

Figure S3

Sequences

Reference sequence

BMPR2 Gene > ENST00000374580 Transcript > Exon number: 10 (137 bp) + 100 intronic nucleotides at exon ends

```

1 caattttttt tgcttacttg gtatcagaaa taccocctggt attctatcat ttatgatagt tagaaatttt attctgtcat tcttttctac a---aatcca
101 cagGGGAATC CGTACCAGAG TACCAGATGG CTTTTCAGAC AGAGGTTGGA AACCATCCCA CTTTGTAGGA TATGCAGGTT CTCGTGTCTA GGGAAAAACA
201 GAGACCCAAG TTCCAGAAG CCTGGAAAGA AAATAGCCTG gtaagaaaaa actaagttat taaagagagg acttatgact gaatatgttt taatgttgtt
301 tgaaacaaaa atagactggt aataacgtta atagtttttg
    
```

Total sequence length: 340 nucleotides

Mutant sequence

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1 caattttttt tgcttacttg gtatcagaaa taccocctggt attctatcat ttatgatagt tagaaatttt attctgtcat tcttttctac aggggaatcca
101 cagGGGAATC CGTACCAGAG TACCAGATGG CTTTTCAGAC AGAGGTTGGA AACCATCCCA CTTTGTAGGA TATGCAGGTT CTCGTGTCTA GGGAAAAACA
201 GAGACCCAAG TTCCAGAAG CCTGGAAAGA AAATAGCCTG gtaagaaaaa actaagttat taaagagagg acttatgact gaatatgttt taatgttgtt
301 tgaaacaaaa atagactggt aataacgtta atagtttttg
    
```

Total sequence length: 340 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
New Acceptor Site	1 - HSF Matrices		<p>Activation of an intronic cryptic acceptor site. Potential alteration of splicing.</p>
	2 - MaxEnt		
New Acceptor Site	1 - HSF Matrices		<p>Activation of an intronic cryptic acceptor site. Potential alteration of splicing.</p>
Broken WT Acceptor Site	1 - MaxEnt		<p>Alteration of the WT acceptor site, most probably affecting splicing.</p>