**Ejemplo comparación de resultados predictores in sillico**
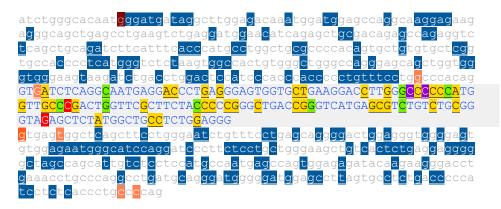
Cambio de estudio DDR1 c.418-287G>T (chr 6:30890686 G/T,  COSV61323560 o NM_001202521.1: c.418-287G>T)

Exón 6 e intrones adyacentes:

atctgggcacaatggggatgatagggcttggagacaaatggatggagccaggcaaggagaag
agggcagctgagcctgaagtctgaggatggaacatcagagctgcgacagagccagaggtc
tcagctgcagatcttcatttcacccatgcctggctgcgccccacagtgctgtgtgctcgg
tgccaccctcatgggtctcttaagtggcactgtgggctgggccaggagcagctggtgg
gtgggaagtaagatctgacctggactccatccacccaccctgtttcctggcccacag
GTGATCTCAGGCAATGAGGACCCTGAGGGAGTGGTGCTGAAGGACCTTGGGCCCCCCATG
GTTGCCCGACTGGTTCGCTTCTACCCCCGGGCTGACCGGGTCATGAGCGTCTGTCTGCGG
GTAGAGCTCTATGGCTGCCTCTGGAGGG
gtgagtggctcagcttcctgggaatctgtttcctgagcaggggactggagggtgggga
gtggagaatgggcatccaggatccctttctcctgctggaagctgtcactctgaggaggg
gctagccagcattgtctcctccatgccaatgagccagtggagagatacaagaagggacct
gaaacctgcccaggcctgatgcagggatggggatggagccttagtgcctctctgacccca
tcctctcaccctgcccag

El cambio se encuentra en la primera línea del intrón 5 (la **g** en color granate).

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'
          237         0      +       0.60      GGAGCAGCTG^GTGGGTGGGA
          248         2      +       0.37      TGGGTGGGAA^GTAAGATCTG
          449         1      +       1.00      CTCTGGAGGG^GTGAGTGGCT H

Donor splice sites, complement strand
-------------------------------------
  pos 3'->5'  pos 5'->3'  phase strand  confidence  5'      exon intron    3'
     697         11        -      -       0.00      CTGGGGCAGG^GTGAGAGGAT
     396         312       0      -       0.51      TCATGACCCG^GTCAGCCCGG
     279         429       2      -       0.32      GAAACAGGGG^GTGGGTGGGA

Acceptor splice sites, direct strand
------------------------------------
        pos 5'->3'  phase strand  confidence  5'      intron exon    3'
          300         0      +       1.00      TGGCCCACAG^GTGATCTCAG H
          310         1      +       0.42      GTGATCTCAG^GCAATGAGGA
          318         0      +       0.20      AGGCAATGAG^GACCCTGAGG
          327         0      +       0.18      GGACCCTGAG^GGAGTGGTGC

Acceptor splice sites, complement strand
----------------------------------------
  pos 3'->5'  pos 5'->3'  phase strand  confidence  5'      intron exon    3'
     493         215       0      -       0.20      CACCCTCCAG^TCCCCTGCTC
     481         227       0      -       0.52      CCCTGCTCAG^GAAACAGATT
     474         234       1      -       0.19      CAGGAAACAG^ATTCCCAGGA
     466         242       0      -       0.25      AGATTCCCAG^GAAGCTGAGC
     462         246       1      -       0.20      TCCCAGGAAG^CTGAGCCACT
     457         251       0      -       0.20      GGAAGCTGAG^CCACTCACCC
     441         267       1      -       0.85      ACCCCTCCAG^AGGCAGCCAT
     439         269       0      -       0.20      CCCTCCAGAG^GCAGCCATAG
     435         273       1      -       0.19      CCAGAGGCAG^CCATAGAGCT
     429         279       1      -       0.19      GCAGCCATAG^AGCTCTACCC
     427         281       0      -       0.18      AGCCATAGAG^CTCTACCCGC
     415         293       0      -       0.14      CTACCCGCAG^ACAGACGCTC
```

**Splice Site Prediction by Neural Network (NNSplice)**

## Donor site predictions for 10.42.2.148.574662.0 :

| Start | End | Score | Exon | Intron |
|-------|-----|-------|------|--------|
| 294 | 308 | 0.46 | cccacag**gt**gatctc | |
| 442 | 456 | 0.97 | tggaggg**gt**gagtgg | |

## Donor site predictions for 10.42.3.123.574674.0 :

| Start | End | Score | Exon | Intron |
|-------|-----|-------|------|--------|
| 294 | 308 | 0.46 | cccacag**gt**gatctc | |
| 442 | 456 | 0.97 | tggaggg**gt**gagtgg | |

## Acceptor site predictions for 10.42.2.148.574662.0 :

| Start | End | Score | Intron | Exon |
|-------|-----|-------|--------|------|
| 280 | 320 | 0.96 | cccctgtttcctggcccac**ag**gtgatctcaggcaatgagga | |
| 468 | 508 | 0.51 | gggaatctgtttcctgagc**ag**gggactggagggtggggagt | |

## Acceptor site predictions for 10.42.3.123.574674.0 :

| Start | End | Score | Intron | Exon |
|-------|-----|-------|--------|------|
| 280 | 320 | 0.96 | cccctgtttcctggcccac**ag**gtgatctcaggcaatgagga | |
| 468 | 508 | 0.51 | gggaatctgtttcctgagc**ag**gggactggagggtggggagt | |

**Spliceman**

| Point mutation | Wildtype (wt) | Mutation (mt) | L1 distance | Ranking (L1) |
|----------------|---------------|---------------|-------------|--------------|
| acaat(g/t)ggatg | tgggat | ttggat | 28131 | 62% |

**Human Splicing Finder**

| Alteration of auxiliary sequences | Significant alteration of ESE / ESS motifs ratio (3) | |
|---|---|---|
| Algorithm/Matix | position | sequence |
| EIE (ESE Site Broken) | chr6:30890681 | ACAATG |
| ESE_SRp40 (ESE Site Broken) | chr6:30890681 | ACAATGG |
| Sironi_motif2 (ESS Site Broken) | chr6:30890682 | CAATGGG |
| IIE (New ESS Site) | chr6:30890683 | AATTGG |
| Fas ESS (ESS Site Broken) | chr6:30890683 | AATGGG |
| Sironi_motif2 (ESS Site Broken) | chr6:30890683 | AATGGGA |
| RESCUE ESE (New ESE Site) | chr6:30890684 | ATTGGA |
| IIE (New ESS Site) | chr6:30890684 | ATTGGA |
| RESCUE ESE (New ESE Site) | chr6:30890685 | TTGGAT |
| EIE (New ESE Site) | chr6:30890686 | TGGATG |
| Fas ESS (ESS Site Broken) | chr6:30890686 | GGGATG |

**SVM-BPfinder**

```
seq_id  agez    ss_dist bp_seq      bp_scr          y_cont          ppt_off ppt_len ppt_scr svm_scr
wt      14      67      ggatgatag   -1.01687700492  0.306451612903  62      0       0       -3.8218986
wt      14      16      agctgagcc   0.905318440489  0.545454545455  11      0       0       0.23613714
mut     15      68      ggatgatag   -1.01687700492  0.301587301587  63      0       0       -3.8867679
mut     15      17      agctgagcc   0.905318440489  0.5             12      0       0       0.15815833
```

**Variant Effect Predictor tool**

| | | | | | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000324771.12 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000376567.6 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000376568.7 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000376569.7 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000376570.8 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000376575.7 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000396342.6 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | downstream_gene_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000412274.6 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | upstream_gene_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000417521.5 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000418800.6 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000421124.6 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000424544.2 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000428153.6 | protein_coding | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant, non_coding_transcript_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000431373.1 | retained_intron | - | - | COSV61323560 | 1 |
| ENST00000376569.7:c.418-287G>T | 6:30890686-30890686 | T | intron_variant | DDR1 | ENSG00000204580 | Transcript | ENST00000437124.6 | protein_coding | - | - | COSV61323560 | 1 |

**ESEfinder**

No se obtiene ningún resultado positive con la posición de interés ni en la secuencia mutante ni en la WT.