## 1.5 unc-13 EMS mutations summary

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **EMS alleles** | Survival | **Mutation**  | **Effect on protein** | **Location (In genomic from AE003847.2)** | Transcripts affected) |
| 7.5 | Adults | 1bp (T) deletionDownstreamGGC  AGC | FrameshiftTruncated proteinGly  Ser | Exon 1DBase 2856Exon 1D | AC |
| 7.96 | Adults | 8bp deletion | FrameshiftTruncated protein | Exon 1DBase 4389 | AC |

## 1.6 unc-13 EMS mutations in detail

### *EMS allele : 7.5*

*Location:*

In AE003847.2

# Exon 1, Base 2856

*Alignment***:**

Exon1-genomic ATATACCTACGACATCTCCATCTTGTACTCAAAAAAATGGCGAGGAAAATTTATATTCGG

7.5\_1 ATATACCTACGACATCTCCATCTTGTACTCAAAAAAATGGCGAGGAAAATT-ATATTCGG

7.5\_2 ATATACCTACGACATCTCCATCTTGTACTCAAAAAAATGGCGAGGAAAATT-ATATTCGG

7.5\_3 ATATACCTACGACATCTCCATCTTGTACTCAAAAAAATGGCGAGGAAAATT-ATATTCGG

 \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*

*Protein translation:*

## Genomic:

acgacatctccatcttgtactcaaaaaaatggcgaggaaaatttatattcggctacagtc

 T T S P S C T Q K N G E E N L Y S A T V

*7.5:*

cccctctataacaacaggatcagaactaatcctaccctaatattgatttcacctgaaaat

 P L Y N N R I R T N P T L I L I S P E N

gtttctagtcgtaacagtaattatatacctacgacatctccatcttgtactcaaaaaaat

 V S S R N S N Y I P T T S P S C T Q K N

ggcgaggaaaattatattcggctacagtccacaataagtcaacgaaatcgaattcatact

 G E E N Y I R L Q S T I S Q R N R I H T

ataatgaagtgggagaaattagttcgaccttagtcagaaatgtttgtgacagttacgata

 I M K W E K L V R P - S E M F V T V T I

atagctgtgatgagatgattttaaccaacgaaatggtaaatatagggatgctcgacagtg

 I A V M R - F - P T K W - I - G C S T V

aatctgagtttggcttaatagaaaattcgtattcgtatcaggtaccagacaatgagcaaa

 N L S L A - - K I R I R I R Y Q T M S K

tcgacagtgttaactcgtataataataagacccaaaatgtaacaaataatagcatagaaa

 S T V L T R I I I R P K M - Q I I A - K

aggcaaatacaaaaaataaaccagtgcctcttcatgaccctcctacgaaaaaagcaagta

 R Q I Q K I N Q C L F M T L L R K K Q V

ctgttggcatgttcggatcaattttgggaaaagctgctgcagcagtgcaatcagcaacgc

*Protein alignment:*

Genomic SVGGLLTSTSSTSIKQTGSETNPTLILISPENVSSRNSNYIPTTSPSCTQKNGEENLYSA

7.5 ----------------------PTLILISPENVSSRNSNYIPTTSPSCTQKNGEENYIRL

 \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*

Genomic TVHNKSTKSNSYYNEVGEISSTLVRNVCDSYDNSYDEMILTNEMVNIGMLDSESEFGLIE

7.5 QSTISQRNRIHTIMKWEKLVRP-SEMFVTVTIIAVMR-F---------------------

### *EMS allele: 7.96*

*Location:*

In AE003847.2

Exon 1, Base 4389

*Alignment:*

Genomic CCGATCTTACTGACTTAAGTAAACTAATCTCACAAAAAAAAAAAACGCTCTTGAGAGGAG

7.96-1 CCGATCTTACTGACTTAAGTAAACTAATCTCACAAAAAT--------CTCTTGAGAGGAG

7.96-2 CCGATCTTACTGACTTAAGTAAACTAATCTCACAAAAAT--------CTCTTGAGAGGAG

7.96-3 CCGATCTTACTGACTTAAGTAAACTAATCTCACAAAAAT--------CTCTTGAGAGGAG

 \*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\*\* \*\*\*\*\*\*\*\*\*\*\*\*\*

*Protein translation:*

*Genomic*

gaagatgaaaacctaagcgatcacgtttccgatcttactgacttaagtaaactaatctca

 E D E N L S D H V S D L T D L S K L I S

caaaaaaaaaaaacgctcttgagaggagaaactgaagaagtcgtaggaggacacatgcag

 Q K K K T L L R G E T E E V V G G H M Q

*7.96*

 gatgaaaacctaagcgatcacgtttccgatcttactgacttaagtaaactaatctca

 D E N L S D H V S D L T D L S K L I S

caaaaatctcttgagaggagaaactgaagaagtcgtaggaggacacatgcaggtgttacg

 Q K S L E R R N - R S R R R T H A G V T

ccaaacggaaatcacagctcggcaacgatggcattgggcttacaataaaataatcatgca

 P N G N H S S A T M A L G L Q - N N H A

gcttaatgtaagtttgtaatacacatttttttaatatttattataatatagtttttgttg

A - C K F V I H I F L I F I I I - F L L

agtgtcacgacttttaaatacctgccctcaattaaagtaggggcaacggaaattatattg

S V T T F K Y L P S I K V G A T E I I L

*Protein alignment:*

Genomic DLTDLSKLISQKKKTLLRGETEEVVGGHMQVLR---QTEITARQRWHWAYN----KIIMQ

7.96 DLTDLSKLISQKSLERRNSTPRSRRRTHAGVTPNGNHSSATMALGLQSTPNNHAASTPCK

 \*\*\*\*\*\*\*\*\*\*\*\*. .. ... \* \* ::. \* : : \* . :

**1.7 Location of EMS mutations (diagram)**

Adapted from diagrams in James' Eurotalk 2003

Felicity/Dunc13/Sequencing of EMS alleles/Dunc13 EMs mutations/Powerpoint – EMS mutations diagram

***Protein truncation***

**7.5**

**1bp**

**deletion**

**A**

**C**

**B**

**7.5**

**360kDa**

**7.96**

**8bp**

**deletion**

**B**

**7.96**

**A**

**C**

**C1**

**A**

**C**

**B**

**C2B**

**C2C**

**P84200**

**A**

**C**

**B**

**Wild Type**

**320kDa**

**320kDa**