Supplemental Figure S1. In-silico analysis for predicted splice site

http://www.fruitfly.org/seq_tools/splice.html

Splice site predictions for 1 sequence with donor score cutoff 0.40, acceptor score cutoff 0.40 (exon/intron boundary shown in larger font):

Donor site predictions for 10.42.3.122.160411.0 :

Start	End	Score	Exon Intron
7	21	0.77	ccaaatagtatgtcc
61	75	0.69	gaatgaggtgtgcag
251	265	0.98	cctccaggtgagagg
427	441	0.99	cacagaagtaagtac
490	504	0.66	gcttcaggtgacacc

Splice site predictions for 1 sequence with donor score cutoff 0.40, acceptor score cutoff 0.40 (exon/intron boundary shown in larger font):

Donor site predictions for 10.42.0.144.160423.0 :

Start	End	Score	Exon Intron
7	21	0.77	ccaaatagtatgtcc
61	75	0.69	gaatgaggtgtgcag
251	265	0.98	cctccaggtgagagg
486	500	0.66	gcttcaggtgacacc

In comparison with the wild type sequence, the normal splicing donor site (red rectangle) is disappeared in the variant sequence.

Wild type

Supplemental Figure S2. In-silico analysis for predicted splice site

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http://www.cbs.dtu.dk/services/NetGene2/
      The sequence: sequence1 has the following composition:
Wild type
      Length: 780 nucleotides.
      26.5% A, 20.1% C, 29.9% G, 23.5% T, 0.0% X, 50.0% G+C
      Donor splice sites, direct strand
                   pos 5'->3'
                             phase strand confidence 5'
                                                                      3'
                                                          exon intron
                                                     TGTCCTCCAG^GTGAGA
                      258
                                           0.49
                                           0.93
                      434
                               n
                                                     AGCCACAGAA^GTAAG
      Variant
      The sequence: sequence1 has the following composition:
      Length: 776 nucleotides.
      26.4% A, 20.2% C, 29.9% G, 23.5% T, 0.0% X, 50.1% G+C
      Donor splice sites, direct strand
                  pos 5'->3' phase strand confidence 5'
                                                                      3'
                                                         exon intron
                                                    TGTCCTCCAG^GTGAGAGGAC
                      258
                              0
                                           0.49
                                    +
```

In comparison with the wild type sequence, the normal splicing donor site (red rectangle) is disappeared in the variant sequence.