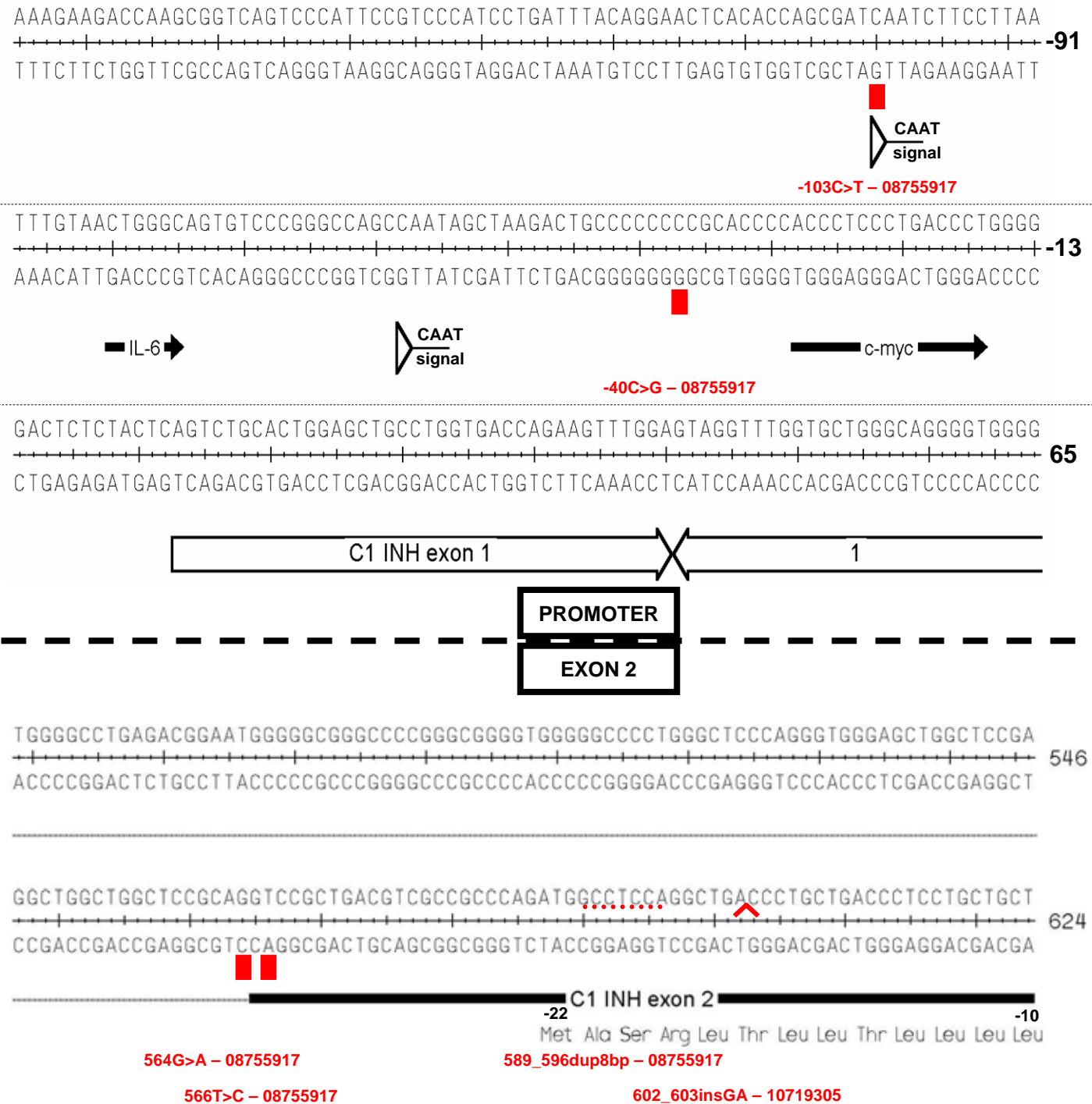


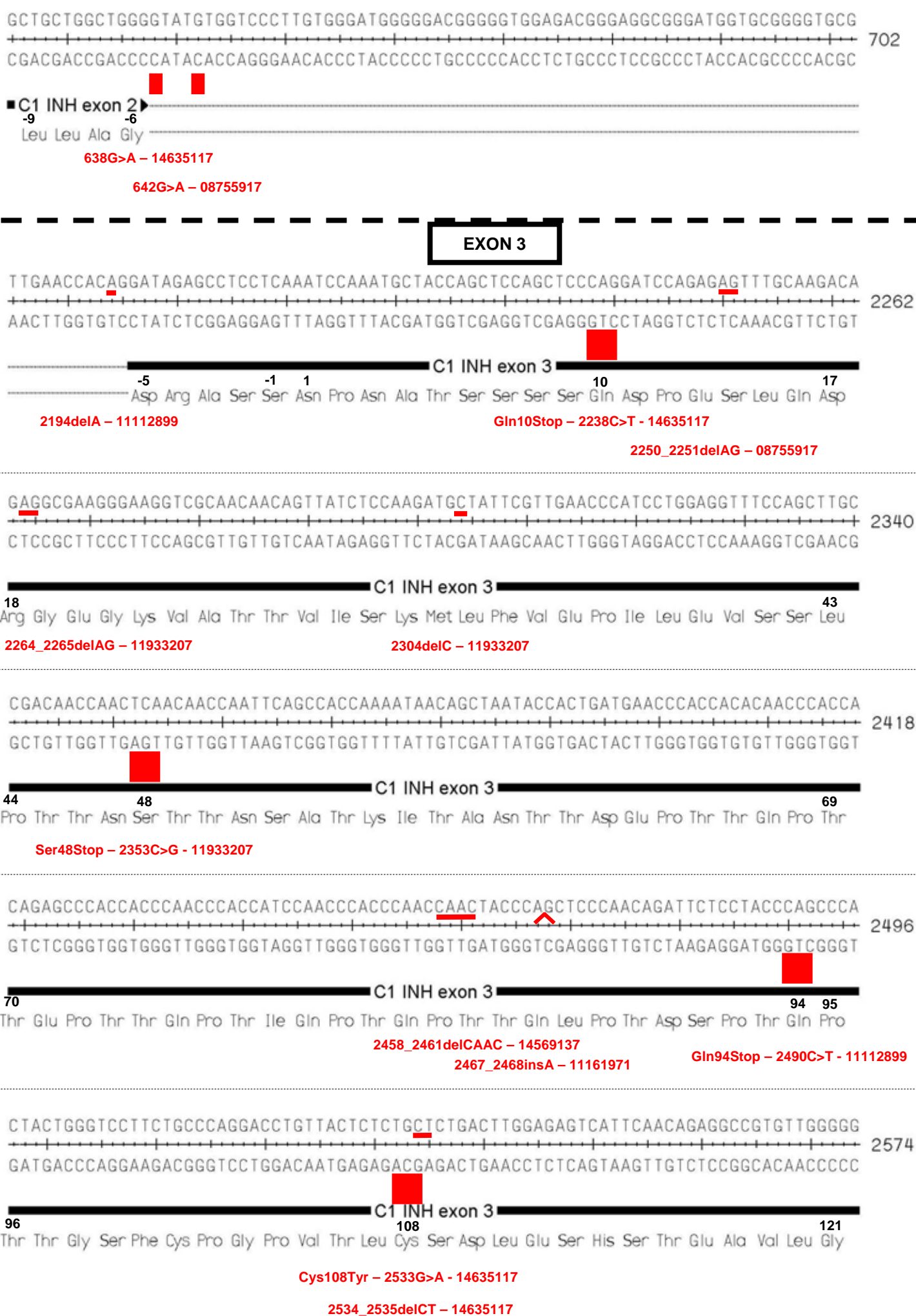
C1 inhibitor gene mutation map (micro mutations)

(with article PubMed identification numbers)

Last Update:
2004.04.22.

- Nucleotide substitution (regulatory, splicing site)
- Missense/nonsense mutation
- Duplication
- ^ Insertion
- Deletion





ATGCTTTGGTAGATTTCTCCCTGAAGCTCTACCACGCCTTCTCAGCAATGAAGAAGGTGGAGACCAACATGGCCTTTT 2652
TACGAAACCATCTAAAGAGGGACTTCGAGATGGTGC GGAAGAGTCGTTACTTCTTCCACCTCTGGTTGTACCGGAAAA
122 127 133 147
C1 INH exon 3
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Phe127Val - 2589T>G - 09069585

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C1 INH exon 3
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Ser148Pro - 2652T>C - 1112899 2679_2680insTT - 08755917
Pro149Leu - 2656C>T - 08755917 Gly162Arg - 2694G>A - 08755917
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EXON 4

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162 167
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168 169 183 193
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194 196 206
C1 INH exon 4
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4467C>T - Gln201Stop - 14635117

EXON 5

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207 C1 INH exon 5 214
 Asp Leu Ala Ile Arg Asp Thr Phe
 Phe214Ser - 8346T>C - 08755917

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215 C1 INH exon 5 240
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 Ser224Arg - 8377C>A - 09069585
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241 C1 INH exon 5 266
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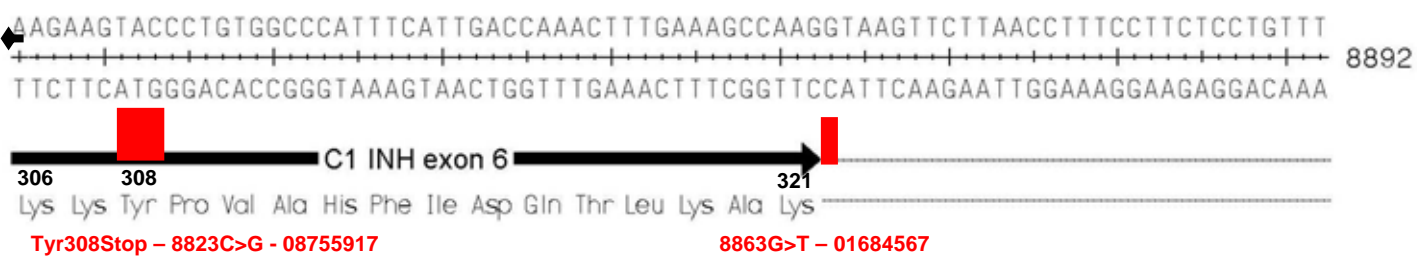
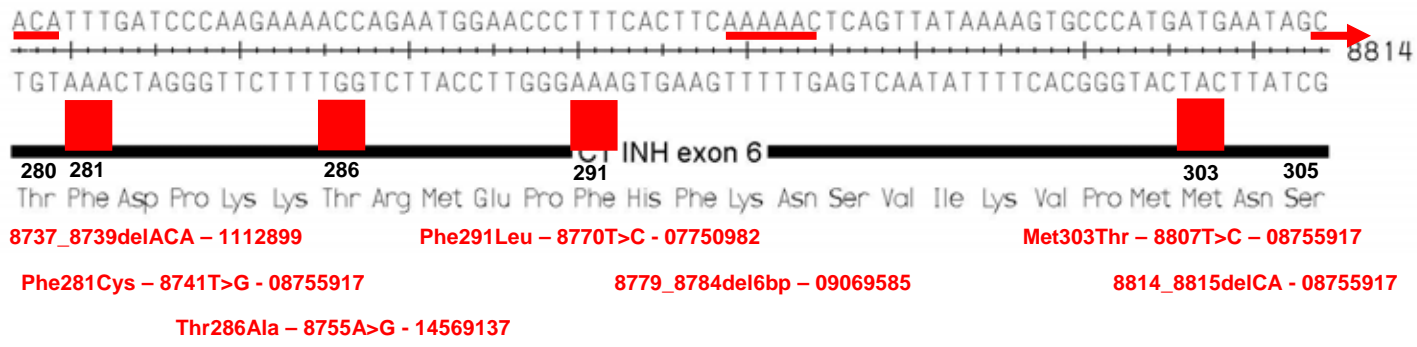
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267 C1 INH exon 5 274
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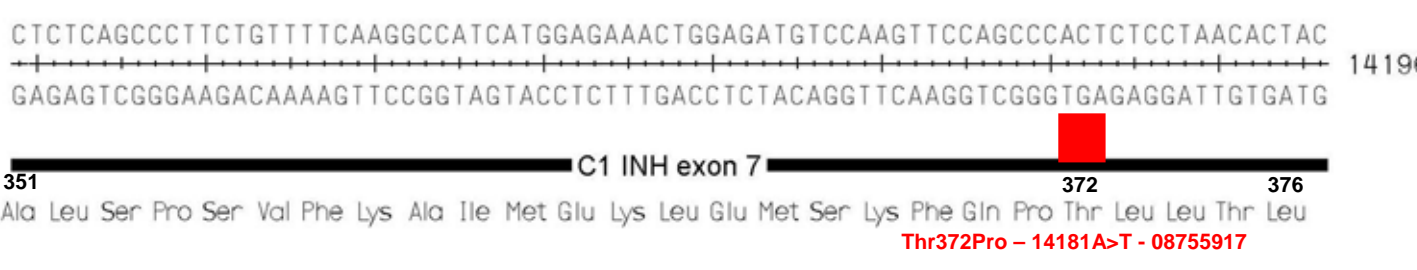
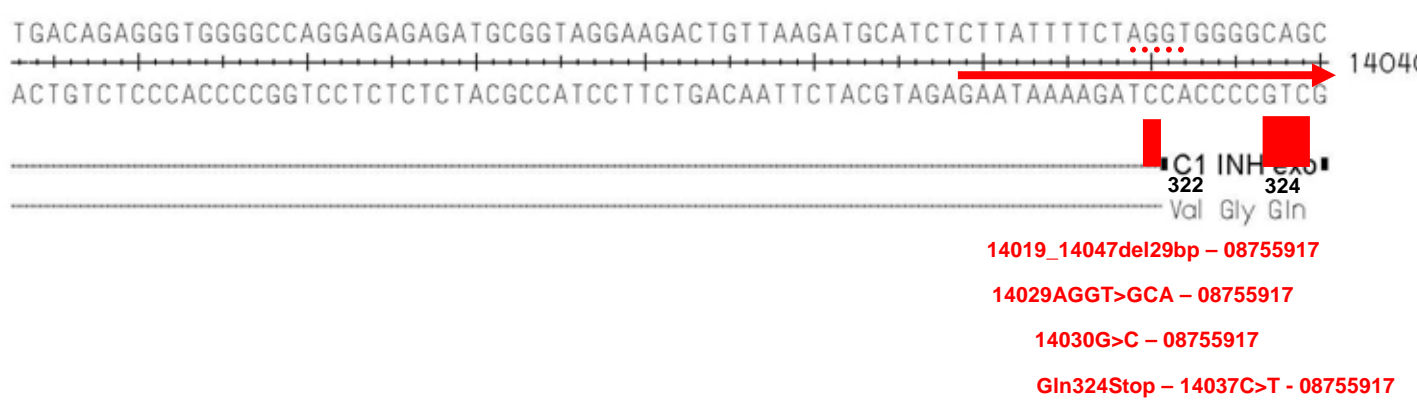
EXON 6

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275 C1 INH exon 6 279
 Ala Lys Trp Lys Thr
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 8722G>A - 15098611
 Thr279Lys - 8735C>A - 09069585
 Thr279Ile - 8735C>T - 09069585



EXON 7



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EXON 8

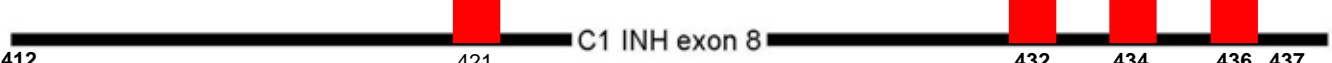
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