Figure S4

BMPR2 c.853-2A>G

▾ Sequences

Reference sequence

BMPR2 Gene > ENST00000374580 Transcript > Exon number: **7** (115 bp) + **100** intronic nucleotides at exon ends

```
  1 agcctatttg caaattcttt ataaggatgc taatttactc ttcatgttaa agtgagttaa ttctacctttt tttttttttc gcatttttttc ctctatatag
101 GGATCTTTAT GCAAGTATTT AAGTCTCCAC ACAAGTGACT GGGTAAGCTC TTGCCGTCTT GCTCATTCTG TTACTAGAGG ACTGGCTTAT CTTCACACAG
201 AATTACCACG AGGAGgtaag atagtcaata gatgaaattg acactcatgt gggttcaaaa ttcacaacag gaaattttaa ttatatcttc aaagtaaaag
301 tataatttat attat
          Total sequence length: 315 nucleotides
```

Mutant sequence

```
  1 agcctatttg caaattcttt ataaggatgc taatttactc ttcatgttaa agtgagttaa ttctacctttt tttttttttc gcatttttttc ctctatatgg
101 GGATCTTTAT GCAAGTATTT AAGTCTCCAC ACAAGTGACT GGGTAAGCTC TTGCCGTCTT GCTCATTCTG TTACTAGAGG ACTGGCTTAT CTTCACACAG
201 AATTACCACG AGGAGgtaag atagtcaata gatgaaattg acactcatgt gggttcaaaa ttcacaacag gaaattttaa ttatatcttc aaagtaaaag
301 tataatttat attat
          Total sequence length: 315 nucleotides
```

The underlined sequences are analyzed by HSF.

▾ Interpreted Data

This table shows only relevant results related to the mutation position and context.
The mutation occurs in the late intronic positions, the following table show results of acceptor splice sites that could be affected by the mutation

Predicted signal	Prediction algorithm	cDNA Position	Interpretation
Broken WT Acceptor Site	1 - HSF Matrices	ttcgcatttttttcctctatatagGGATC	Alteration of the WT acceptor site, most probably affecting splicing.
	2 - MaxEnt		