

Ejemplo comparación de resultados predictores in silico

Cambio de estudio KCNJ11 c.1054G>A (chr11:17387038 G/A, CM1111083 o NM_000525.4: c.1054G>A)

Exón 1 e intrones adyacentes:

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aaagcggcgggggcgctcgggaggggtggagttaggacataggggcgccactggaggag
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TGAGGCTGGTATTAAAGAAGTGAAGTGGA CCGAGGTGGAGTAAGGAAGAGTCTGGTGGG
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CACCGAGAGGACTCTGCAGTGAGGCCTAGGCCACGTCCGAGGGGTGCCTCCGATGGGGG
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caggtggccagccctgtcagcctgtctggccaactcagcctttgggcactcaccaggct
ttgcagccctgggctctgtctctaattcccagggaacctgctggaaggctggagtgeccagg

Cambio de estudio

NetGene2

Donor splice sites, direct strand										Donor splice sites, direct strand									
-----										-----									
pos 5'->3'		phase	strand	confidence	5'	exon	intron	3'		pos 5'->3'		phase	strand	confidence	5'	exon	intron	3'	
532		-	+	0.00		GGACCATGTG	^GTATGTAGCN			532		-	+	0.00		GGACCATGTG	^GTATGTAGCN		
Donor splice sites, complement strand										Donor splice sites, complement strand									
-----										-----									
pos 3'->5'	pos 5'->3'	phase	strand	confidence	5'	exon	intron	3'		pos 3'->5'	pos 5'->3'	phase	strand	confidence	5'	exon	intron	3'	
220	321	0	-	0.41		CATCGGCCAG	^GTAGGAGGTG			220	321	0	-	0.41		CATCGGCCAG	^GTAGGAGGTG		
Acceptor splice sites, direct strand										Acceptor splice sites, direct strand									
-----										-----									
No acceptor site predictions above threshold.										No acceptor site predictions above threshold.									
Acceptor splice sites, complement strand										Acceptor splice sites, complement strand									
-----										-----									
pos 3'->5'	pos 5'->3'	phase	strand	confidence	5'	intron	exon	3'		pos 3'->5'	pos 5'->3'	phase	strand	confidence	5'	intron	exon	3'	
295	246	1	-	0.07		CTTGGAGTAG	^TCCACAGAGT			295	246	1	-	0.07		CTTGGAGTAG	^TCCACAGAGT		
288	253	2	-	0.18		TAGTCCACAG	^AGTAACGTCC			288	253	2	-	0.18		TAGTCCACAG	^AGTAACGTCC		
286	255	1	-	0.18		GTCCACAGAG	^TAACGTCCGT			286	255	1	-	0.18		GTCCACAGAG	^TAACGTCCGT		
267	274	2	-	0.34		TCCTCCTCAG	^CTACAATGGG			267	274	2	-	0.34		TCCTCCTCAG	^CTACAATGGG		
250	291	1	-	0.18		GGGCACAAAG	^CGCTGGCCCC			250	291	1	-	0.18		GGGCACAAAG	^CGCTGGCCCC		
236	305	0	-	0.18		GGCCCCACAG	^GATCTCATCG			236	305	0	-	0.18		GGCCCCACAG	^GATCTCATCG		

Splice Site Prediction by Neural Network (NNSplice)

Donor site predictions for 85.53.81.31.12339.0 :

Start	End	Score	Exon	Intron
525	539	0.86	ccatgtg	g tatgtag

Donor site predictions for 85.53.81.31.12274.0 :

Start	End	Score	Exon	Intron
525	539	0.86	ccatgtg	g tatgtag

Acceptor site predictions for 85.53.81.31.12339.0 :

Start	End	Score	Intron	Exon
246	286	0.59	agcgctttgtgcccattgt a g	ctgaggaggacggacgttac
378	418	0.56	aagctctgaccctcgcc a g	cccgcgggccctgcgcaag
447	487	0.81	ccaagttcagcatctctcc a g	attccctgtcctgagccatg

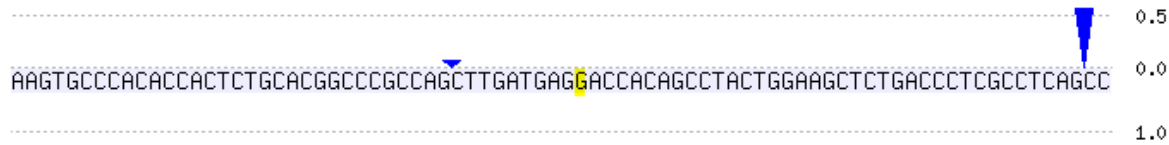
Acceptor site predictions for 85.53.81.31.12274.0 :

Start	End	Score	Intron	Exon
246	286	0.59	agcgctttgtgcccattgt a g	ctgaggaggacggacgttac
378	418	0.56	aagctctgaccctcgcc a g	cccgcgggccctgcgcaag
447	487	0.81	ccaagttcagcatctctcc a g	attccctgtcctgagccatg

Spliceman

Point mutation	Wildtype (wt)	Mutation (mt)	L1 distance	Ranking (L1)
atgag(g/a)accac	tgagga	tgagaa	24919	42%

CRYP-SKIP



Human Splicing Finder

Type	↑↓	Interpretation	↑↓
	No significant impact on splicing signals.	No significant impact on splicing signals.	

seq_id	agez	ss_dist	bp_seq	bp_scr	y_cont	ppt_off	ppt_len	ppt_scr	svm_scr	seq_id	agez	ss_dist	bp_seq	bp_scr	y_cont	ppt_off	ppt_len	ppt_scr	svm_scr
wt	13	407	cgctgatca	2.37514887589	0.54228857214	1	9	13	1.5647118	mut	13	407	cgctgatca	2.37514887589	0.54228857214	1	9	13	1.5647118
wt	13	404	tgcgtcatct	-1.73967262935	0.541353383459	26	10	14	-1.6198794	mut	13	404	tgcgtcatct	-1.73967262935	0.541353383459	26	10	14	-1.6198794
wt	13	392	atgtcattg	-1.95880536641	0.537467700258	14	10	14	-0.94735534	mut	13	392	atgtcattg	-1.95880536641	0.537467700258	14	10	14	-0.94735534
wt	13	388	cattgatgc	1.74739133819	0.537859007833	10	10	14	0.75711549	mut	13	388	cattgatgc	1.74739133819	0.537859007833	10	10	14	0.75711549
wt	13	323	agatcatcg	-2.26800510867	0.534591194969	1	10	16	-0.22784085	mut	13	323	agatcatcg	-2.26800510867	0.534591194969	1	10	16	-0.22784085
wt	13	317	tcgtcatcc	-1.19884322252	0.532051282051	31	9	11	-1.7555626	mut	13	317	tcgtcatcc	-1.19884322252	0.532051282051	31	9	11	-1.7555626
wt	13	284	gcataccaca	-0.673617040347	0.548387096774	10	16	23	-0.10358086	mut	13	284	gcataccaca	-0.673617040347	0.548387096774	10	16	23	-0.10358086
wt	13	250	cgatgagat	-1.27372607449	0.538775510204	17	15	20	-0.81269197	mut	13	250	cgatgagat	-1.27372607449	0.538775510204	17	15	20	-0.81269197
wt	13	211	agctgagga	-0.953646914644	0.538834951456	13	9	15	-0.48073257	mut	13	211	agctgagga	-0.953646914644	0.538834951456	13	9	15	-0.48073257
wt	13	196	acgttactc	-1.675598472	0.560209424084	47	9	12	-2.9365979	mut	13	196	acgttactc	-1.675598472	0.560209424084	47	9	12	-2.9365979
wt	13	161	ccgtcaaaag	-1.73500560039	0.557692307692	12	9	12	-0.74523028	mut	13	161	ccgtcaaaag	-1.73500560039	0.557692307692	12	9	12	-0.74523028
wt	13	124	gcttgatga	-0.104270961811	0.554621848739	24	16	24	-0.75550196	mut	13	124	gcttgatga	-0.104270961811	0.554621848739	24	16	24	-0.75550196
wt	13	121	tgatgagga	-2.43834989987	0.560344827586	21	16	24	-1.4776609	mut	13	121	tgatgagga	-2.43834989987	0.560344827586	21	16	24	-1.4776609
wt	13	95	ctctgaccc	3.92167998102	0.588888888889	1	10	18	2.2318817	mut	13	95	ctctgaccc	3.92167998102	0.588888888889	1	10	18	2.2318817
wt	13	84	gccttcagcc	-0.909174006341	0.556962025316	60	9	17	-2.701885	mut	13	84	gccttcagcc	-0.909174006341	0.556962025316	60	9	17	-2.701885
wt	13	26	agttcacga	-0.38458682825	0.714285714286	2	9	17	0.12211276	mut	13	26	agttcacga	-0.38458682825	0.714285714286	2	9	17	0.12211276

ENST00000339994.4:c.1054G>A	11:17387038-17387038	T	missense_variant	KCNJ11	ENSG00000187486	Transcript	ENST00000339994.4	protein_coding	1/1	1622	1054	352	D/N	GAC/AAC	rs1242156391, CM1111083, COSV60594722
ENST00000339994.4:c.1054G>A	11:17387038-17387038	T	upstream_gene_variant	KCNJ11	ENSG00000187486	Transcript	ENST00000526747.1	processed_transcript	-	-	-	-	-	-	rs1242156391, CM1111083, COSV60594722
ENST00000339994.4:c.1054G>A	11:17387038-17387038	T	downstream_gene_variant	KCNJ11	ENSG00000187486	Transcript	ENST00000526912.1	protein_coding	-	-	-	-	-	-	rs1242156391, CM1111083, COSV60594722
ENST00000339994.4:c.1054G>A	11:17387038-17387038	T	missense_variant	KCNJ11	ENSG00000187486	Transcript	ENST00000528731.1	protein_coding	2/2	967	793	265	D/N	GAC/AAC	rs1242156391, CM1111083, COSV60594722
ENST00000339994.4:c.1054G>A	11:17387038-17387038	T	downstream_gene_variant	KCNJ11	ENSG00000187486	Transcript	ENST00000528992.1	protein_coding	-	-	-	-	-	-	rs1242156391, CM1111083, COSV60594722

Las puntuaciones para las matrices 5'SS y 3'SS salen todas con puntuaciones negativas.

En los sitios ESE, el único resultado donde descienden las puntuaciones es en el 1920, que pasan a ser negativas, por lo que podría estar perdiéndose un sitio ESE.

1916 (-1486)	GATGAGG	-3.45732	1916 (-1486)	GATGAGG	-3.63559	1916 (-1486)	GATGAGGA	-1.33389	1916 (-1486)	GATGAGG	-1.21825
1917 (-1485)	ATGAGGA	2.11103	1917 (-1485)	ATGAGGA	1.24471	1917 (-1485)	ATGAGGAC	-6.05007	1917 (-1485)	ATGAGGA	-4.15139
1918 (-1484)	TGAGGAC	-7.41570	1918 (-1484)	TGAGGAC	-4.30267	1918 (-1484)	TGAGGACC	-4.38589	1918 (-1484)	TGAGGAC	-0.18649
1919 (-1483)	GAGGACC	-5.45113	1919 (-1483)	GAGGACC	-4.58432	1919 (-1483)	GAGGACCA	-0.48166	1919 (-1483)	GAGGACC	-3.92803
1920 (-1482)	AGGACCA	0.81271	1920 (-1482)	AGGACCA	0.53481	1920 (-1482)	AGGACCAC	-3.58941	1920 (-1482)	AGGACCA	-4.14892
1921 (-1481)	GGACCAC	-3.52338	1921 (-1481)	GGACCAC	-2.57394	1921 (-1481)	GGACCACA	3.06984	1921 (-1481)	GGACCAC	-0.14892
1922 (-1480)	GACCACA	-1.08323	1922 (-1480)	GACCACA	-1.46471	1922 (-1480)	GACCACAG	4.59528	1922 (-1480)	GACCACA	-3.52956

1916 (-1486)	GATGAGA	-2.73054	1916 (-1486)	GATGAGA	-3.20022	1916 (-1486)	GATGAGAA	-0.03700	1916 (-1486)	GATGAGA	-3.59998
1917 (-1485)	ATGAGAA	-0.46670	1917 (-1485)	ATGAGAA	-0.53494	1917 (-1485)	ATGAGAAC	-6.41156	1917 (-1485)	ATGAGAA	-4.71922
1918 (-1484)	TGAGAAC	-9.33532	1918 (-1484)	TGAGAAC	-6.00905	1918 (-1484)	TGAGAACC	-3.96063	1918 (-1484)	TGAGAAC	2.36807
1919 (-1483)	GAGAACC	-2.54255	1919 (-1483)	GAGAACC	-2.86457	1919 (-1483)	GAGAACCA	-0.48166	1919 (-1483)	GAGAACC	-3.77273
1920 (-1482)	AGAACCA	-1.25107	1920 (-1482)	AGAACCA	-0.84894	1920 (-1482)	AGAACCAC	-2.29252	1920 (-1482)	AGAACCA	-1.54012
1921 (-1481)	GAACCAC	-3.07616	1921 (-1481)	GAACCAC	-2.87216	1921 (-1481)	GAACCACA	2.70899	1921 (-1481)	GAACCAC	-1.59409
1922 (-1480)	AACCACA	-2.01111	1922 (-1480)	AACCACA	-1.70226	1922 (-1480)	AACCACAG	2.84321	1922 (-1480)	AACCACA	-2.07484

EX-SKIP

No se puede analizar porque la secuencia es demasiado larga.

HOT-SKIP

>wt

aaagcggcgggggcgctccgggaggggtggagtaggacatagggggcgacacctggaggagagacggggcgggggtggcca
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