

## Ejemplo comparación de resultados predictores in silico

Cambio de estudio COL25A1 c.1020+571C>T (chr 4:108883607 C/T, COSV67652663 o NM\_198721: c.1020+571C>T)

Exón 19 e intrones adyacentes:

```
aataatgaaatattgggagttttcttctcttttgactcctcttttctcgscatctgacggagat
accaatgtgcagtgaactgaaaggaaagagagaaaaatcccaaaaaatctgtgaagagggtca
accattttctaaatcaagagttgactgaattgcataatctcatcaaagtttttgaaagta
gacggttttttttatttatctttgcagaaagtattgagccaggaaacatttgccatgttttt
ctccacataattgtgactgagtgaaatacattctataaatgctatgatattttctgtgtag
GGTGAAACAGGGCTTCCTGGGCTTCCTGGACTTCCGGGATAAAG
gtaaatactgcacggacagtctcggcttcadagaactgttaattatgagattcaccaaaat
tggaggaaaatagggaataatgaaaataaaaaatagaattgctgttctctaagtactctcc
agccatctcaactaactaatttttaaaattaaaaattaagacagatacctctaaagttt
gcatggcaatatggattgtttattttctccatttaacttactactgcccctaaataaat
gtaaatgtcccttcatttgaagtcaaggaattctttaagtcctaaaatgtctctttacc
ttcatatttttagttttattcttaagacccctatcagattttgatcagaattagagtcttaa
catcccataaaagctaacaatttaggcagctgtttttgtgattcactcattataataatct
ctctaaaacatttatcttctacttctctcagtaattatcactgcaaagtacaaatcata
ttactaaaatattttgaccacaatacactgtccccataaaactttctaataatctttt
gaaataattgcttaagcttagtttgaaaaacatttggttttaaatggatgtaaacactta
aaaatgttaaaaacattttaaaaaatgttttgaattttttgcatggcaactgacaagac
```

El cambio se encuentra en la penúltima línea del intrón 19 (la **c** en color rojo subrayada de azul).

Se va a obtener los resultados que produce analizar esta variable con los diferente predictores y ver cuál de ellos es más preciso.

## NetGene2

### Donor splice sites, direct strand

pos	5'→3'	phase	strand	confidence	5'	exon	intron	3'
346		0	+	0.92	GGGGATAAAG	^GTAATACTG	H	

### Donor splice sites, complement strand

pos	3'→5'	pos	5'→3'	phase	strand	confidence	5'	exon	intron	3'
644		362		1	-	0.41	AAATATGAAG	^GTAAAGAGAC		
566		440		1	-	0.41	AGGGGCAGTA	^GTAAGTTAAA		

### Acceptor splice sites, direct strand

pos	5'→3'	phase	strand	confidence	5'	intron	exon	3'
206		1	+	0.56	ATCTTTGCAG	^AAAGTATTGA		
300		0	+	0.97	TCCTGTGTAG	^GGTGAACCAG		
797		1	+	0.27	TTCCTCTCAG	^TAATTATCAC		

### Acceptor splice sites, complement strand

pos	3'→5'	pos	5'→3'	phase	strand	confidence	5'	intron	exon	3'
807		199		2	-	0.16	TACTTTGCAG	^TGATAATTAC		
77		929		1	-	0.00	TTCCTTTTCAG	^TTCACGTGCAC		
50		956		1	-	0.00	TCTCCGTCAG	^ATGGCGAGAA		

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Splice Site Prediction by Neural Network (NNSplice)

Donor site predictions for wt :

Start	End	Score	Exon	Intron
339	353	1.00	gataaag	gt <sup>a</sup> aaatac
579	593	0.44	aataaat	gt <sup>a</sup> aaatgt

Donor site predictions for mut :

Start	End	Score	Exon	Intron
339	353	1.00	gataaag	gt <sup>a</sup> aaatac
579	593	0.44	aataaat	gt <sup>a</sup> aaatgt

Acceptor site predictions for wt :

Start	End	Score	Intron	Exon
186	226	0.98	tttttttatttatctttgc	ag <sup>a</sup> aaagtattgagccaggaaca
280	320	0.98	ctatatgattttcctgtgt	agg <sup>a</sup> gtgaaccagggcttcctgg
637	677	0.94	ctctttaccttcataat	ag <sup>a</sup> ttttattcctaagaccctat
650	690	0.77	tatttttagttttattccta	ag <sup>a</sup> accctatcagattttgatca
777	817	0.91	ttatcttctacttcctctc	ag <sup>a</sup> taattatcactgcaaagtac


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Start	End	Score	Intron	Exon
186	226	0.98	tttttttatttatctttgc	ag <sup>a</sup> aaagtattgagccaggaaca
280	320	0.98	ctatatgattttcctgtgt	agg <sup>a</sup> gtgaaccagggcttcctgg
637	677	0.94	ctctttaccttcataat	ag <sup>a</sup> ttttattcctaagaccctat
650	690	0.77	tatttttagttttattccta	ag <sup>a</sup> accctatcagattttgatca
777	817	0.91	ttatcttctacttcctctc	ag <sup>a</sup> taattatcactgcaaagtac

Spliceman

Point mutation	Wildtype (wt)	Mutation (mt)	L1 distance	Ranking (L1)
gaaaa(c/t)atttg	aaaaca	aaaata	30508	77%

## Human Splicing Finder

 No significant impact on splicing signals.	No significant impact on splicing signals.
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## SVM-BPfinder

seq_id	agez	ss_dist	bp_seq	bp_scr	y_cont	ppt_off	ppt_len	ppt_scr	svm_scr		
wt	88	84	gtttgaaaa		-1.91554835644	0.46835443038	4	10	19	-0.27317731	
wt	88	70	gttttaaat		-4.42972677405	0.446153846154	45	6	13	-3.9158933	
wt	88	69	ttttaaatg		-0.95804607955	0.453125	44	6	13	-2.4910165	
wt	88	59	atgtaaacc		-0.129652361591	0.481481481481	34	6	13	-1.5245197	
wt	88	51	cacttaaaa		-1.91602907304	0.45652173913	26	6	13	-1.7256453	
wt	88	50	acttaaaaa		-2.58519839667	0.466666666667	25	6	13	-1.9210819	
wt	88	42	atgttaaaa		-4.40087465531	0.486486486486	17	6	13	-2.1192171	
wt	88	41	tgttaaaaa		-1.07961071764	0.5	16	6	13	-0.75112255	
wt	88	31	attttaaaa		-5.34640335052	0.5	6	6	13	-1.7887902	
wt	88	30	ttttaaaaa		-1.98390511134	0.52	5	6	13	-0.4024556	
wt	88	18	ttttgaaat		-1.37529902636	0.615384615385	3	8	18	0.039824763	
mut	88	84	gtttgaaaa		-1.91554835644	0.46835443038	4	10	20	-0.26386146	
mut	88	70	gttttaaat		-4.42972677405	0.446153846154	45	6	13	-3.9158933	
mut	88	69	ttttaaatg		-0.95804607955	0.453125	44	6	13	-2.4910165	
mut	88	59	atgtaaacc		-0.129652361591	0.481481481481	34	6	13	-1.5245197	
mut	88	51	cacttaaaa		-1.91602907304	0.45652173913	26	6	13	-1.7256453	
mut	88	50	acttaaaaa		-2.58519839667	0.466666666667	25	6	13	-1.9210819	
mut	88	42	atgttaaaa		-4.40087465531	0.486486486486	17	6	13	-2.1192171	
mut	88	41	tgttaaaaa		-1.07961071764	0.5	16	6	13	-0.75112255	
mut	88	31	attttaaaa		-5.34640335052	0.5	6	6	13	-1.7887902	
mut	88	30	ttttaaaaa		-1.98390511134	0.52	5	6	13	-0.4024556	
mut	88	18	ttttgaaat		-1.37529902636	0.615384615385	3	8	18	0.039824763	

Variant Effect Predictor tool

ENST00000399132.5:c.1020+571C>T	<a href="#">4:108883607-108883607</a>	A	 intron_variant	COL25A1	<a href="#">ENSG00000188517</a>	Transcript	<a href="#">ENST00000399126.1</a>	protein_coding	-	-	-	-	-	-	<a href="#">COSV67652663</a>
ENST00000399132.5:c.1020+571C>T	<a href="#">4:108883607-108883607</a>	A	 intron_variant	COL25A1	<a href="#">ENSG00000188517</a>	Transcript	<a href="#">ENST00000399127.5</a>	protein_coding	-	-	-	-	-	-	<a href="#">COSV67652663</a>
ENST00000399132.5:c.1020+571C>T	<a href="#">4:108883607-108883607</a>	A	 intron_variant	COL25A1	<a href="#">ENSG00000188517</a>	Transcript	<a href="#">ENST00000399132.6</a>	protein_coding	-	-	-	-	-	-	<a href="#">COSV67652663</a>
ENST00000399132.5:c.1020+571C>T	<a href="#">4:108883607-108883607</a>	A	 intron_variant  NMD_transcript_variant	COL25A1	<a href="#">ENSG00000188517</a>	Transcript	<a href="#">ENST00000494183.5</a>	nonsense_mediated_decay	-	-	-	-	-	-	<a href="#">COSV67652663</a>
ENST00000399132.5:c.1020+571C>T	<a href="#">4:108883607-108883607</a>	A	 intron_variant	COL25A1	<a href="#">ENSG00000188517</a>	Transcript	<a href="#">ENST00000610288.4</a>	protein_coding	-	-	-	-	-	-	<a href="#">COSV67652663</a>
ENST00000399132.5:c.1020+571C>T	<a href="#">4:108883607-108883607</a>	A	 intron_variant	COL25A1	<a href="#">ENSG00000188517</a>	Transcript	<a href="#">ENST00000622134.4</a>	protein_coding	-	-	-	-	-	-	<a href="#">COSV67652663</a>
ENST00000399132.5:c.1020+571C>T	<a href="#">4:108883607-108883607</a>	A	 intron_variant	COL25A1	<a href="#">ENSG00000188517</a>	Transcript	<a href="#">ENST00000642955.1</a>	protein_coding	-	-	-	-	-	-	<a href="#">COSV67652663</a>

ESEfinder

No hay resultados positivos con la posición de interés para ninguna de las secuencias.