## Intron 33 cryptic splice sites:

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45626782 GAATGAAAGTTCGTATTTCGTTGCTCATACATGGTCCCTGGACCAGCAGCATCAGCATCA 45626723
45626722 GCATCAGTATCACCCCTGGGAAGGGGTGAGAGATGCAGAGTCTCGGGCTCCGTGCCAGGC 45626663
45626662 CTCCTCAATTGCAAGGCACATAACAAGATGCCAGGTGACATTCACAGGCTCATTAAAGTG 45626603
45626602 AGGAATGCTGAGCTTCTGCAGTCTGTCTGGGATCACCCTGGTCCCACTGTCAAAGTTCAT 45626543
45626542 CAGGGTTGGCTTTTACTGTCCCTGCGATCATCTGCAGTTTCTTATCCAGGCATGTGGCTG 45626483
45626482 AATCAGGGCTGGCCTGCCAGCCTCCCACCCCCATGTGCTCCCTGGGGCCACCAGGCAGCT 45626423
45626422 GTGAGAACCACTGCTCAGAATTCCTTCCCCAGCTGGGCTCCTAGGACTGAAAGACCTTCC 45626363
45626362 ACACTAGCTGCCAAGAGGGCAGGGTTTTCTCTGTGAAAAGAGGAGCCCCCGCCACCCCTC 45626303
45626302 CGAGTCCCTCTTGGCTGCTCAGCAAGGGCCAGGAGGGGAGTCTGTGTGACCTGTGCAGCTT 45626243
45626242 GCTCAGGTTTGTGCCCAGTGCTGCTTTGCCAGTCGTACCAGCTCCCTCCCCTCCAGTCTT 45626183
45626182 GTGCCCCATCTGCCACAGGCCACTGCAACAGGCCACCTCCCTGCTCTGTACCAACCCAGT 45626123
45626122 CCAGCATCCCCCTGCAGAGCCAGGAAGAGTCTTAGAGGAGAGAGTCCTGTGGGAAAGAGG 45626063
45626062 AGAAATCTTTGGTGGTCTGATCCTCTGGACCCTGAGCCAGCAGCTCCCTTCTTTATGGCA 45626003
45626002 CCTTAGTGAAATGGGATGATGACACTGACCTGCCTGCCCCATGATGTGAAGTGTGTCAAA 45625943
45625942 GGCGAAATGCTGTGTTAAAACTGCAAAACCCTTCTCAAATGTGAGGCTTTCAAAGGAGTG 45625883
45625882 GTAGCTTTTGCAGGAATATTTGAAGTTCAAATAAGAGGCAAGGCAAGCATTATTTTAGAG 45625823
45625822 AAACTTATGTAAGACTGAAGTCCGACTCTGCTGAGCACTGTGCCTAGGGCCGAGGGACCC 45625763
45625762 AGGAAGAACAGATTGTGACATCTGCCTTAAACAGATTTGCTCCAGCTGTTTGGTTTGTGT 45625703
45625702 GTTTTTAGTAGCCTGCTTCTTAGATGTATTTTGCTCTTTTAAAAAGCTTAATCTCCCCTA 45625643
45625642 CAGTTGTCCAGCAATTAATTAAATGTGGTGTAAGTTTATCTTAAACTTCCTTAAACAGAC 45625583
45625582 TGCATCTTGTTCAAATGCAATTAGGAACAAACCTTTCATCTCTGGTGAATGAAACTTTCA 45625523
45625522 ATAAATAAGGCTGCTTGTGTCAGACAGGTGGCTCCCTCCTGCCATTTGCCAGGGTTTTTA 45625463
45625462 CAGAGAAAACCCTGCCATCAGCATGAGATGACCCTTGCTCAGGTGGCCCCAGGAAAGGTC 45625403
45625402 ATCTCCTGCTGAGCTACCTAGGAAAATCCTGCAGTCATCTTCCCTCCAAAAGACCCCATC 45625343
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45625222 AGCTGGGGCACCCAAACAAGCTTTATTTAATCACAATAGAACATTCCAAGACAGGACAAG 45625163
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45624802 GCCTACAGGTGGCTCATGAGGCCTCGTTTGGGAGGGTATGAGTGTAAAGAACAAGGGCTG 45624743
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45624682 CCTGTCATCTTGCCTTTGCACCAGGCCTTTCCCCCTAGTTCTCTGGCCTTCTACCACCAC 45624623
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45624142 GGAGCTAGGCCCTGTCCTCAGCCTTCCACTTGCCATGGGCCCCTGTTGGCTGCGGACTGA 45624083
45624082 GGCATCTGTGTAACTTGCAGGCACTTTTCCTCCATATTGTTAGGCTGAGGCATTAGAAGC 45624023
45624022 CTGTTTCAGGGTTATAGTGCAGACAATTAGGAATGTGGTTTTGTTTGTTTGTGTTTTTAA 45623963
45623962 CCACAGTGTCTGAAACTACTGGTAGGCATGCTCGGGAGTTTGGGTACCCTGGGTGGCTTG 45623903
45623902 TCTCCCAATCCTCTTTGAGACATCTATTCCTAGATGAGCTATGGGGAAATATTGAAAACA 45623843
45623842 CGTCTATTCAGATGAGGATTCTGACAGCCTCCATTGTCCCCCATAGATCTAACTTGCCAT 45623783
45623782 TTGCTGCTCTCGTTGCCCTTAG
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http://www.fruitfly.org/seq_tools/splice.html

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**TCCCCCATAG**ATCTAACTTGCCATTGCTGCTCTCGTTGCCCTTAGacttgaagccccagatgctgtcgggcacagtgccc
tgt

Yellow, selected by software tool
Bold yellow, best scores ( > 0.88)
Green, sequence primer Ex34F
Small print, exon 34

Supplementary Figure 7. Identification of cryptic splice acceptor sites in the intronic sequence upstream of the c.4460-1G>C mutation and exon 34. The upper panel shows the manually annotated consensus splice acceptor (SA) sites, while the lower panel indicates those selected by the software tool from www.fruitfly.org. Upper panel: SA dinucleotide AG (yellow), consensus SA (pink), sequence primer g.E_34F (green). Lower panel: Software selected consensus SA (yellow) with high ranking scores in bold.