SMN2 exon 7 splicing is inhibited by binding of hnRNP A1 to a common ESS motif that spans the 3psplice site

DOI: 10.1002/humu.21419
Human Mutation, 2011, 32, 220-30.

#	Paper	IF	Citations
35	A leaky splicing mutation affecting SMN1 exon 7 inclusion explains an unexpected mild case of spinal muscular atrophy. *Human Mutation*, **2011**, 32, 989-94	4.7	15
34	A synonymous mutation in SPINK5 exon 11 causes Netherton syndrome by altering exonic splicing regulatory elements. *Journal of Human Genetics*, **2012**, 57, 311-5	4.3	12
33	Guidelines for splicing analysis in molecular diagnosis derived from a set of 327 combined in silico/in vitro studies on BRCA1 and BRCA2 variants. *Human Mutation*, **2012**, 33, 1228-38	4.7	171
32	A synonymous polymorphic variation in ACADM exon 11 affects splicing efficiency and may affect fatty acid oxidation. *Molecular Genetics and Metabolism*, **2013**, 110, 122-8	3.7	18
31	hnRNP A1: the Swiss army knife of gene expression. *International Journal of Molecular Sciences*, **2013**, 14, 18999-9024	6.3	170
30	Novel GUCA1A mutations suggesting possible mechanisms of pathogenesis in cone, cone-rod, and macular dystrophy patients. *BioMed Research International*, **2013**, 2013, 517570	3	22
29	Functional analysis of a large set of BRCA2 exon 7 variants highlights the predictive value of hexamer scores in detecting alterations of exonic splicing regulatory elements. *Human Mutation*, **2013**, 34, 1547-57	4.7	40
28	Exon first nucleotide mutations in splicing: evaluation of in silico prediction tools. *PLoS ONE*, **2014**, 9, e89570	3.7	14
27	Absence of an intron splicing silencer in porcine Smn1 intron 7 confers immunity to the exon skipping mutation in human SMN2. *PLoS ONE*, **2014**, 9, e98841	3.7	3
26	Targeting SR proteins improves SMN expression in spinal muscular atrophy cells. *PLoS ONE*, **2014**, 9, e115205	3.7	34
25	The ETFDH c.158A>G variation disrupts the balanced interplay of ESE- and ESS-binding proteins thereby causing missplicing and multiple Acyl-CoA dehydrogenation deficiency. *Human Mutation*, **2014**, 35, 86-95	4.7	22
24	Global identification of hnRNP A1 binding sites for SSO-based splicing modulation. *BMC Biology*, **2016**, 14, 54	7.3	41
23	Quantitative proteomics suggests metabolic reprogramming during ETHE1 deficiency. *Proteomics*, **2016**, 16, 1166-76	4.8	10
22	The prevalent deep intronic c. 639+919 G>A GLA mutation causes pseudoexon activation and Fabry disease by abolishing the binding of hnRNPA1 and hnRNP A2/B1 to a splicing silencer. *Molecular Genetics and Metabolism*, **2016**, 119, 258-269	3.7	13
21	Differential hnRNP D isoform incorporation may confer plasticity to the ESSV-mediated repressive state across HIV-1 exon 3. *Biochimica Et Biophysica Acta - Gene Regulatory Mechanisms*, **2017**, 1860, 205-217	6	8
20	RNA-sequencing of a mouse-model of spinal muscular atrophy reveals tissue-wide changes in splicing of U12-dependent introns. *Nucleic Acids Research*, **2017**, 45, 395-416	20.1	63
19	Transcriptional and Splicing Regulation of Spinal Muscular Atrophy Genes. **2017**, 75-97		7

18	Blocking of an intronic splicing silencer completely rescues IKBKAP exon 20 splicing in familial dysautonomia patient cells. *Nucleic Acids Research*, **2018**, 46, 7938-7952	20.1	9
17	Mechanism of Splicing Regulation of Spinal Muscular Atrophy Genes. *Advances in Neurobiology*, **2018**, 20, 31-61	2.1	51
16	Splicing Enhancers at Intron-Exon Borders Participate in Acceptor Splice Sites Recognition. *International Journal of Molecular Sciences*, **2020**, 21,	6.3	2
15	Identification of SRSF10 as a regulator of SMN2 ISS-N1. *Human Mutation*, **2021**, 42, 246-260	4.7	3
14	A Comprehensive Analysis of the Role of hnRNP A1 Function and Dysfunction in the Pathogenesis of Neurodegenerative Disease. *Frontiers in Molecular Biosciences*, **2021**, 8, 659610	5.6	10
13	HNRNP A1 Promotes Lung Cancer Cell Proliferation by Modulating Translation. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	0
12	A high-throughput genome-wide RNAi screen identifies modifiers of survival motor neuron protein. *Cell Reports*, **2021**, 35, 109125	10.6	0
11	hnRNP A/B Proteins: An Encyclopedic Assessment of Their Roles in Homeostasis and Disease. *Biology*, **2021**, 10,	4.9	5
10	Heterogeneous Nuclear Ribonucleoproteins: Implications in Neurological Diseases. *Molecular Neurobiology*, **2021**, 58, 631-646	6.2	11
9	Identification of six novel PTH1R mutations in families with a history of primary failure of tooth eruption. *PLoS ONE*, **2013**, 8, e74601	3.7	35
8	The GENDULF algorithm: mining transcriptomics to uncover modifier genes for monogenic diseases. *Molecular Systems Biology*, **2020**, 16, e9701	12.2	1
7	Tandem hnRNP A1 RNA recognition motifs act in concert to repress the splicing of survival motor neuron exon 7. *ELife*, **2017**, 6,	8.9	34
6	Nucleotides in both donor and acceptor splice sites are responsible for choice in NAGNAG tandem splice sites. *Cellular and Molecular Life Sciences*, **2021**, 78, 6979-6993	10.3	0
5	Vulnerable exons, like ACADM exon 5, are highly dependent on maintaining a correct balance between splicing enhancers and silencers.. *Human Mutation*, **2021**,	4.7	0
4	Effects of inhibitors of SLC9A-type sodium-proton exchangers on Survival Motor Neuron2 (SMN2) mRNA splicing and expression. *Molecular Pharmacology*, MOLPHARM-AR-2022-000529	4.3	
3	Position-dependent effects of hnRNP A1/A2 in SMN1/2 exon7 splicing. **2022**, 1865, 194875		0
2	A splicing silencer in SMN2 intron 6 is critical in spinal muscular atrophy.		0
1	Type I PRMT inhibitor MS023 promotes SMN2 exon 7 inclusion and synergizes with nusinersen to rescue the phenotype of SMA mice.		0