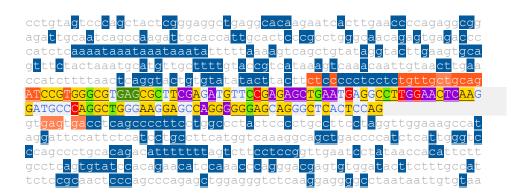
Ejemplo comparación de resultados predictores in sillico

Cambio de estudio TP53 c.1024C>T (chr17:7670685 C/T, COSV526654870 NM_000546.6: c.1024C>T)

Exón 10 e intrones adyacentes:



El cambio se encuentra en la primera línea del exón 10 (la ${\bf c}$ en color rojo).

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

NetGene2

ites, direct	strand	I				•				
		strand +	confidence 0.93					strand +	confidence 0.86	5' exon intron 3' CTCACTCCAG^GTGAGTGACC
ites, comple	ment st	rand						rand		
pos 5'->3' 433	phase 0	strand -	confidence 0.93		pos 3'->5' 275			strand -	confidence 0.93	5' exon intron 3' AGGGGGAGAA^GTAAGTATAT H
sites, dir	ect str	and			Acceptor splic	ce sites, dir	rect str	and		
			confidence	E' introp even 3'			phase			
•	pnase						1	+	0.33	TTTAACTCAG^GTACTGTGTA
	1					300	0	+	1.00	GTTGCTGCAG^ATCCGTGGGC H
						315	0	+	0.20	TGGGCGTGAG^CGCTTCGAGA
						324	0	+	0.07	GCGCTTCGAG^ATGTTCTGAG
		+				454	1	+	0.53	GCCTTCCTAG^GTTGGAAAGC
454	_	+				605	2	+	0.16	GTATCCACAG^AACATCCAAA
605	2	+	0.16	GTATCCACAG^AACATCCAAA						
sites com	nlement	strand					•			
									confidence	5' intron exon 3'
pos 5'->3'	phase	strand	confidence	5' intron exon 3'	451	257	2	-	0.17	TCCAACCTAG^GAAGGCAGGG
257	2	-	0.17	TCCAACCTAG^GAAGGCAGGG	371	337	2	-	0.51	TCCTTCCCAG^CCTGGGCATC
337	2	-	0.67	TCCTTCCCAG^CCTGGGCATC	355	353	2	-	0.17	CATCCTTGAG^TTCCAAGGCC
353	2	-	0.18	CATCCTTGAG^TTCCAAGGCC	348	360	0	-	0.18	GAGTTCCAAG^GCCTCATTCA
360	0	-	0.18	GAGTTCCAAG^GCCTCATTCA	337	371	2	-	0.17	CCTCATTCAG^CTCTCAGAAC
371	2	-	0.17	CCTCATTCAG^CTCTCGGAAC	330	378	0	-	0.19	CAGCTCTCAG^AACATCTCGA
	pos 5'->3' 408 ites, comple pos 5'->3' 433 e sites, dir pos 5'->3' 256 300 315 324 454 605 e sites, com pos 5'->3' 256 300 315 324 454 605	pos 5'->3' phase 408 2 Ites, complement st pos 5'->3' phase 433 0 sites, direct str pos 5'->3' phase 256 1 300 0 315 0 324 0 454 1 605 2 sites, complement pos 5'->3' phase 257 2 337 2 353 2 360 0	pos 5'->3' phase strand 408 2 + tes, complement strand pos 5'->3' phase strand 433 0 - sites, direct strand complement strand sites, direct strand complement strand 256 1 + 300 0 + 315 0 + 324 0 + 454 1 + 605 2 + sites, complement strand pos 5'->3' phase strand complement strand complem	pos 5'->3' phase strand confidence 408 2 + 0.93 Ites, complement strand pos 5'->3' phase strand confidence 433 0 - 0.93 Ites, direct strand pos 5'->3' phase strand confidence 256 1 + 0.43 300 0 + 1.00 315 0 + 0.19 324 0 + 0.07 454 1 + 0.53 605 2 + 0.16 Is sites, complement strand pos 5'->3' phase strand confidence 257 2 - 0.16 Ites ites, complement strand confidence 257 2 - 0.17 337 2 - 0.67 353 2 - 0.18 360 0 - 0.18	pos 5'->3' phase strand confidence 5' exon intron 3' 408 2 + 0.93 CTCACTCCAG^GTGAGTGACC H Ites, complement strand pos 5'->3' phase strand confidence 5' exon intron 3' 433 0 - 0.93 AGGGGGAGAA^GTAAGTATAT H e sites, direct strand pos 5'->3' phase strand confidence 5' intron exon 3' 256 1 + 0.43 TTTAACTCAG^GTACTGTGTAT 300 0 + 1.00 GTTGCTGCAG^ATCCGTGGGC H 315 0 + 0.19 TGGGCGTGAG^CGCTTCGAGA 324 0 + 0.07 GCGCTTCGAG^ATGTTCCGAG 454 1 + 0.53 GCCTTCCTAG^GTTGGAAAGC 605 2 + 0.16 GTATCCACAG^AACATCCAAA e sites, complement strand pos 5'->3' phase strand confidence 5' intron exon 3' 257 2 - 0.17 TCCAACCTAG^GAAGGCAGGG 337 2 - 0.67 TCCTTCCCAG^CCTGGGCATC 353 2 - 0.18 CATCCTTGAG^TTCCAAGGCC 360 0 - 0.18 GAGTTCCAAG^GCCTCATTCA	pos 5'->3' phase strand confidence 5' exon intron 3' 408 2 + 0.93 CTCACTCCAG^GTGAGTGACC H Ites, complement strand pos 5'->3' phase strand confidence 5' exon intron 3' pos 3'->5' 275 433 0 - 0.93 AGGGGGAGAA^GTAAGTATAT H Pos 5'->3' phase strand confidence 5' intron exon 3' 256 1 + 0.43 TTTAACTCAG^GTACTGTGTA 300 0 + 1.00 GTTGCTGCAG^ATCCGTGGGC H 315 0 + 0.19 TGGGCGTGAG^CGCTTCGAGA 454 1 + 0.53 GCCTTCCTAG^GTTGGAAAGC 605 2 + 0.16 GTATCCACAG^ATCGTAGAAACCCAAA esistes, complement strand pos 5'->3' phase strand confidence 5' intron exon 3' Acceptor splice strand acceptor sp	pos 5'->3' phase strand confidence 5' exon intron 3' 408 2 + 0.93 CTCACTCCAG^GTGAGTGACC H Ltes, complement strand pos 5'->3' phase strand confidence 5' exon intron 3' 433 0 - 0.93 AGGGGGAGAA^GTAAGTATAT H sites, direct strand pos 5'->3' phase strand confidence 5' intron exon 3' 275 433 Pos 5'->3' phase strand confidence 5' intron exon 3' 256 1 + 0.43 TTTAACTCAG^GTACTGTGTA 300 300 0 + 1.00 GTTGCTGCAG^ATCCGTGGGC H 315 0 + 0.19 TGGGGTGAGA^GCCTTCGAGA 324 0 + 0.07 GCGCTTCGAGAAGCCGTTCGAGA 454 1 + 0.53 GCCTTCCTAGAGATCCCAGA 454 605 2 + 0.16 GTATCCACAG^ATCCAAA Sites, complement strand Pos 5'->3' phase strand confidence 5' intron exon 3' 454 605 605 2 + 0.16 GTATCCACAG^AACATCCAAA Sites, complement strand Pos 5'->3' phase strand confidence 5' intron exon 3' 451 257 257 2 - 0.17 TCCAACCTAG^GAAGGCAGGG 371 337 337 337 2 - 0.67 TCCTTCCCAG^CCTTGGACATC 355 353 353 2 - 0.18 GAGTTCCAAG^CCTCATTCA 337 371	pos 5'->3' phase strand confidence 5' exon intron 3' 408 2 + 0.93 CTCACTCCAG^GTGAGTGACC H Les, complement strand pos 5'->3' phase strand confidence 5' exon intron 3' 408 2 Les, complement strand pos 5'->3' phase strand confidence 5' exon intron 3' 275 pos 5'->3' phase 3'->5' pos 5'->3' pha	pos 5'->3' phase strand confidence 5' exon intron 3' 408 2 + 0.93 CTCACTCCAG^GTGAGTGACC H pos 5'->3' phase strand 408 2 + 0.93 CTCACTCCAG^GTGAGTGACC H pos 5'->3' phase strand confidence 5' exon intron 3' 275 433 0 - 0.93 AGGGGGAGAA^GTAAGTATAT H pos 5'->3' phase strand confidence 5' exon intron 3' 275 433 0 - 0.93 AGGGGGAGAA^GTAAGTATAT H pos 5'->3' phase strand confidence 5' intron exon 3' 275 433 0 - 0.93 AGGGGGAGAA^GTAAGTATAT H pos 5'->3' phase strand confidence 5' intron exon 3' 256 1 + 0.43 TTTAACTCAG^GTACTGTGTA 300 0 + 1.00 GTTGCTGCAG^ATCCGTGGGC H 315 0 + 0.19 TGGGCGTGAG^ATCGTTGGAG 324 0 + 0.19 TGGGCGTGAG^ATGTTCCGAG 454 1 + 0.53 GCCTTCCAGAG^ATGTTCCGAG 454 1 + 0.54 1 + 0.55 GCCTTCCAGAG^ATGTTCCGAG 454 1 + 0.55 GCCTTCCAGAGAAGC 605 2 + 0.16 GTATCCACAG^AACATCCAAA pos 5'->3' phase strand confidence 5' intron exon 3' 451 257 2 - 0.57 CCAACCTAGAGAGGCAGGG 371 337 2 - 0.57 TCCAACCTAGAGAGGCAGGG 371 337 2 - 0.57 TCCAACCTTAGAGAGGCC 348 360 0 - 0.18 GAGTTCCAAGAGGCCC 348 360 0 0 - 0.18 GAGTTCCAAGAGGCCC 348 360 0 0 - 0.18 GAGTTCCAAGAGGCCCTATTCA 337 371 2 - 0.57 CCAACCTCATCACAGAGGCCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGAGCCC 348 360 0 0 - 0.18 GAGTTCCAAGAGGCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGAGCCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGAGCCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGACCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGAGCCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGAGCCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGACCCTCTATCACCCTATTCA 337 371 2 - 0.58 CATCCTTGAGACCCCTATTCA 337 371 2 - 0.58 CATCCTTGAGACCCCTATTCATTCAAGCCCC 348 360 0 0 - 0.58 GAGTTCCAAGAGCCCCTATTCATTCAAGACCCCCTATTCATTC	pos 5'->3' phase strand

Aparece un nuevo sitio *acceptor* en la secuencia mutante. Si se empleara este en vez del normal, se perderían 39 nt del exón.

Splice Site Prediction by Neural Network (NNSplice)

Donor site predictions for 85.53.81.31.8031.0:

Donor site predictions for 85.53.81.31.8053.0:

Start	End	Score	Exon Intron	Start	End	Score	Exon Intron
105	119	0.53	caacaga gt gagacc	105	119	0.53	caacaga gt gagacc
160	174	0.76	tgtatag gt acttga	160	174	0.76	tgtatag gt acttga
250	264	0.90	aactcag gt actgtg	250	264	0.90	aactcag gt actgtg
401	415	1.00	actccag gt gagtga	401	415	1.00	actccag gt gagtga

Acceptor site predictions for 85.53.81.31.8031.0:

Acceptor site predictions for 85.53.81.31.8053.0:

Start	End	Score	Intron	Exon	Start	End	Score	Intron	Exon
236	276	0.73	ttgaaccatctt	ttaactc ${f ag}$ gtactgtgtatatacttact	236	276	0.73	ttgaaccatctt	ttaactc ag gtactgtgtatatacttact
280	320	0.96	cccctcctctg	ttgctgc ${f ag}$ atccgtgggcgtgagcgctt	280	320	0.96	cccctcctctg	ttgctgc ag atccgtgggcgtgagcgctt
434	474	0.98	cctactcccctg	ccttcct ag gttggaaagccataggattc	434	474	0.98	cctactcccctg	ccttcct ag gttggaaagccataggattc
585	625	0.49	cttgcctcagtg	tatccac ag aacatccaaacccagggacg	585	625	0.49	cttgcctcagtg	tatccac ag aacatccaaacccagggacg

Spliceman

Point mutation	Wildtype (wt)	Mutation (mt)	L1 distance	Ranking (L1)
tgttc(c/t)gagag	ttccga	ttctga	31375	83%

CRYP-SKIP

Results for sequence wt

Exon length (bp)	107	EXSK		CR-E
PESS (<=-2.62) density	0.00	EAGR	0.5	ON L
NN 5'ss score density	0.05			
SF2/ASF score density	8.11	×	\mathcal{X}	
FAS-ESS (hex2) density	5.61	//	. / //	
EIE score density	364.52	- //	\ \	1
Probability of cryptic splice site activation (PcR-E)	0.33	0	1	1
>wt				1.0
•····•••••••••••••••••••••••••••••••••				0.5
cccctcctctgttgctgcagATCCGTGGGCGTGAGCGCTTC GATGCCCAGGCTGGGAAGGAGCCAGGGGGAGCAGGGCTCA	GAGATGTTCCGAG CTCCAGgtgagtg	AGCTGAATGAG (GCCTTGGAACTCAF	iG 0.0

Parece que hay un sitio críptico de *splicing* dentro del propio exón, pero el cambio de interés no lo toma en consideración, por lo que no debe considerar que tenga algún efecto en el *splicing*.

Human Splicing Finder

Туре	ţ	Interpretation	↑↓
 No significant impact on splicing signals. 		No significant impact on splicing signals.	

SVM-BPfinder

seq_id	agez	ss_dis	st bp_seq bp_scr	y_cont ppt_off	ppt_len ppt_scr	svm_scr				seq_id	agez	ss_dist	t bp_seq bp_scr	y_cont ppt_off	ppt_len ppt_scr	svm_scr			
wt	39	195	gcgtgagcg	0.0665569413265	0.505263157895	102	37	60	-5.3064548	mut	39	195	gcgtgagcg	0.0665569413265	0.505263157895	102	37	60	-5.3064548
wt	39	169	agctgaatg	0.654980928792	0.512195121951	76	37	60	-3.4280638	mut	39	176	ttctgagag	0.230625415505	0.502923976608	83	37	60	-4.0403017
wt	39	165	gaatgaggc	-1.14357913713	0.51875 72	37	60	-3.8769	9748	mut	39	169	agctgaatg	0.654980928792	0.512195121951	76	37	60	-3.4280638
wt	39	151	aactcaagg	-0.651503836258	0.520547945205	58	37	60	-2.7975469	mut	39	165	gaatgaggc	-1.14357913713	0.51875 72	37	60	-3.8769	748
wt	39	108	ggctcactc	1.92343889622	0.621359223301	15	37	60	0.9650518	mut	39	151	aactcaagg	-0.651503836258	0.520547945205	58	37	60	-2.7975469
wt	39	98	aggtgagtg	-0.804796826506	0.634408602151	5	37	60	0.53401634	mut	39	108	ggctcactc	1.92343889622	0.621359223301	15	37	60	0.9650518
wt	39	94	gagtgacct	2.14773755094	0.651685393258	1	37	60	1.9488463	mut	39	98	aggtgagtg	-0.804796826506	0.634408602151	5	37	60	0.53401634
wt	39	89	acctcagcc	0.174971006769	0.642857142857	2	31	55	1.0636861	mut	39	94	gagtgacct	2.14773755094	0.651685393258	1	37	60	1.9488463
wt	39	27	ttctcatcc	0.982359041544	0.590909090909	1	12	21	1.1095986	mut	39	89	acctcagcc	0.174971006769	0.642857142857	2	31	55	1.0636861
wt	39	16	ccttcatgg	0.0233227581103	0.363636363636	11	0	0	-0.1679278	mut	39	27	ttctcatcc	0.982359041544	0.590909090909	1	12	21	1.1095986
										mut	39	16	ccttcatgg	0.0233227581103	0.363636363636	11	0	0	-0.1679278

Aparece un nuevo BP, pero tiene una puntuación muy negativo, por lo que no lo tendremos en cuenta.

Variant Effect Predictor tool

ENST00000445888.6:c.1024C>T	17:7670685- 7670685	Α	3_prime_UTR_variant	TP53	ENSG00000141510 Transcript	ENST00000610623.4	protein_coding	7/8	966	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.1024C>T	<u>17:7670685-</u> <u>7670685</u>	Α	3_prime_UTR_variant	TP53	ENSG00000141510 Transcript	ENST00000618944.4	protein_coding	7/8	1039	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.1024C>T	<u>17:7670685-</u> <u>7670685</u>	Α	stop_gained	TP53	ENSG00000141510 Transcript	ENST00000619186.4	protein_coding	6/7	906	547	183	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.1024C>T	<u>17:7670685-</u> <u>7670685</u>	Α	stop_gained	TP53	ENSG00000141510 Transcript	ENST00000619485.4	protein_coding	10/11	1160	907	303	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.1024C>T	<u>17:7670685-</u> <u>7670685</u>	Α	stop_gained	TP53	ENSG00000141510 Transcript	ENST00000620739.4	protein_coding	10/11	1214	907	303	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.1024C>T	<u>17:7670685-</u> <u>7670685</u>	Α	3 prime_UTR_variant	TP53	ENSG00000141510 Transcript	ENST00000622645.4	protein_coding	11/12	1290	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.1024C>T	<u>17:7670685-</u> <u>7670685</u>	Α	stop gained, NMD transcript variant	TP53	ENSG00000141510 Transcript	ENST00000635293.1	nonsense_mediated_decay	10/12	1167	907	303	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487

ENST00000445888.6:c.102	7:7670685- 670685	Α	stop_gained	TP53	ENSG00000141510 Transcript	ENST00000269305.9	protein_coding	10/11	1166	1024	342	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	intron_variant	TP53	ENSG00000141510 Transcript	ENST00000359597.8	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	intron_variant	TP53	ENSG00000141510 Transcript	ENST00000413465.6	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	3_prime_UTR_variant	TP53	ENSG00000141510 Transcript	ENST00000420246.6	protein_coding	11/12	1290	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	stop_gained	TP53	ENSG00000141510 Transcript	ENST00000445888.6	protein_coding	10/11	1160	1024	342	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	3_prime_UTR_variant	TP53	ENSG00000141510 Transcript	ENST00000455263.6	protein_coding	11/12	1217	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	downstream_gene_variant	TP53	ENSG00000141510 Transcript	ENST00000503591.1	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	3_prime_UTR_variant	TP53	ENSG00000141510 Transcript	ENST00000504290.5	protein_coding	7/8	966	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	stop_gained	TP53	ENSG00000141510 Transcript	ENST00000504937.5	protein_coding	6/7	906	628	210	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	downstream_gene_variant	TP53	ENSG00000141510 Transcript	ENST00000505014.5	retained_intron	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	downstream_gene_variant	TP53	ENSG00000141510 Transcript	ENST00000508793.5	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	downstream_gene_variant	TP53	ENSG00000141510 Transcript	ENST00000509690.5	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	3_prime_UTR_variant	TP53	ENSG00000141510 Transcript	ENST00000510385.5	protein_coding	7/8	1039	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	downstream_gene_variant	TP53	ENSG00000141510 Transcript	ENST00000514944.5	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	downstream_gene_variant	TP53	ENSG00000141510 Transcript	ENST00000574684.1	processed_transcript	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	intron_variant	TP53	ENSG00000141510 Transcript	ENST00000576024.1	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	downstream_gene_variant	TP53	ENSG00000141510 Transcript	ENST00000604348.5	protein_coding	-	-	-	-	-	-	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	stop_gained	TP53	ENSG00000141510 Transcript	ENST00000610292.4	protein_coding	9/10	1274	907	303	R/*	CGA/TGA	rs730882029, CM004908, COSV52665487
ENST00000445888.6:c.102	7:7670685- 670685	Α	3 prime UTR variant	TP53	ENSG00000141510 Transcript	ENST00000610538.4	protein_coding	11/12	1217	-	-	-	-	rs730882029, CM004908, COSV52665487

ESEfinder

Para los sitios de *splicing*, no aparece ningún resultado con la posición de interés que tenga puntuaciones positivas, ni tampoco los equivalentes en la secuencia mutante, por lo que no afecta a los sitios de *splicing*.

325 ATGTT (-383)	CC -7.04382	325 (-383) ATGTTCC -5.09141	325 ATGTTCCG 0.10981	325 ATGTTCC -1.42468
326	CG -4.87402	326	326	326
(-382) TGTTC		(-382) TGTTCCG -3.21801	(-382) TGTTCCGA 0.83234	(-382) TGTTCCG 0.02835
327	GA 0.58569	327	327	327
(-381) GTTCC		(-381) GTTCCGA -0.40321	(-381) GTTCCGAG 3.29663	GTTCCGA -1.65798
328	AG -2.38611	328	328	328
(-380) TTCCG		(-380) TTCCGAG -0.96415	(-380) TTCCGAGA -1.19004	TTCCGAG 0.49441
329 TCCGA (-379)	GA -3.51916	329 (-379) TCCGAGA -2.01825	329 TCCGAGAG -2.74304	329 TCCGAGA -0.17721
330	AG 0.70877	330	330	330
(-378)		(-378) CCGAGAG 1.91666	(-378) CCGAGAGC -9.37330	(-378) CCGAGAG -1.33250
331	GC -3.80709	331	331	331
(-377)		(-377) CGAGAGC -1.12435	(-377) CGAGAGCT -3.57801	(-377) CGAGAGC 2.57920
		_		
325	CT -5.19043	325	325	325
ATGTT		(-383) ATGTTCT -3.89471	(-383) ATGTTCTG 0.75425	ATGTTCT -2.96378
326	TG -4.41632	326	326	326
(-382) TGTTC		(-382) TGTTCTG -3.11401	(-382) TGTTCTGA -0.89667	TGTTCTG -1.68857
327 GTTCT (-381)	GA -1.93803	327 (-381) GTTCTGA -2.21500	327 (-381) GTTCTGAG 2.53739	327 (-381) GTTCTGA -0.16673
328	AG -3.84343	328	328	328
(-380)		(-380) TTCTGAG -2.21891	(-380) TTCTGAGA -1.42088	(-380) TTCTGAG -1.89023
329	GA -5.82917	329	329	329
TCTGA		(-379) TCTGAGA -4.08711	TCTGAGAG -3.31197	TCTGAGA 1.17107
330	AG 1.31170	330	330	330
(-378)		(-378) CTGAGAG 2.16544	(-378) CTGAGAGC -7.98489	(-378) CTGAGAG -1.64456
331	GC -6.75758	331	331	331
(-377)		(-377) TGAGAGC -4.22940	(-377) TGAGAGCT -3.59914	(-377) TGAGAGC 2.93590

En cuanto a las ESE, los resultados no cambian mucho entre secuencias.

EX-SKIP

Seq	PESS (count)	FAS-ESS hex2 (count)	FAS-ESS hex3 (count)	IIE (count)	IIE (sum)	NI-ESS trusted (count)	NI-ESS all (sum)	PESE (count)	RESCUE -ESE (count)	EIE (count)	EIE (sum)	NI-ESE trusted (count)	NI-ESE all (sum)	ESS (total)	ESE (total)	ESS/ESE (ratio)
wt	0	6	5	21	245.4405	6	-8.2198	4	7	35	376.7962	38	53.3183	38	84	0.45
mut	0	5	5	23	280.0633	5	-8.0016	4	7	33	354.2475	39	54.5823	38	83	0.46

Allele mut has a higher chance of exon skipping than allele wt.

HOT-SKIP

117	31	С	GATGTTCCGAGAGCT	TGTTCCGAGAG	0	2	1	1	7.4238	2	-1.9999	0	0	2	22.5487	1	1.8161	6	3	2.00
118	31	Α	GATGTTCAGAGAGCT	TGTTCAGAGAG	0	0	0	1	8.6203	1	-0.9999	1	1	2	23.1995	3	4.1370	2	7	0.29
119	31	T	GATGTTCTGAGAGCT	TGTTCTGAGAG	0	1	1	3	42.0465	1	-1.7817	0	0	0	0.0000	2	3.0801	6	2	3.00
120	31	G	GATGTTCGGAGAGCT	TGTTCGGAGAG	0	0	0	0	0.0000	0	-1.5335	0	0	3	23.8134	2	2.7246	0	5	0.00