

Supplemental Figure 1

a. Exon 1/2 junction:

Exon 1  
46999-46612 (genomic)

Exon 2  
42022-41949 (genomic)

|         |  |                                      |
|---------|--|--------------------------------------|
| genomic | GCCCAGCGGAGCATGTCTCGGAGGACAGCGACATGGAGAAAGCCATCAAGGTAAGCGGGA | TTTTTAATAGGAGACCTCCATTTTAGAAGAATATA/ |
|         |  |                                      |
| MK5.1   | <u>ATGTCGGAGGACAGCGACATGGAGAAAGCCATCAAG</u>                  | <u>GAGACCTCCATTTTAGAAGAATATA/</u>    |
|         |  |                                      |
| MK5.2   | <u>ATGTCGGAGGACAGCGACATGGAGAAAGCCATCAAG</u>                  | <u>GAGACCTCCATTTTAGAAGAATATA/</u>    |
|         |  |                                      |
| MK5.3   | <u>ATGTCGGAGGACAGCGACATGGAGAAAGCCATCAAG</u>                  | <u>GAGACCTCCATTTTAGAAGAATATA/</u>    |
|         |  |                                      |
| MK5.4   | <u>ATGTCGGAGGACAGCGACATGGAGAAAGCCATCAAG</u>                  | -----/                               |
|         |  |                                      |
| MK5.5   | <u>ATGTCGGAGGACAGCGACATGGAGAAAGCCATCAAG</u>                  | -----/                               |
|         |  |                                      |
|         | M S E D S D M E K A I K                                      | E T S I L E E Y /                    |

b. Exon 6/7 junction:

Exon 6  
37390-37301 (genomic)

Exon 7  
35877-35782 (genomic)

|         |   |                                     |
|---------|---|-------------------------------------|
| genomic | /CAGAGACCTCAAGCCTGAAAATCTGCTTTTCAAGGATAACTCTCTGGTGAGAACAA | TTTCTTTCAGGACGCCCTGTGAAATTATGTGACT/ |
|         |   |                                     |
| MK5.1   | <u>/CAGAGACCTCAAGCCTGAAAATCTGCTTTTCAAGGATAACTCTCTG</u>    | <u>GACGCCCTGTGAAATTATGTGACT/</u>    |
|         |   |                                     |
| MK5.2   | <u>/CAGAGACCTCAAGCCTGAAAATCTGCTTTTCAAGGATAACTCTCTG</u>    | <u>GACGCCCTGTGAAATTATGTGACT/</u>    |
|         |   |                                     |
| MK5.3   | <u>/CAGAGACCTCAAGCCTGAAAATCTGCTTTTCAAGGATAACTCTCTG</u>    | <u>GACGCCCTGTGAAATTATGTGACT/</u>    |
|         |   |                                     |
| MK5.4   | /-----  | <u>GACGCCCTGTGAAATTATGTGACT/</u>    |
|         |   |                                     |
| MK5.5   | /-----  | <u>GACGCCCTGTGAAATTATGTGACT/</u>    |
|         |   |                                     |
|         | / R D L K P E N L L F K D N S L                           | D A P V K L C D /                   |

c. Exon 11/12/13 junction:

|         | Exon 11<br>28668-28538 (genomic) | Exon 12<br>28307-28186                       | Exon 13<br>27214-27110       |
|---------|----------------------------------|--|------------------------------|
| genomic | /CTGCTGGGGTGAGTGAGCGCATTCTTT     | ATTTGTAAAGCACCAAGCC//CAGGCTGGTAAAGGTCATGCCAC | TTGCCACAGGAGAGAATGAA/        |
|         |                                  | //   |                              |
| MK5.1   | /CTGCTGGG-----                   | <u>CACCAAGCC</u> //CAGGCTGGTAAAG             | <u>GAGAGAATGAA</u> /         |
|         |                                  | //   |                              |
| MK5.2   | /CTGCTGGG-----                   | <u>CACCAAGCC</u> //CAGGCTG-----              | <u>GAGAGAATGAA</u> /         |
|         | -       - -   -                  | //   |                              |
| MK5.3   | /CTGCTGGG <b>GAGAGAATGA</b>      | -----//-----                                 | <u>GAGAGAATGA</u> <b>A</b> / |
|         |                                  | //   |                              |
| MK5.4   | /CTGCTGGG-----                   | <u>CACCAAGCC</u> //CAGGCTGGTAAAG             | <u>GAGAGAATGAA</u> /         |
|         |                                  | //   |                              |
| MK5.5   | /CTGCTGGG-----                   | <u>CACCAAGCC</u> //CAGGCTG-----              | <u>GAGAGAATGAA</u> /         |
|         |                                  | //   |                              |
|         | /L L G                           | T K P //Q A G K G                            | E N E /<br>R E *             |

Supplemental Figure 2

