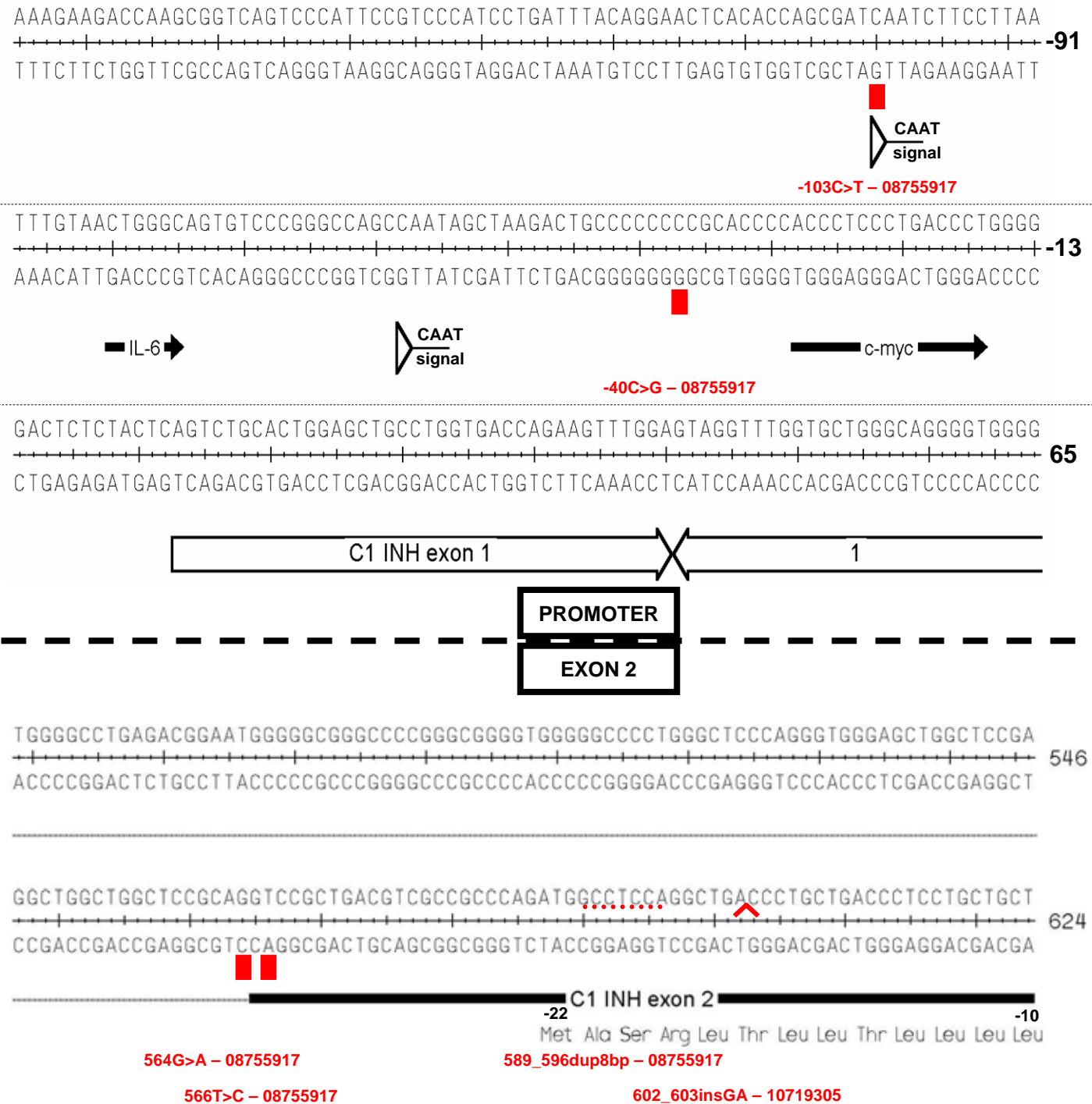


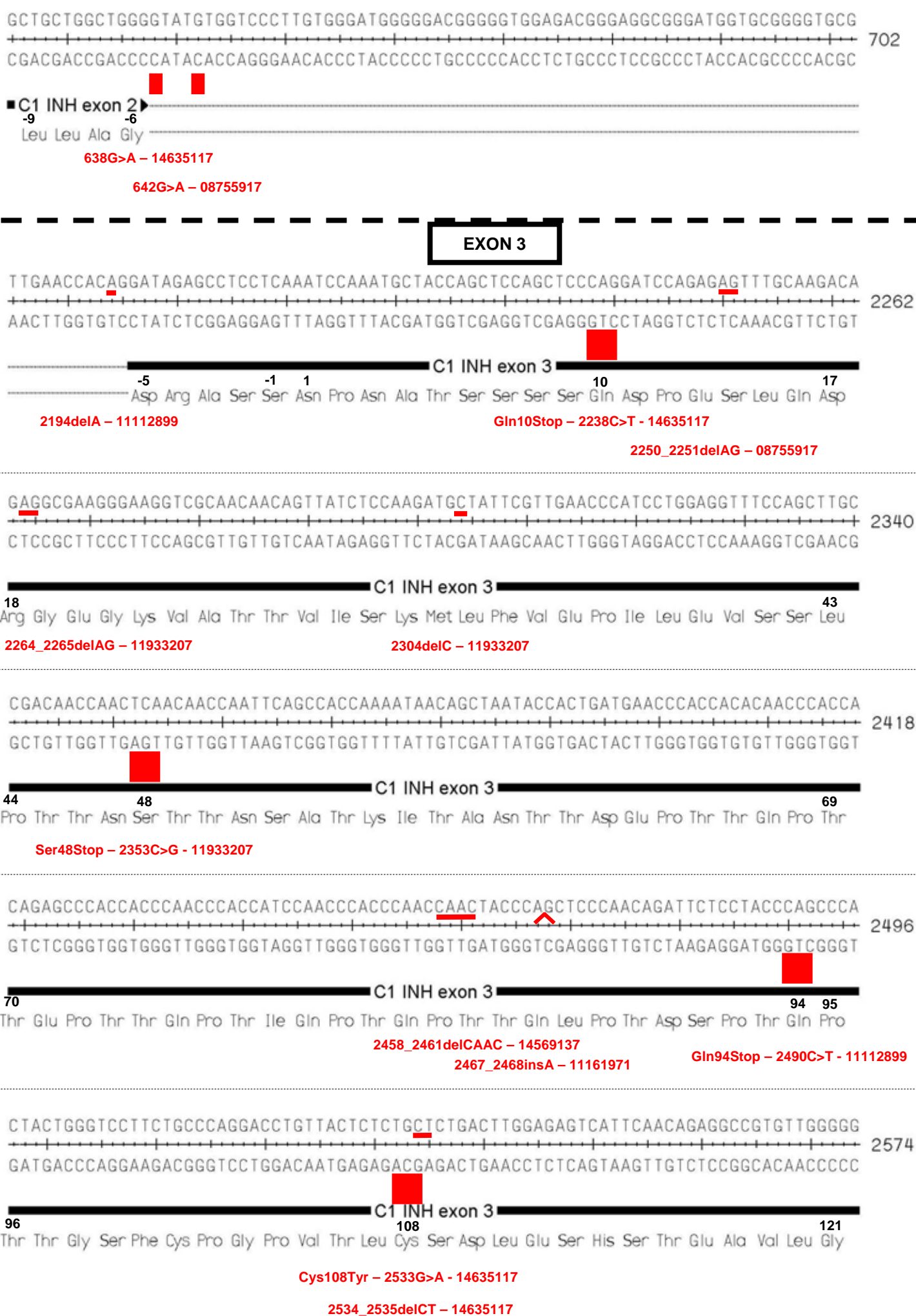
C1 inhibitor gene mutation map (micro mutations)

(with article PubMed identification numbers)

Last Update:
2004.04.22.

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





ATGCTTTGGTAGATTTCTCCCTGAAGCTCTACCACGCCTTCTCAGCAATGAAGAAGGTGGAGACCAACATGGCCTTTT 2652
 TACGAAACCATCTAAAGAGGGACTTCGAGATGGTGCGGAAGAGTCGTTACTTCTTCCACCTCTGGTTGTACCGGAAAA

C1 INH exon 3

122 127 133 147

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 Phe127Val - 2589T>G - 09069585

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C1 INH exon 3

148 149 155 161

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Ser148Pro - 2652T>C - 1112899 2679_2680insTT - 08755917
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C1 INH exon 4

162 167

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Gly162Glu - 4351G>A - 10719305

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C1 INH exon 4

168 169 183 193

Lys Thr Asn Leu Glu Ser Ile Leu Ser Tyr Pro Lys Asp Phe Thr Cys Val His Gln Ala Leu Lys Gly Phe Thr Thr

Thr169Pro - 4371A>G - 09069585 4400delC - 08755917 4428delC - 10719305
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C1 INH exon 4

194 196 206

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Val196Asp - 4453T>A - 10719305
 4460_4461insA - 10719305
 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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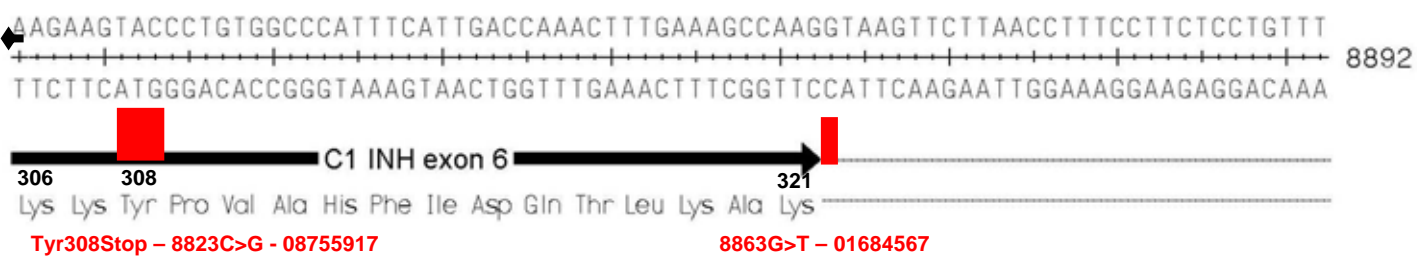
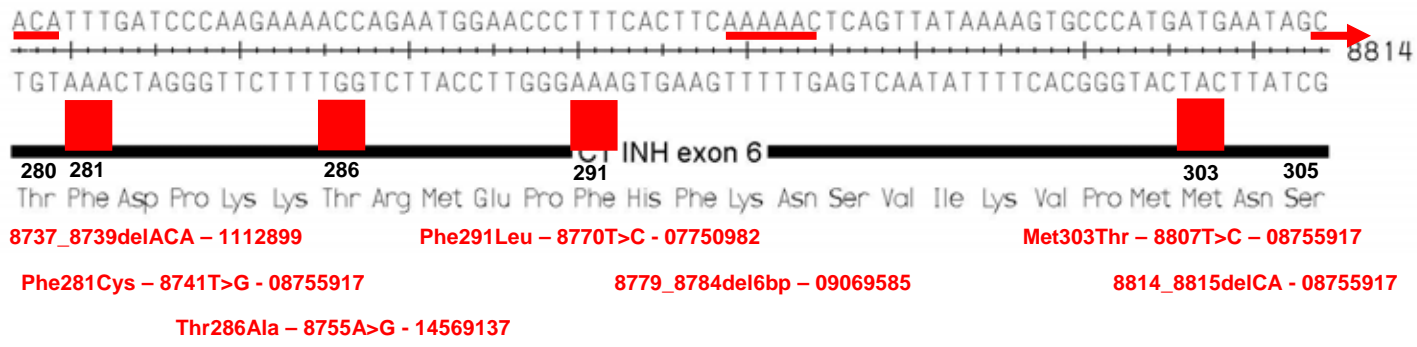
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267 C1 INH exon 5 274
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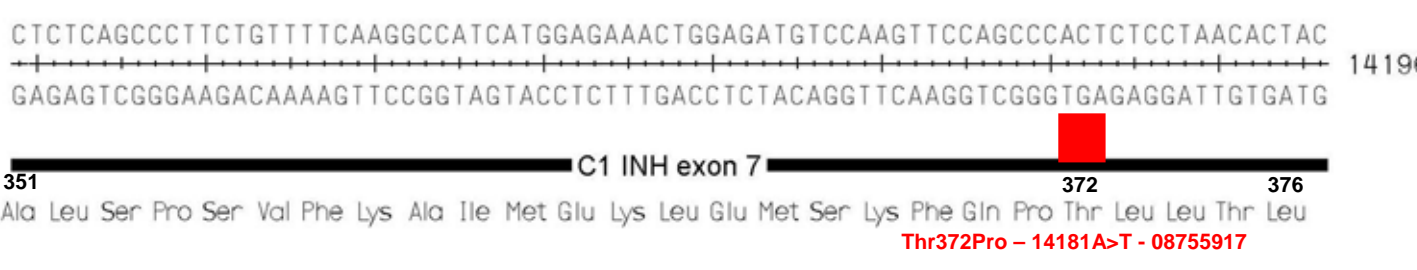
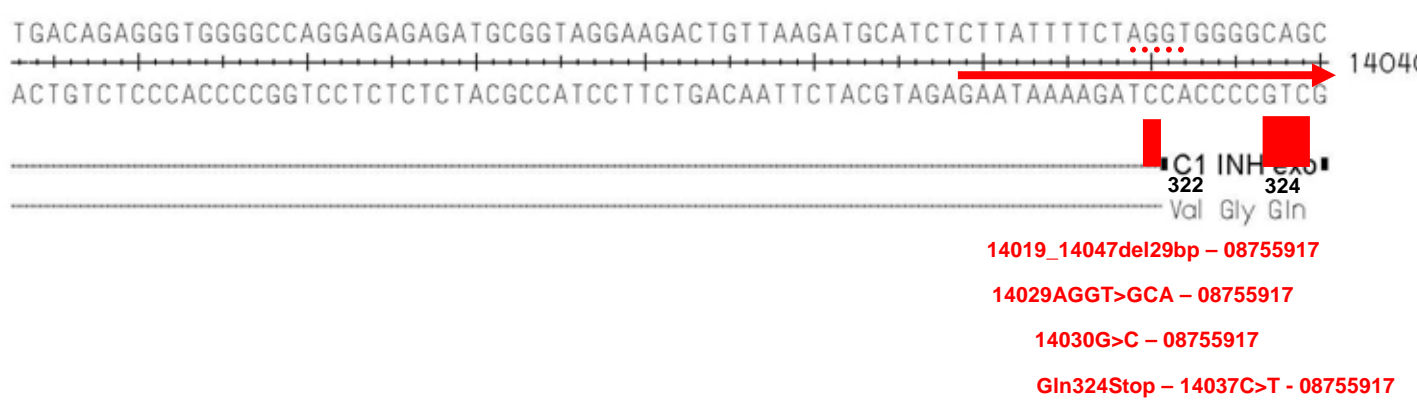
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275 C1 INH exon 6 279
 Ala Lys Trp Lys Thr
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 8722G>A - 15098611
 Thr279Lys - 8735C>A - 09069585
 Thr279Ile - 8735C>T - 09069585



EXON 7



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Pro Arg Ile Lys Val Thr Thr Ser Gln Asp Met Leu Ser Ile Met Glu Lys Leu

Pro377Ser - 14196C>T - 09069585 Asp386Val - 14224A>T - 14635117

Pro377Ala - 14196C>G - 09069585 Ser389Stop - 14233C>A - 11112899

Lys380Ile - 14206A>T - 09069585

14252T>A - 09579556

EXON 8

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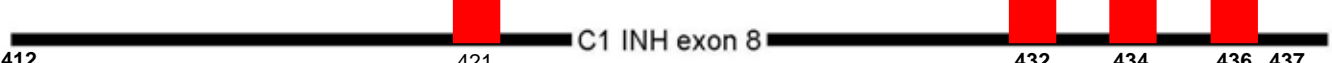
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 Ala443Val - 16786G>A - 07883978 Phe455Ser - 16822T>C - 08755917
 Arg444Cys - 16788C>T - 02563376 Phe457Leu - 16827T>C - 11161971
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 Arg444Pro - 16789G>C 12402344
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464 467 468 470 471 472 C1 INH exon 8 476 479
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 Gly471Arg - 16869G>A - 11315937
 Gly471Glu - 16870G>A - 12402344
 Arg472Stop - 16872C>T - 08755917