (ESE References). A hybrid computational/experimental method, RESCUE-ESE, was recently developed for identifying sequences with ESE activity. In this approach, specific hexanucleotide sequences are identified as candidate ESEs on the basis that they have both significantly higher frequency of occurrence in exons than in introns and also significantly higher frequency in exons with weak (non-consensus) splice sites than in exons with strong (consensus) splice sites. Representative hexamers from ten different classes of candidate ESEs, together with 6 or 7 bases of flanking sequence context on each side, were introduced into a weak (poorly spliced) exon in a splicing reporter construct. These reporter minigenes were then transfected into cultured cells, where they are transcribed and spliced, and the relative level of inclusion of the test exon was assayed by quantitative (radio-labeled) RT-PCR. Point mutants of these sequences were also analyzed to confirm the precise motifs responsible for ESE activity. The RESCUE-ESE approach identified 238 hexamers as candidate ESEs using a large database of human genes of known exon-intron structure containing over 30,000 nonredudant exons. In more recent analyses by Yeo et al., the RESCUE-ESE approach was utilized to predict hexamers as candidate ESEs in other vertebrate genes, namely, Fugu rubipes, Zebrafish and Mouse. This allows the identification of motifs that are conserved in vertebrates. This web server allows a sequence to be checked for presence of these candidate ESE hexamers.

Synonyms: RESCUE-ESE

Resource Type: portal, database, organization portal, data or information resource

Keywords: bio.tools, FASEB list

Funding:

Resource Name: RESCUE-ESE

Resource ID: SCR_008496

Alternate IDs: biotools:rescue-ese, nif-0000-31403

Alternate URLs: https://bio.tools/rescue-ese

Old URLs: http://genes.mit.edu/burgelab/rescue-ese/

Record Creation Time: 20220129T080247+0000

Record Last Update: 20250417T065331+0000

Ratings and Alerts

No rating or validation information has been found for RESCUE-ESE.

No alerts have been found for RESCUE-ESE.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 95 mentions in open access literature.

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

Recinos Y, et al. (2024) Lineage-specific splicing regulation of MAPT gene in the primate brain. Cell genomics, 4(6), 100563.

Chatterjee S, et al. (2022) Whole exome sequencing identifies a novel splice-site mutation in IMPG2 gene causing Stargardt-like juvenile macular dystrophy in a north Indian family. Gene, 816, 146158.

Orlans HO, et al. (2021) Mirtron-mediated RNA knockdown/replacement therapy for the treatment of dominant retinitis pigmentosa. Nature communications, 12(1), 4934.

Mekki C, et al. (2021) Prenatal Ultrasound Suspicion of Cystic Fibrosis in a Multiethnic Population: Is Extensive CFTR Genotyping Needed? Genes, 12(5).

Riolo G, et al. (2021) What's Wrong in a Jump? Prediction and Validation of Splice Site Variants. Methods and protocols, 4(3).

Coutinho MF, et al. (2020) Molecular Characterization of a Novel Splicing Mutation underlying Mucopolysaccharidosis (MPS) type VI-Indirect Proof of Principle on Its Pathogenicity. Diagnostics (Basel, Switzerland), 10(2).

Cascio L, et al. (2020) Abnormalities in the genes that encode Large Amino Acid Transporters increase the risk of Autism Spectrum Disorder. Molecular genetics & genomic medicine, 8(1), e1036.

Odgerel Z, et al. (2019) Whole genome sequencing and rare variant analysis in essential tremor families. PloS one, 14(8), e0220512.

Tonin R, et al. (2019) Progressive myoclonus epilepsy in Gaucher Disease due to a new Gly-Gly mutation causing loss of an Exonic Splicing Enhancer. Journal of neurology, 266(1), 92.

InanlooRahatloo K, et al. (2019) Whole-Transcriptome Analysis Reveals Dysregulation of Actin-Cytoskeleton Pathway in Intellectual Disability Patients. Neuroscience, 404, 423.

Anna A, et al. (2018) Splicing mutations in human genetic disorders: examples, detection, and confirmation. Journal of applied genetics, 59(3), 253.

Girardelli M, et al. (2018) Genetic profile of patients with early onset inflammatory bowel disease. Gene, 645, 18.

Suarez-Artiles L, et al. (2018) Splicing Analysis of Exonic OCRL Mutations Causing Lowe Syndrome or Dent-2 Disease. Genes, 9(1).

Brasil S, et al. (2018) Improving the diagnosis of cobalamin and related defects by genomic analysis, plus functional and structural assessment of novel variants. Orphanet journal of rare diseases, 13(1), 125.

Rodrigues M, et al. (2018) Outlier response to anti-PD1 in uveal melanoma reveals germline MBD4 mutations in hypermutated tumors. Nature communications, 9(1), 1866.

Lykens NM, et al. (2017) AMPA GluA1-flip targeted oligonucleotide therapy reduces neonatal seizures and hyperexcitability. PloS one, 12(2), e0171538.

Prykhozhij SV, et al. (2017) A rapid and effective method for screening, sequencing and reporter verification of engineered frameshift mutations in zebrafish. Disease models & mechanisms, 10(6), 811.

Kapplinger JD, et al. (2017) KCNQ1 p.L353L affects splicing and modifies the phenotype in a founder population with long QT syndrome type 1. Journal of medical genetics, 54(6), 390.

Álvarez-Satta M, et al. (2017) Functional analysis by minigene assay of putative splicing variants found in Bardet-Biedl syndrome patients. Journal of cellular and molecular medicine, 21(10), 2268.

Razeen MM, et al. (2016) Correlating Photoreceptor Mosaic Structure to Clinical Findings in Stargardt Disease. Translational vision science & technology, 5(2), 6.