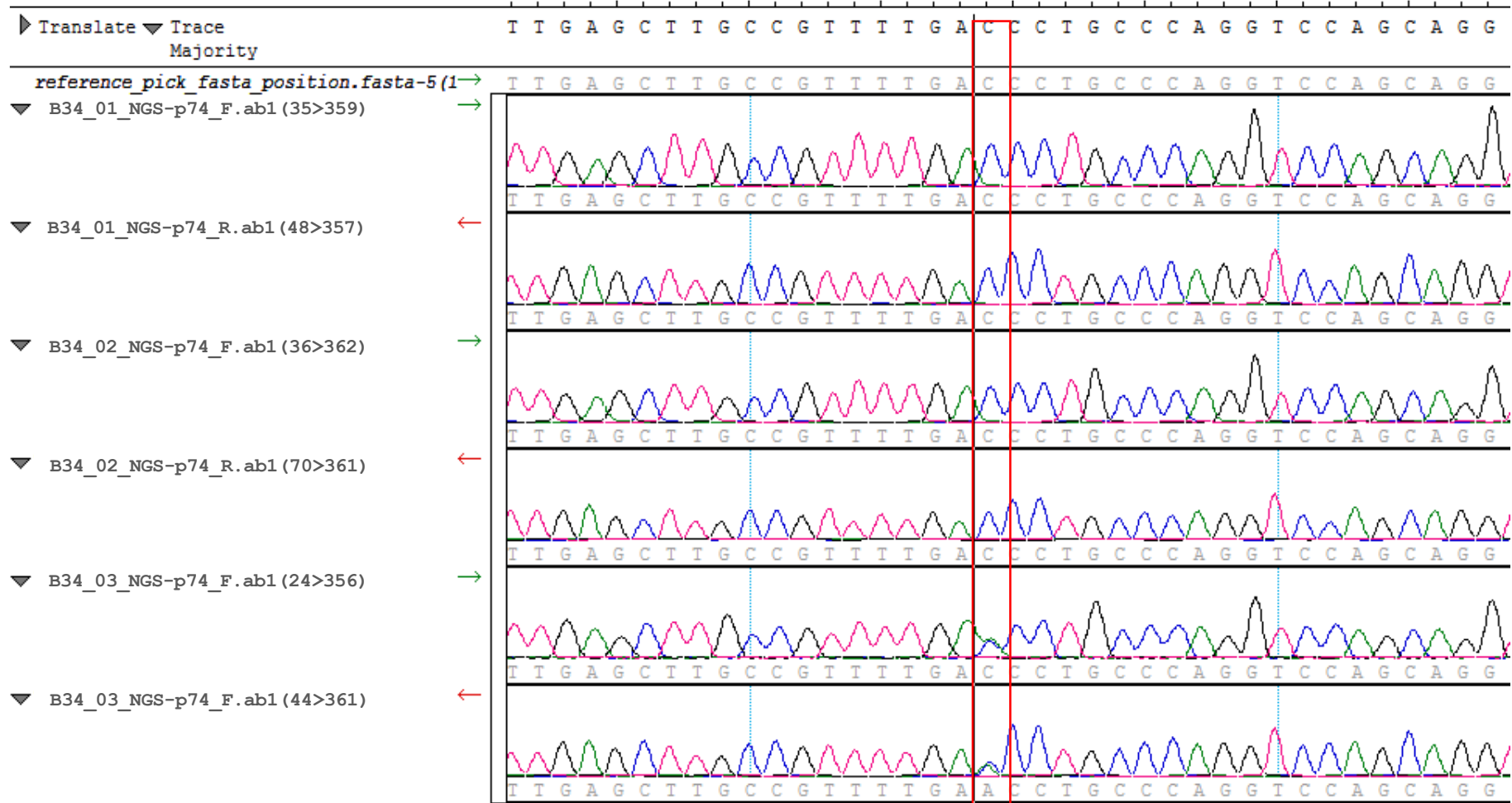
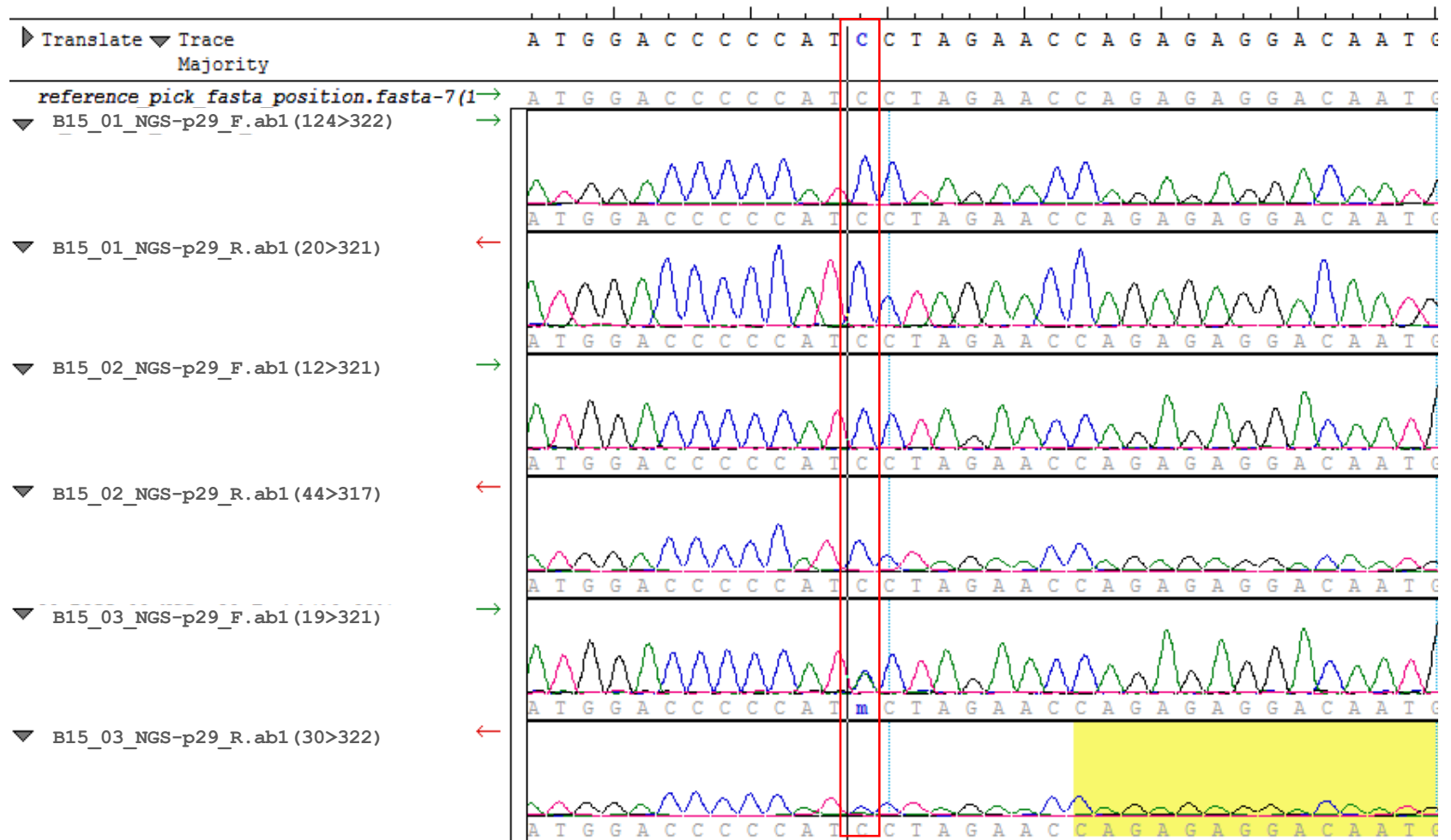


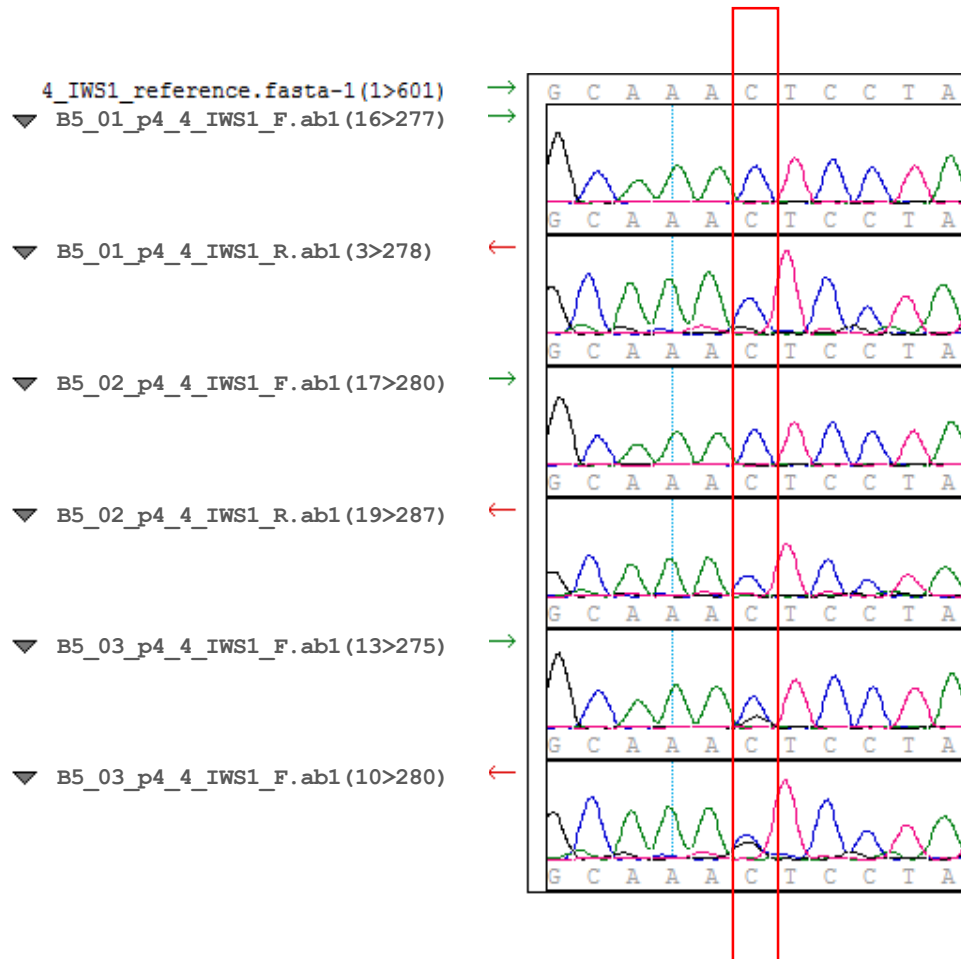
AMIGO1



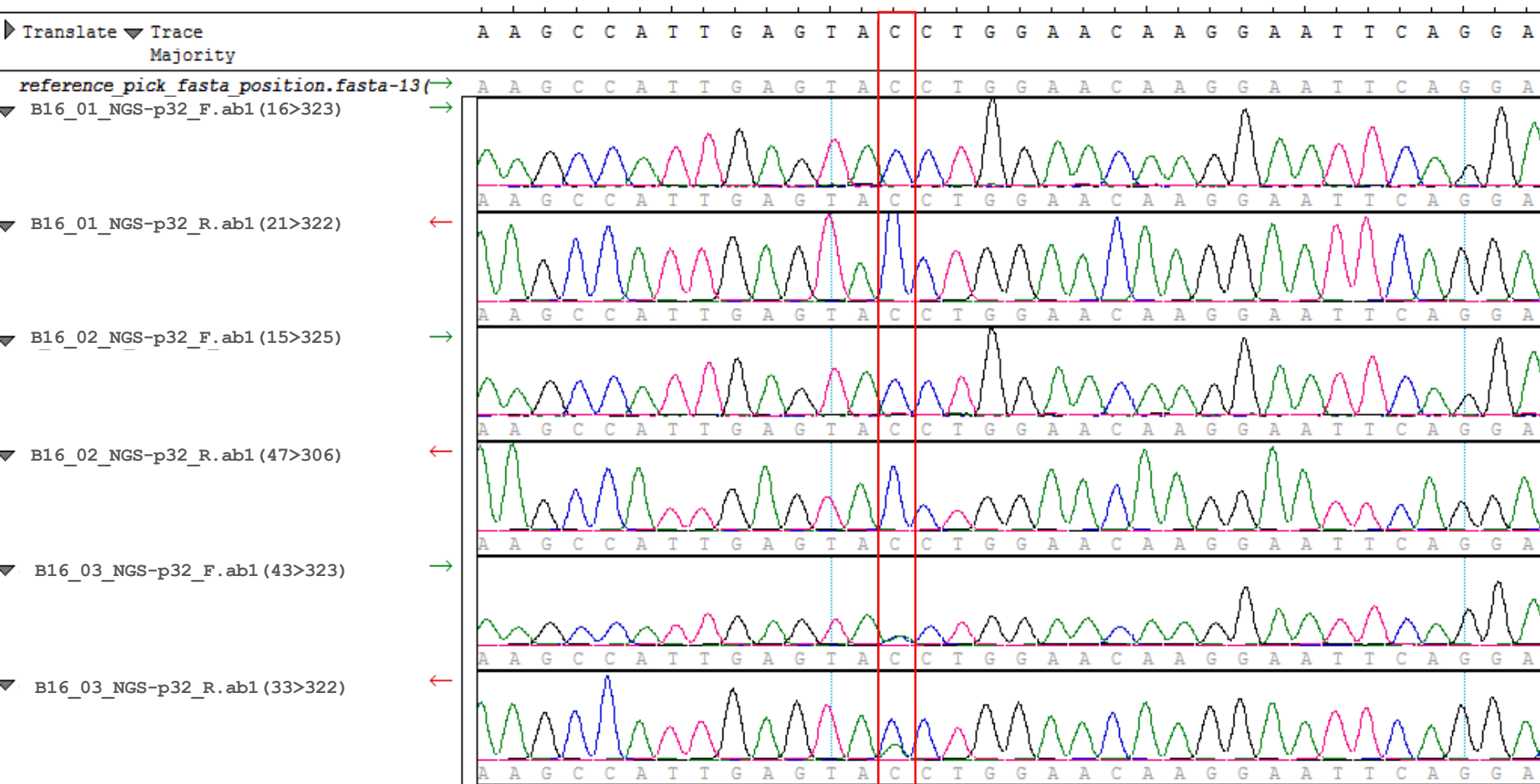
HAX1



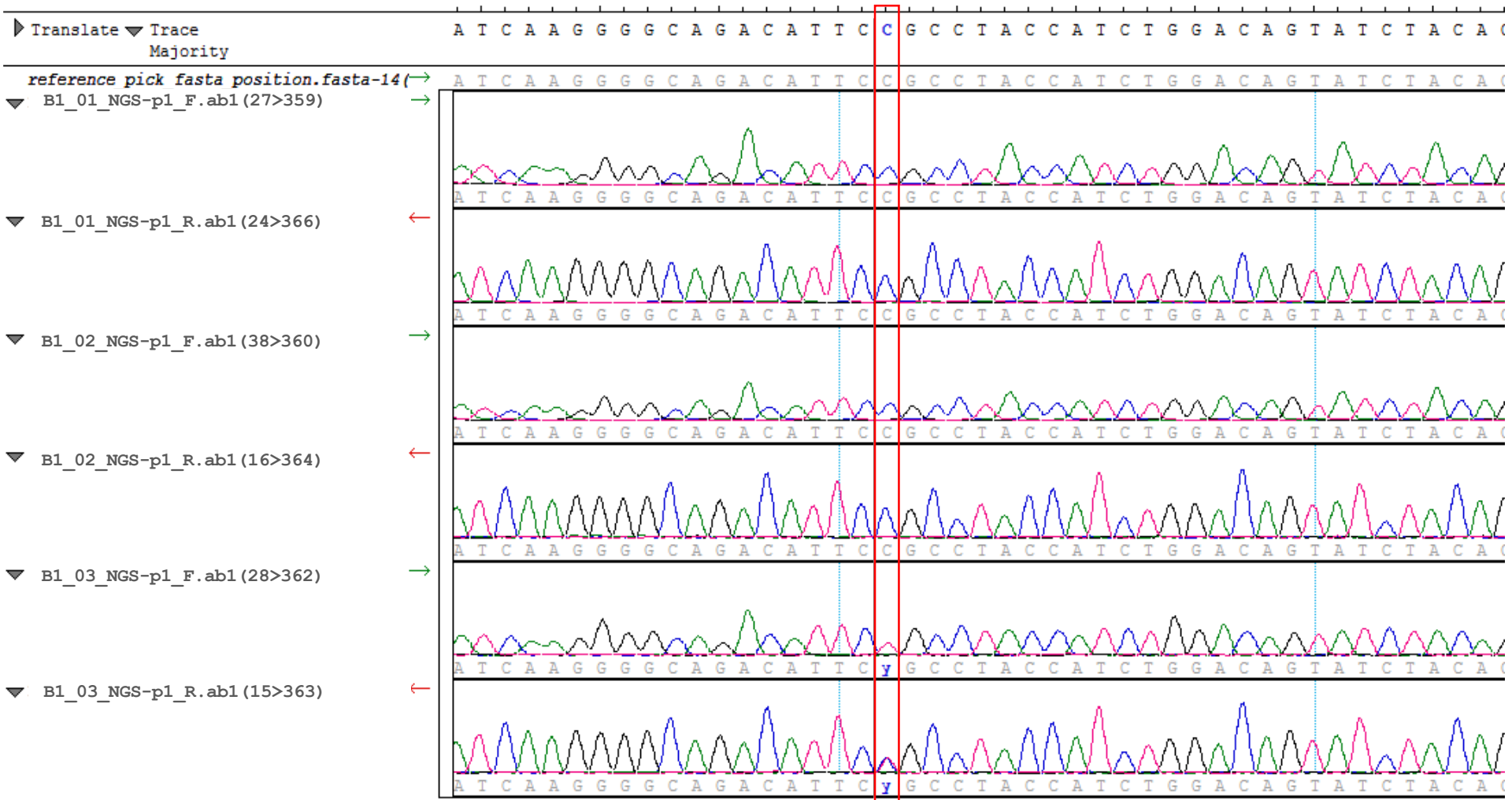
IWS1



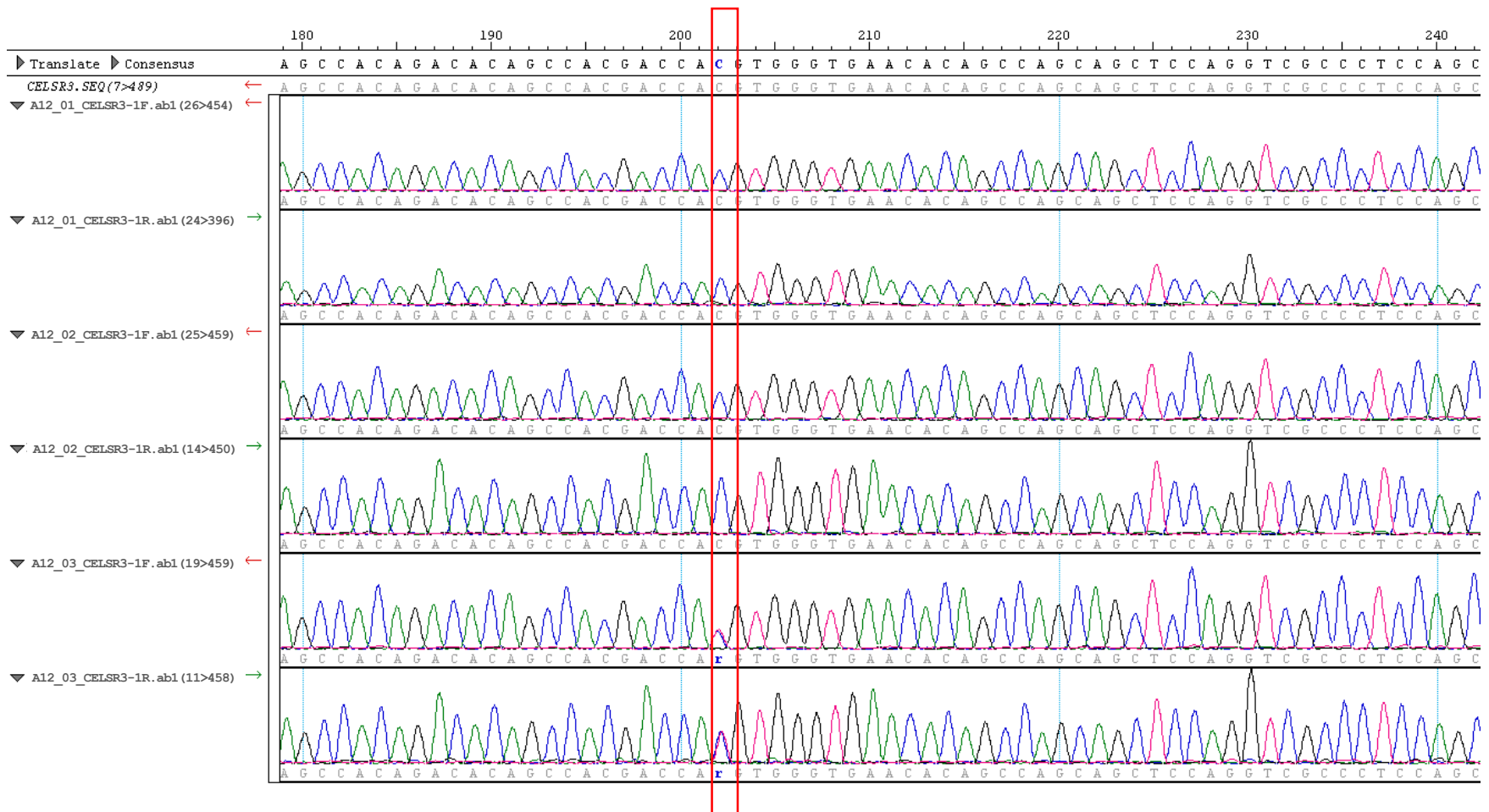
TTC21A



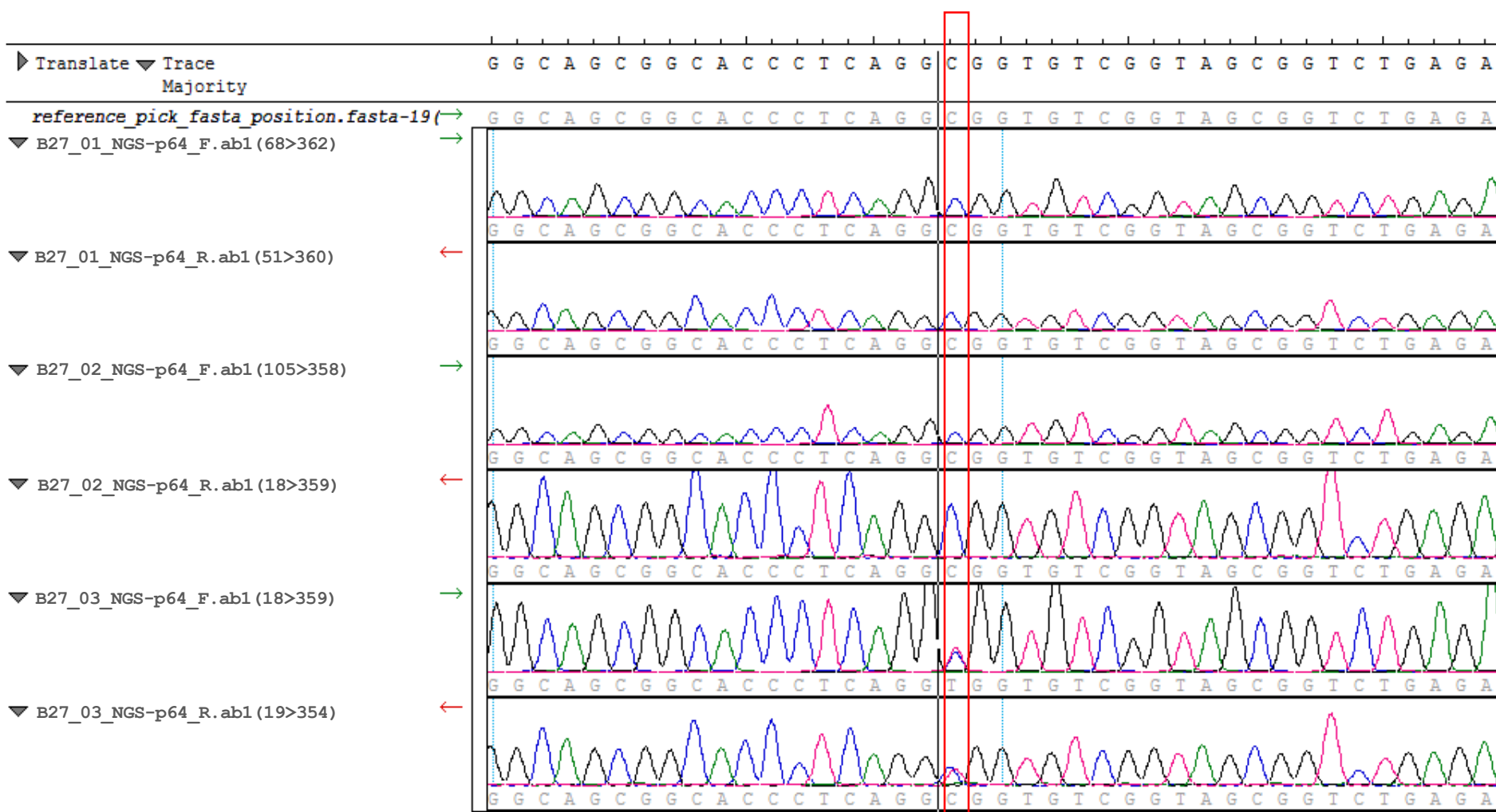
LARS2



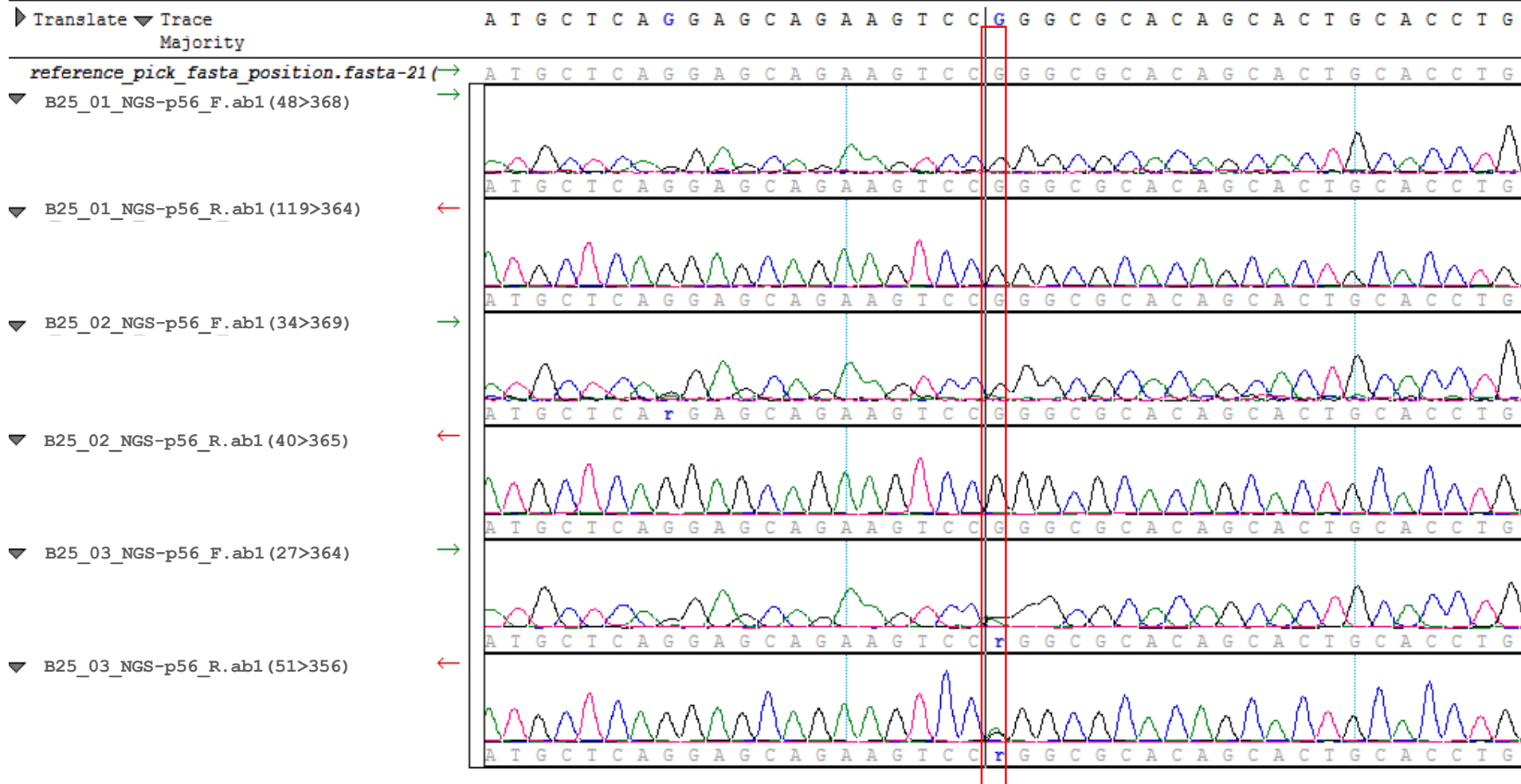
CELSR3



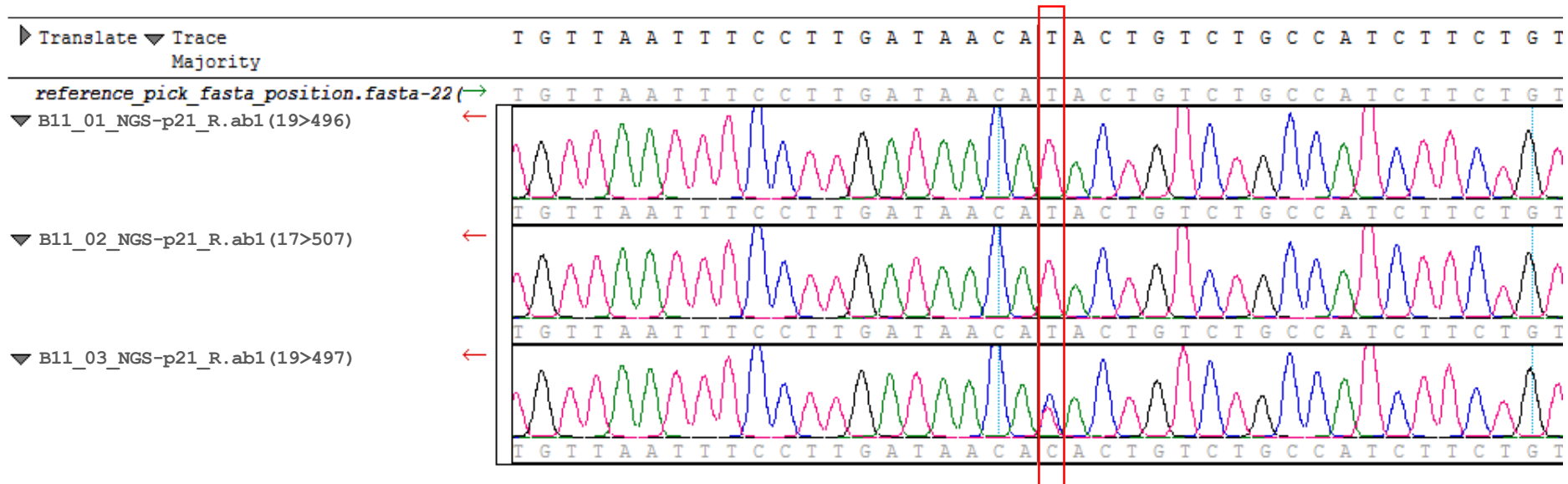
SLC26A1



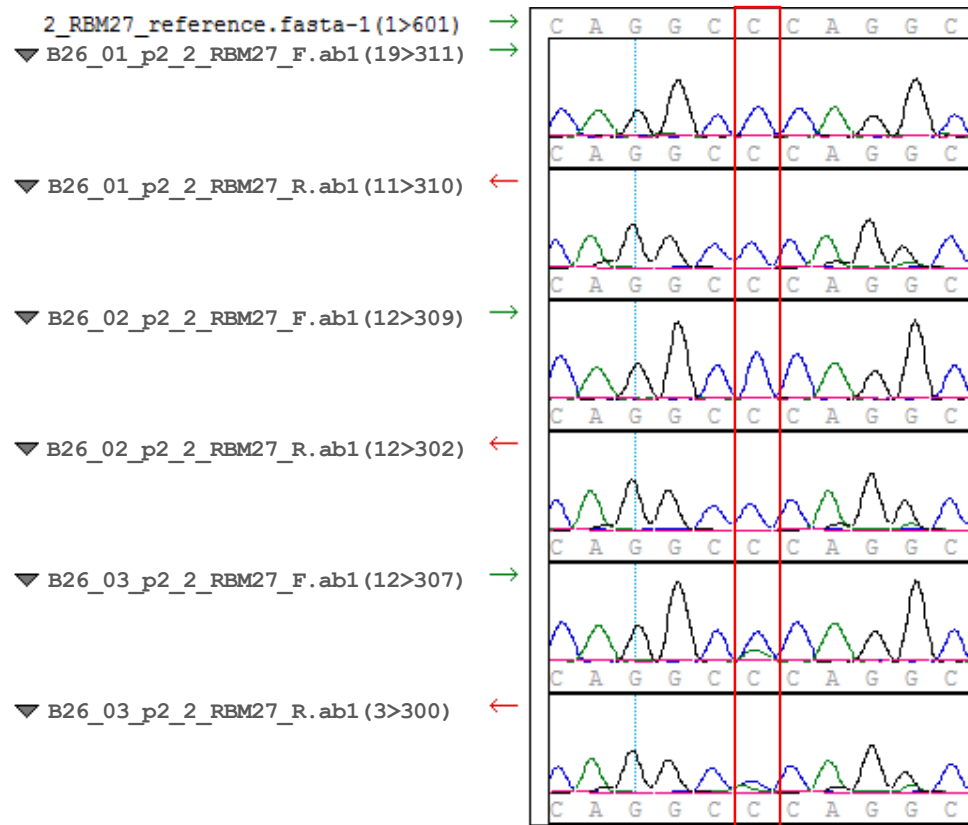
NFKB1



NEK1



RBM27



► Translate ▼ Trace
Majority

reference_pick_fasta_position.fasta-26(→ C C T T C T T C C C G T C C T G T A G C A C C A C T G C C G G A A C T G T G G C C A C A T C T T

▼ B29_01_NGS-p68_F.ab1 (31>370) →
C C T T C T T C C C G T C C T G T A G C A C C A C T G C C G G A A C T G T G G C C A C A T C T T

▼ B29_01_NGS-p68_R.ab1 (35>367) ←
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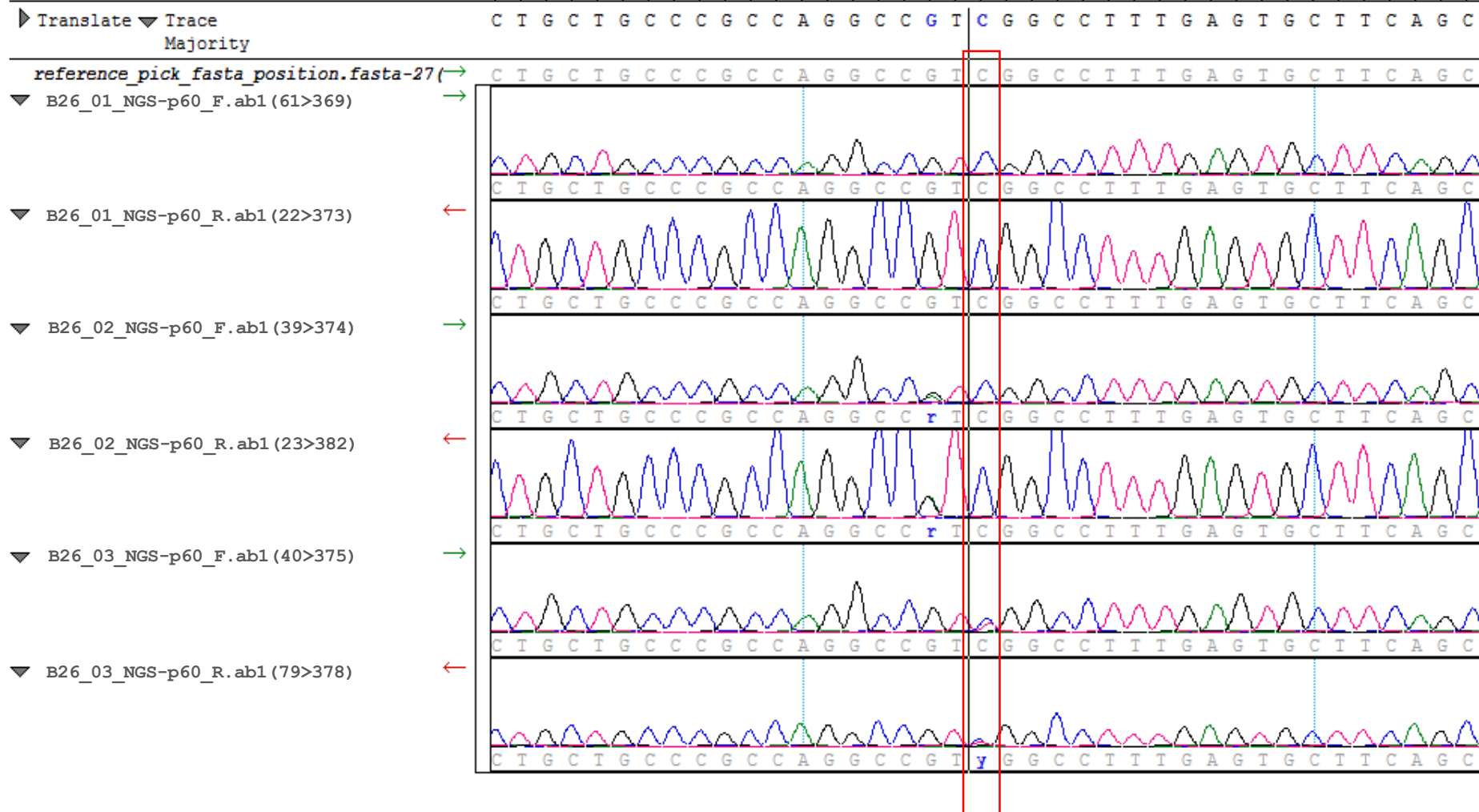
▼ B29_02_NGS-p68_F.ab1 (29>371) →
C C T T C T T C C C G T C C T G T A G C A C C A C T G C C G G A A C T G T G G C C A C A T C T T

▼ B29_02_NGS-p68_R.ab1 (50>369) ←
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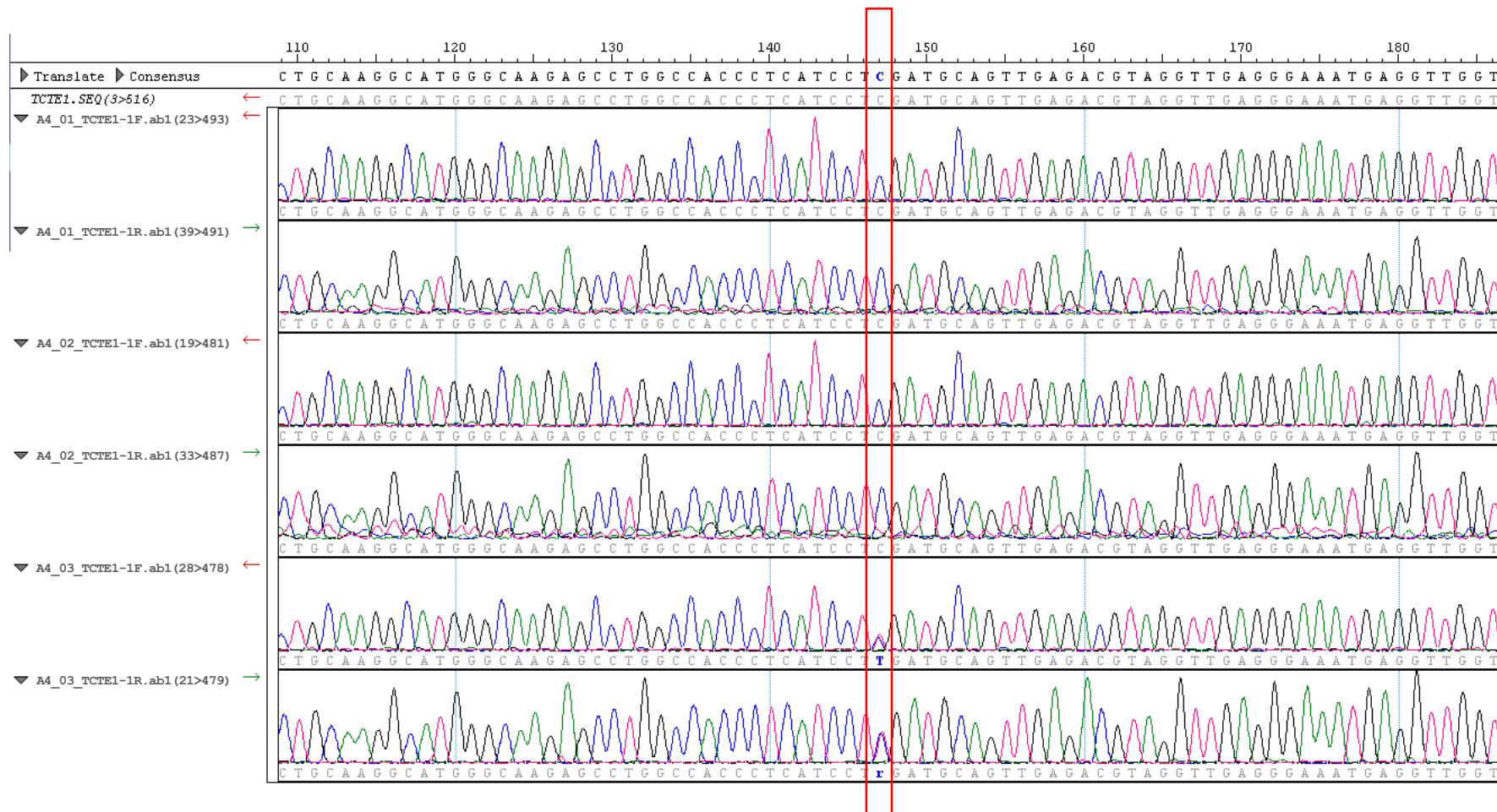
▼ B29_03_NGS-p68_F.ab1 (31>373) →
C C T T C T T C C C G T C C T G T A G C A C C r C T G C C G G A A C T G T G G C C A C A T C T T

▼ B29_03_NGS-p68_R.ab1 (93>364) ←
C C T T C T T C C C G T C C T G T A G C A C C r C T G C C G G A A C T G T G G C C A C A T C T T

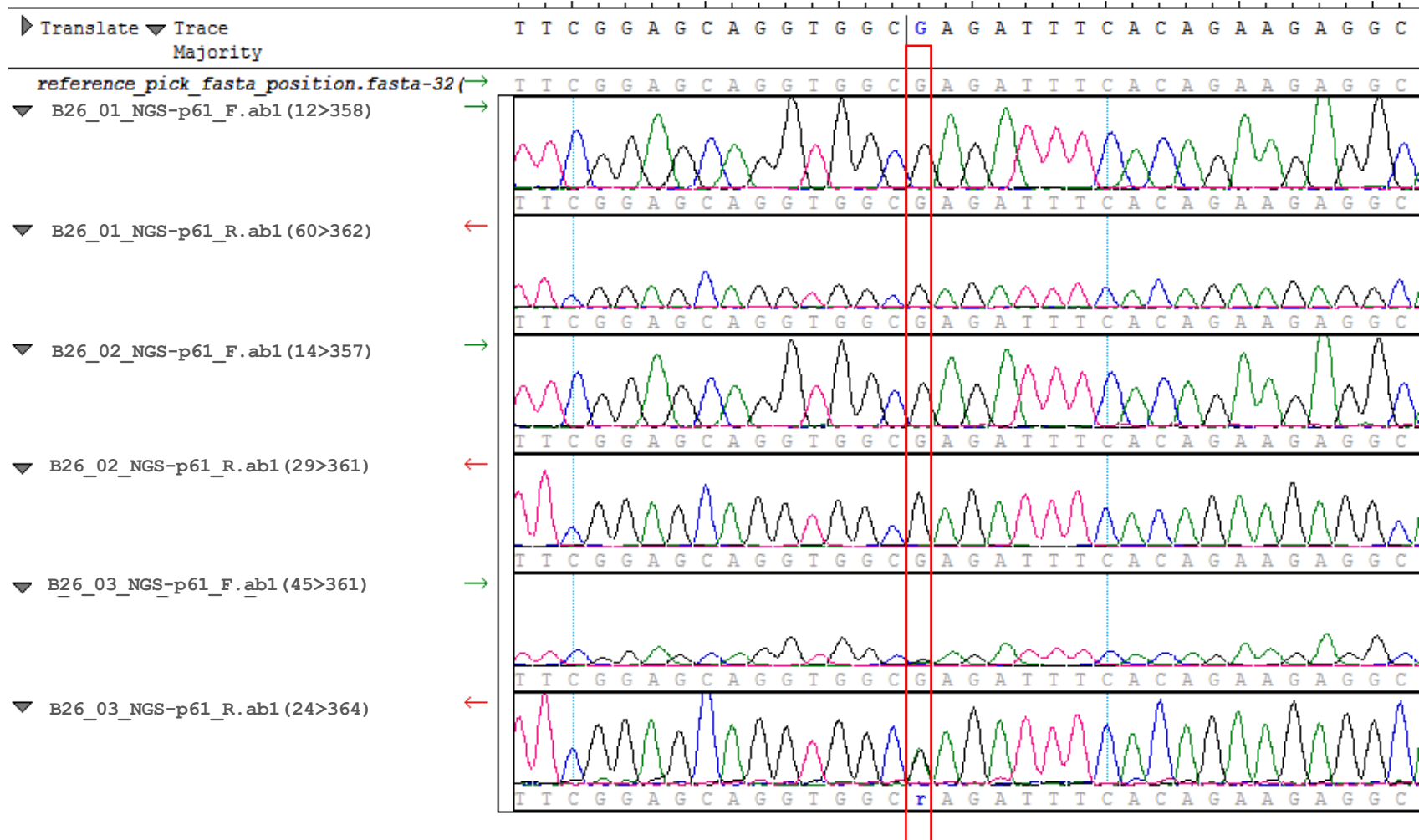
ATXN1



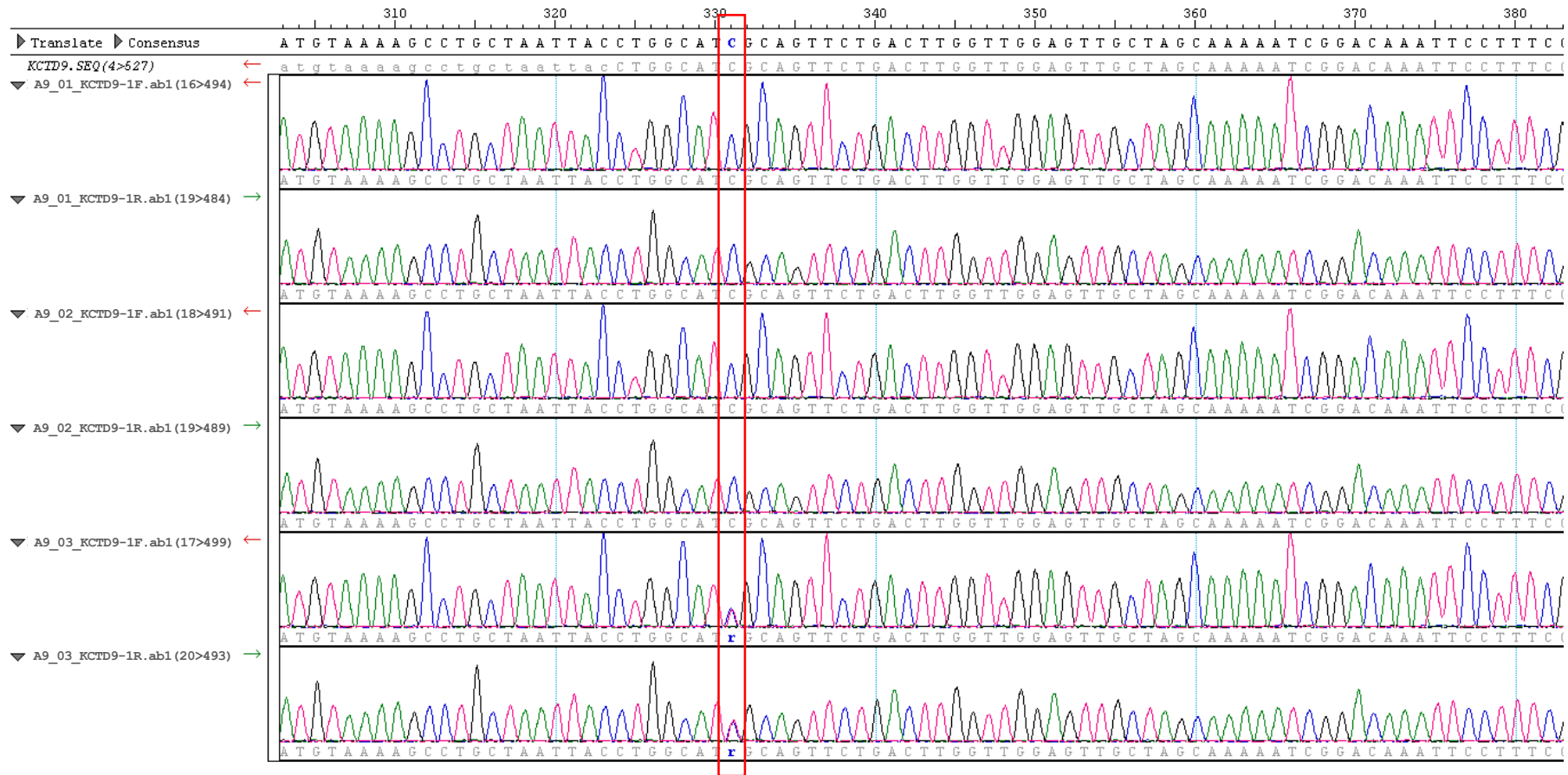
TCTE1



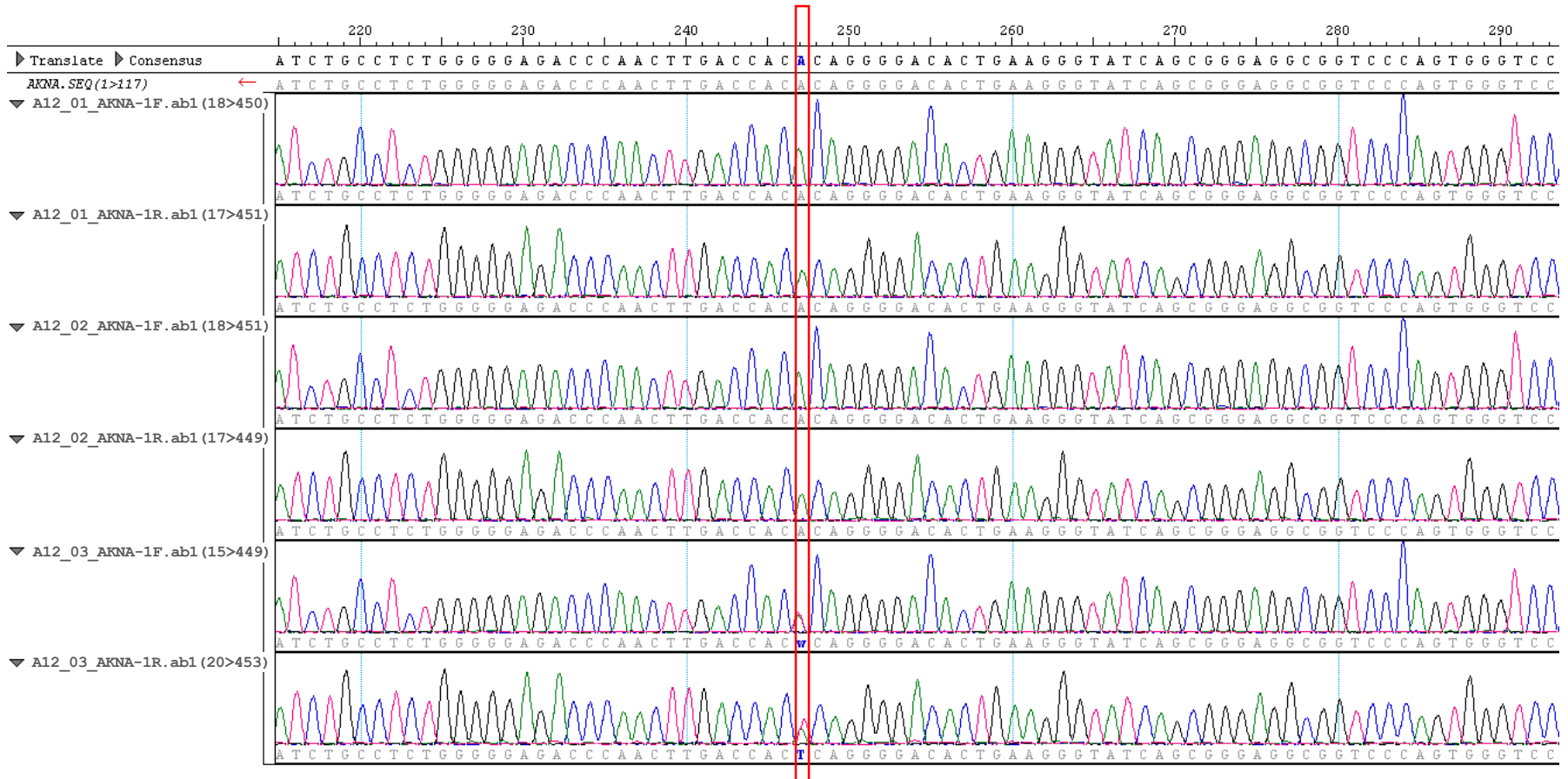
MTUS1



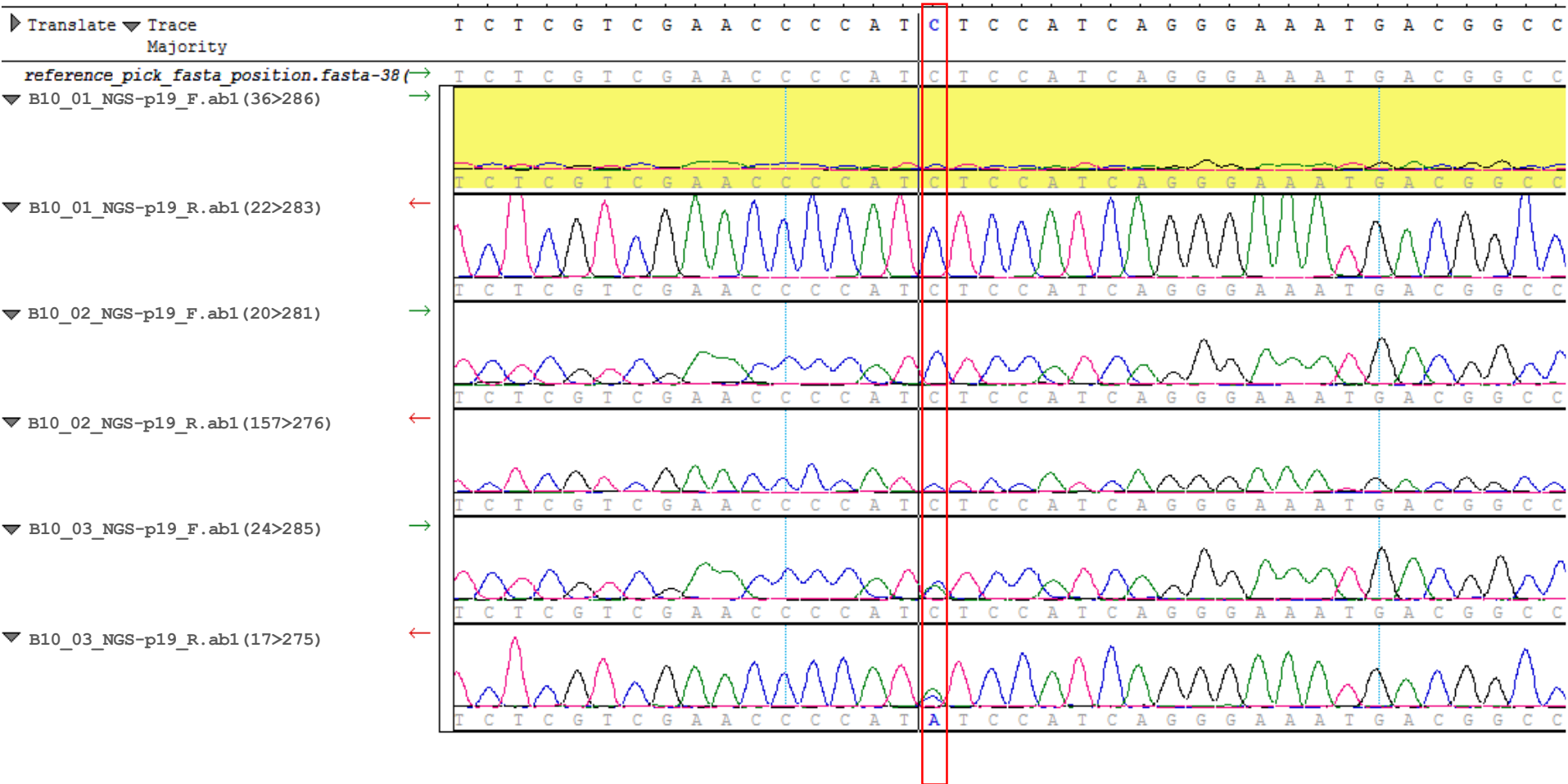
KCTD9



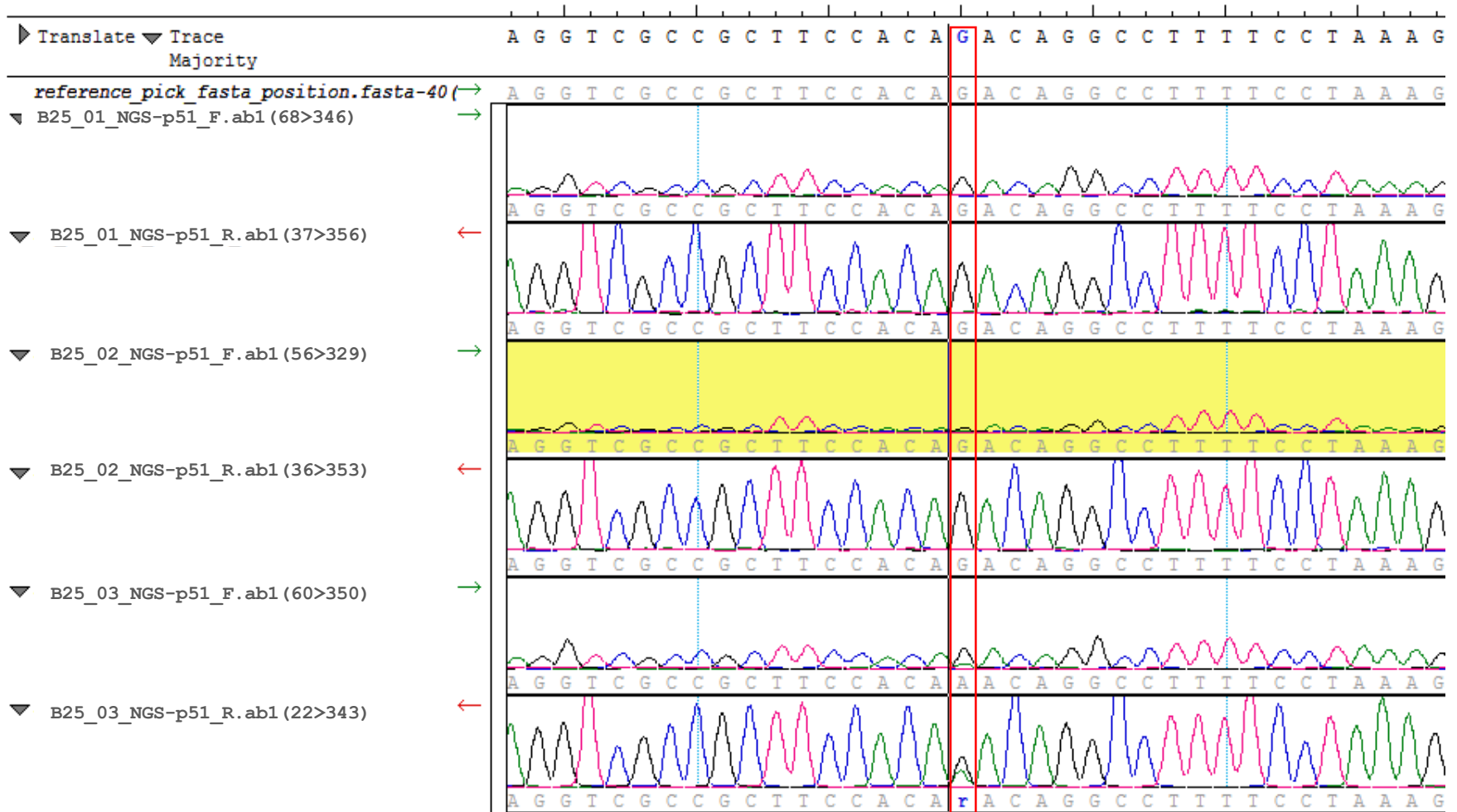
AKNA



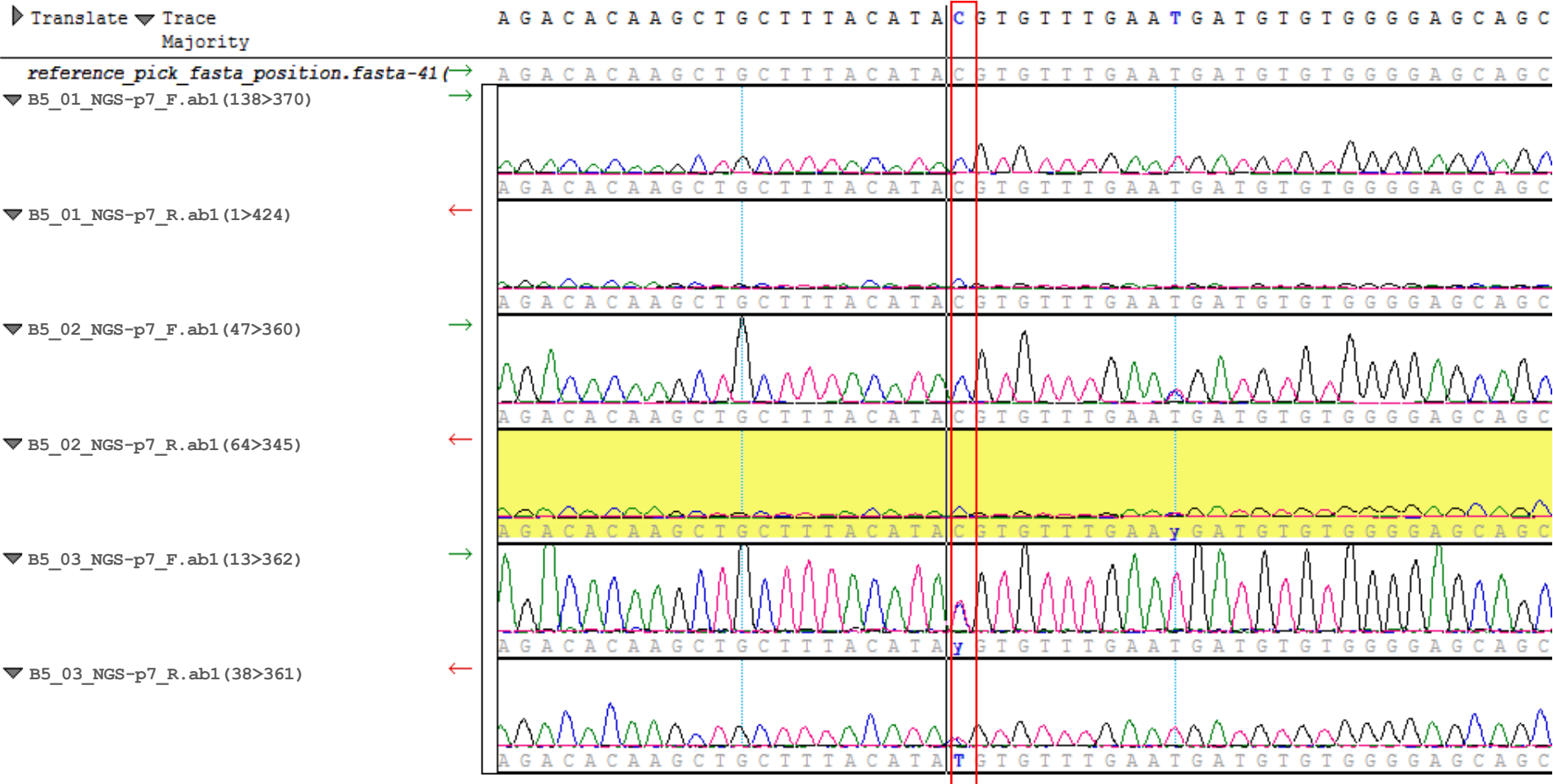
UBAC1



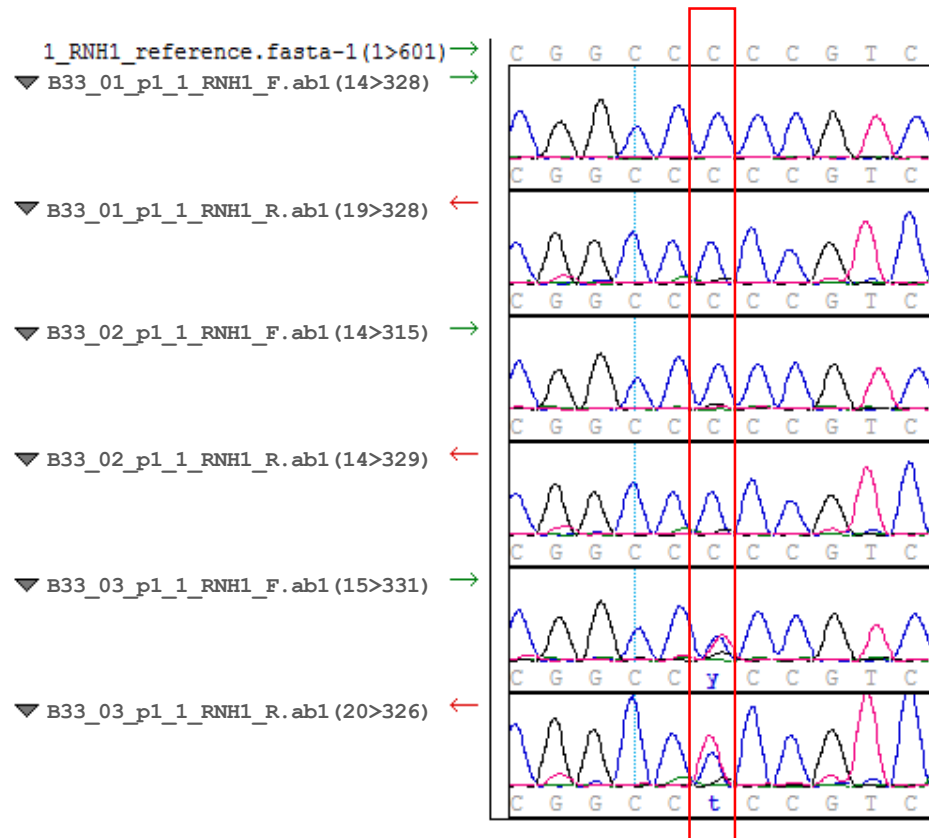
PITRM1-AS1



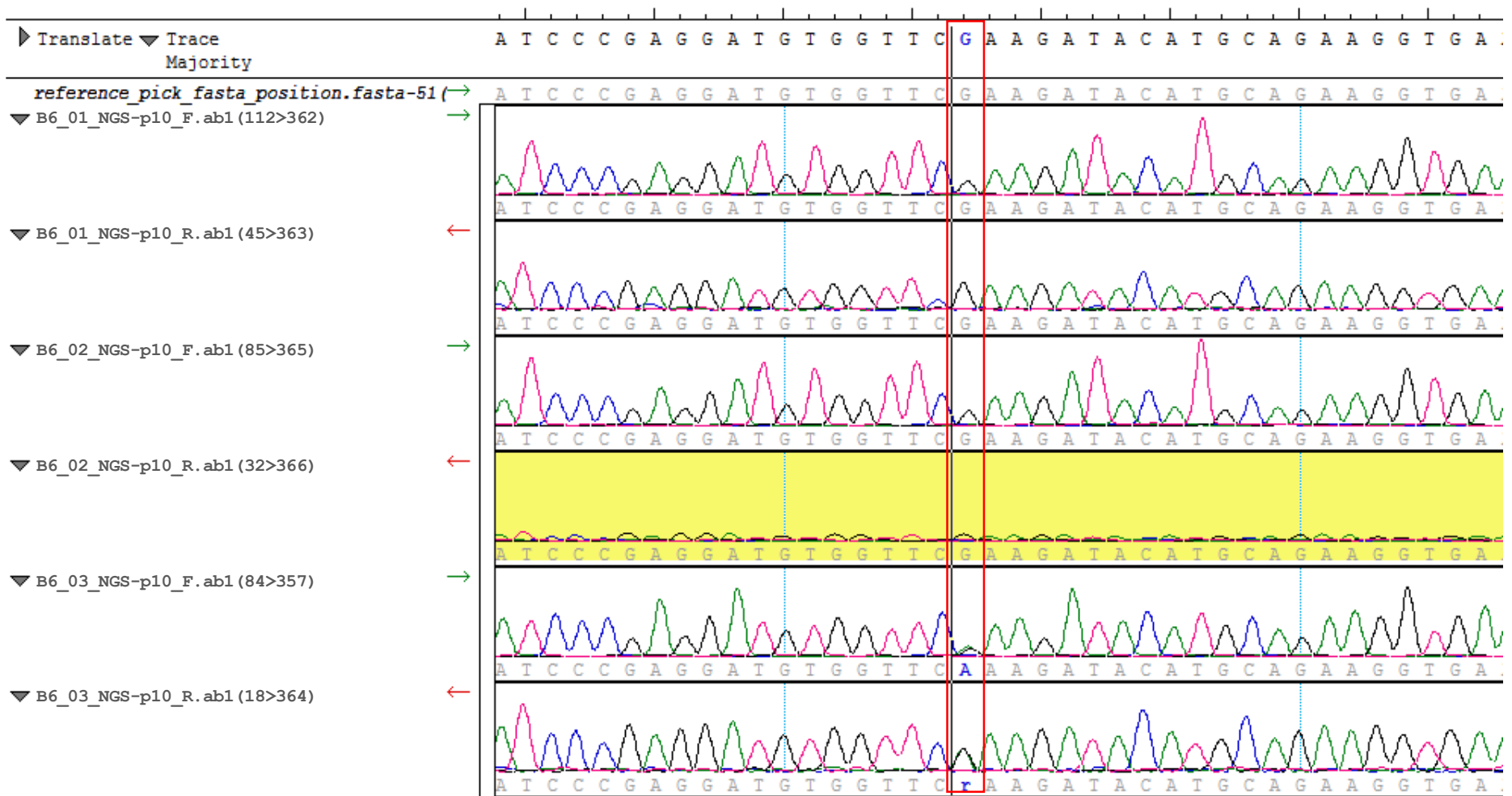
SFMBT2



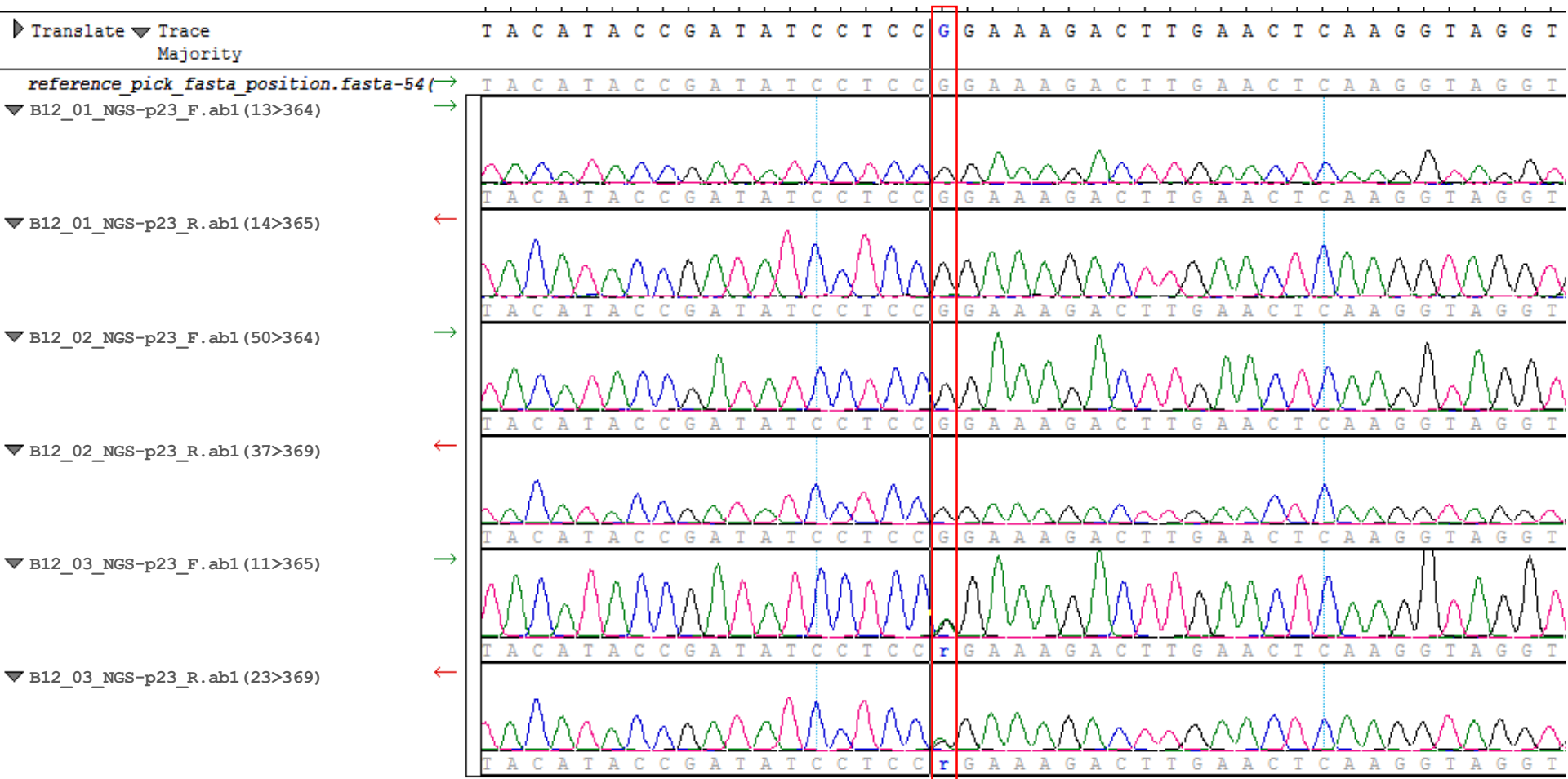
RNH1



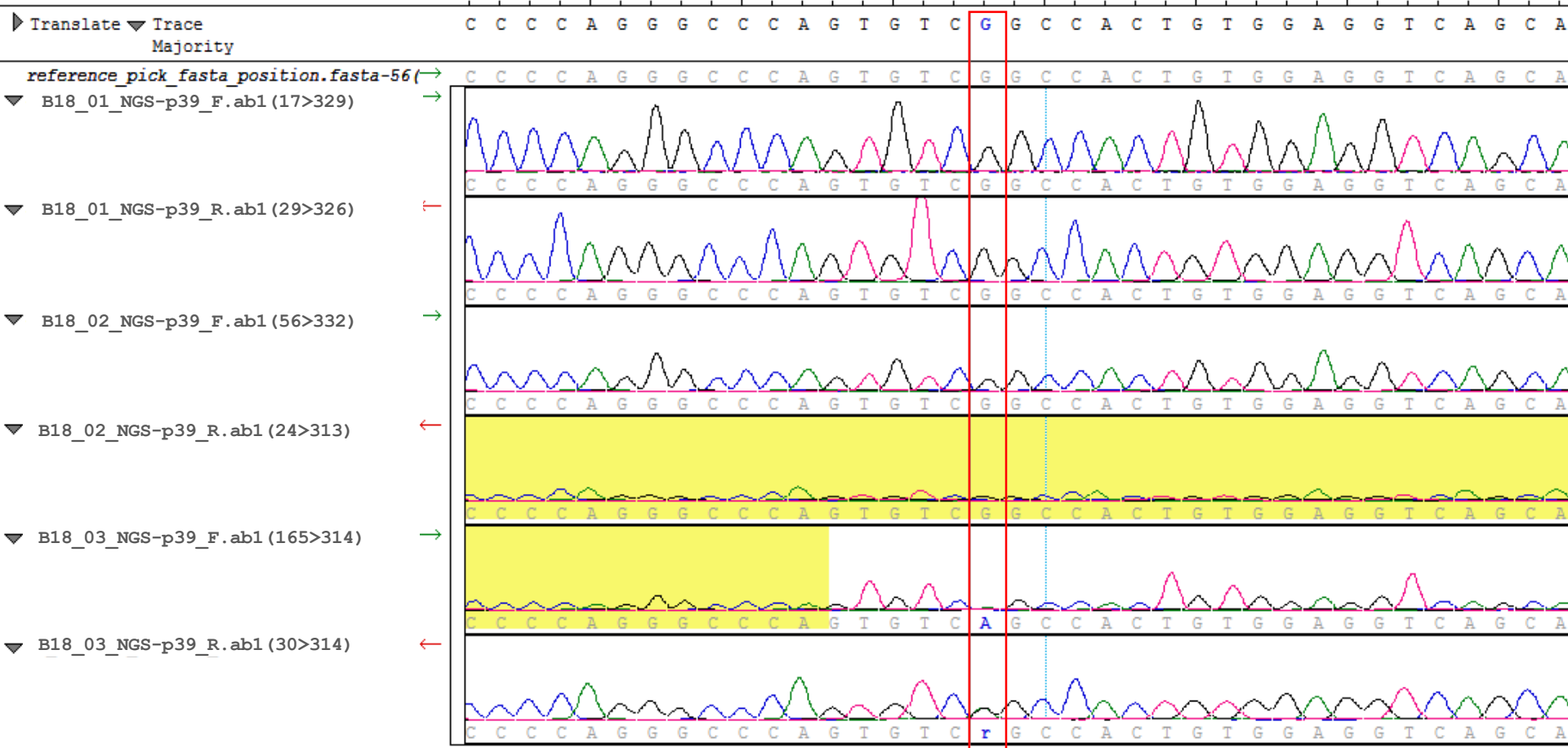
DTX1



RASGRP1



SYNM



TMEM8A

► Translate ▼ Trace
Majority

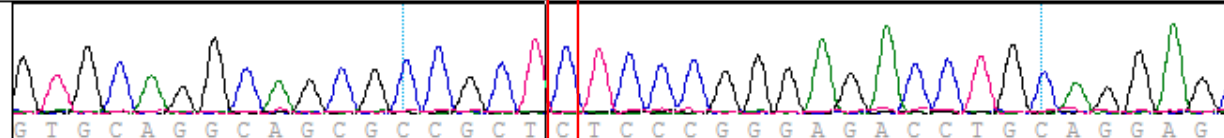
G T G C A G G C A G C G C C G C T C T C C C G G G A G A C C T G C A G G A G

reference_pick_fasta_position.fasta-57(→

G T G C A G G C A G C G C C G C T C T C C C G G G A G A C C T G C A G G A G

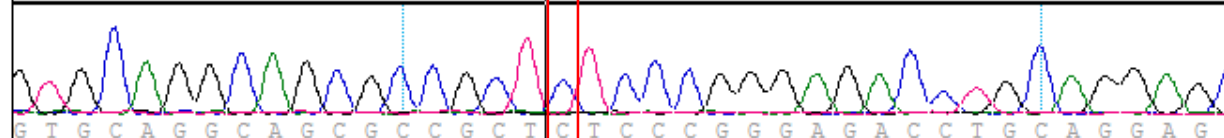
▼ B25_01_NGS-p52_F.ab1 (29>390)

→



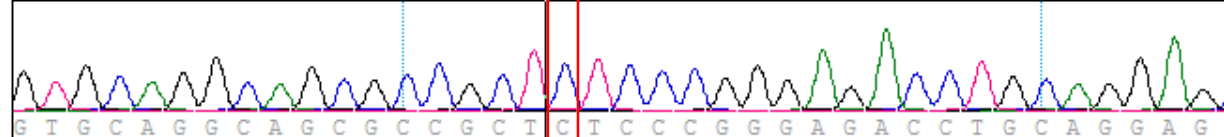
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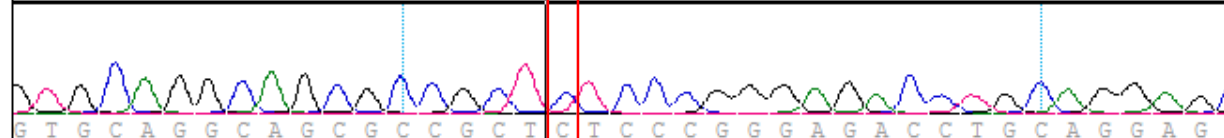
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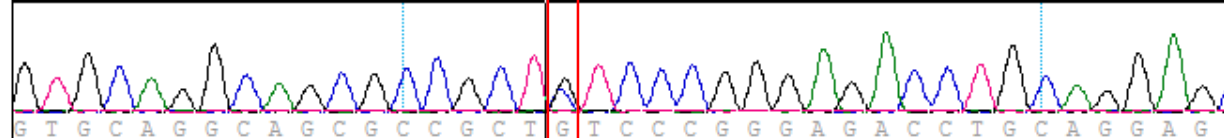
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←



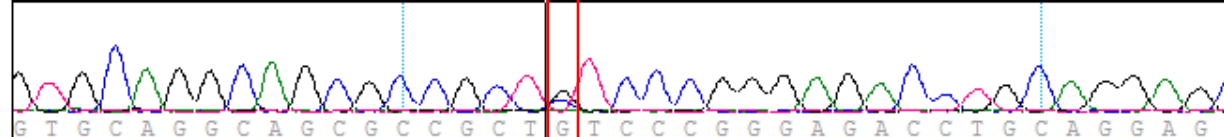
▼ B25_03_NGS-p52_F.ab1 (24>383)

→

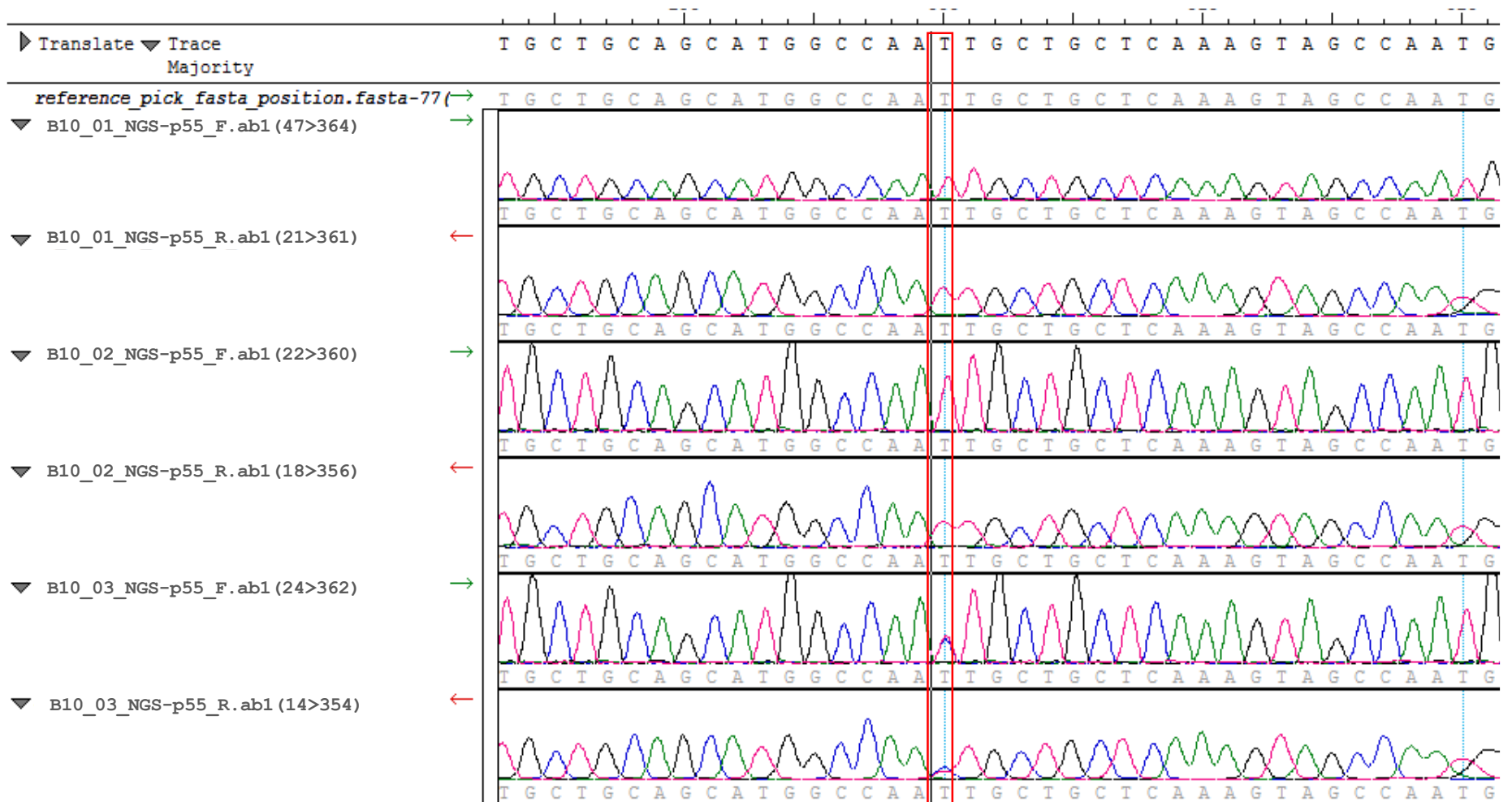


▼ B25_03_NGS-p52_R.ab1 (32>399)

←



ADCY7

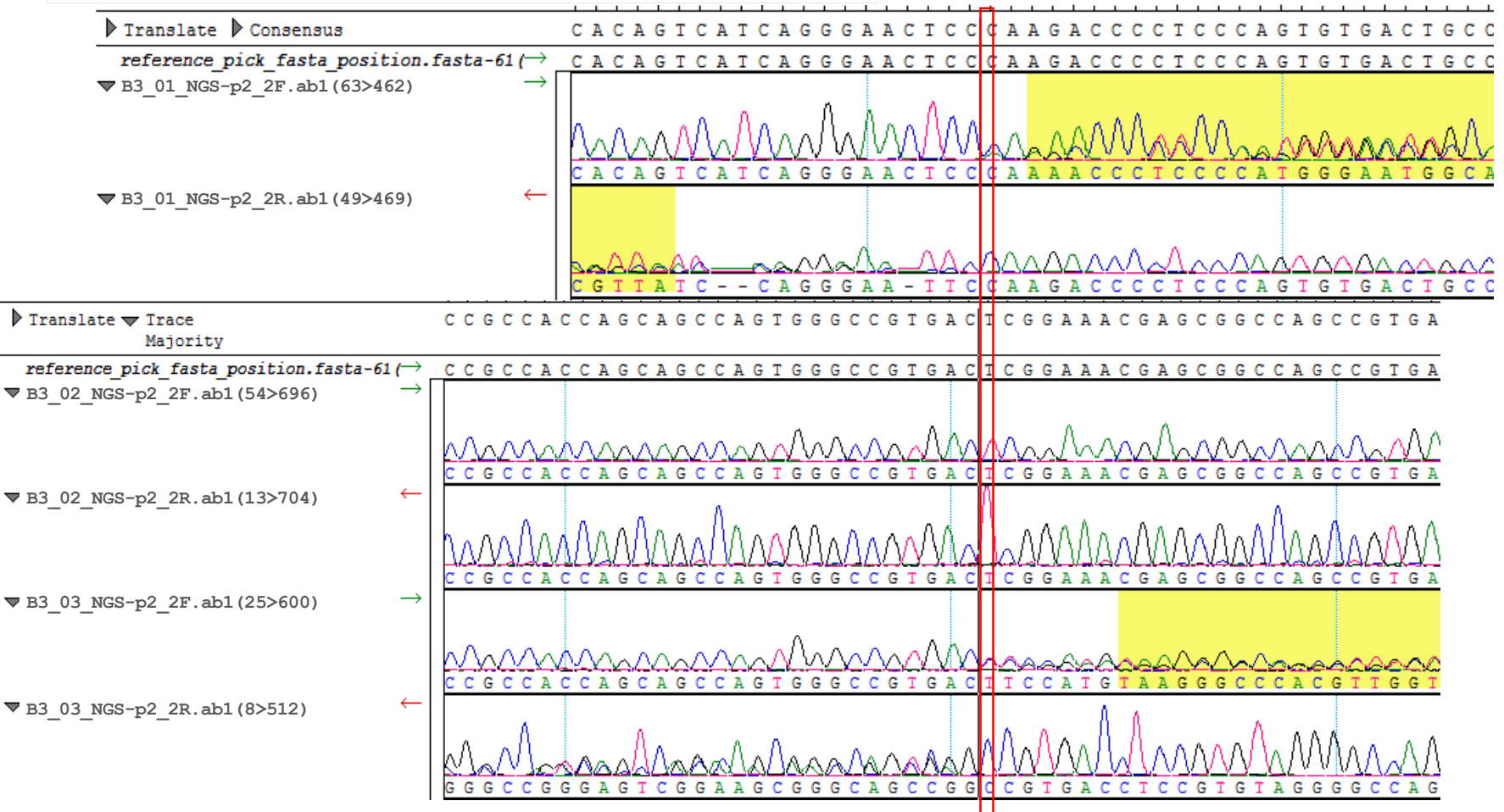


PIEZO1

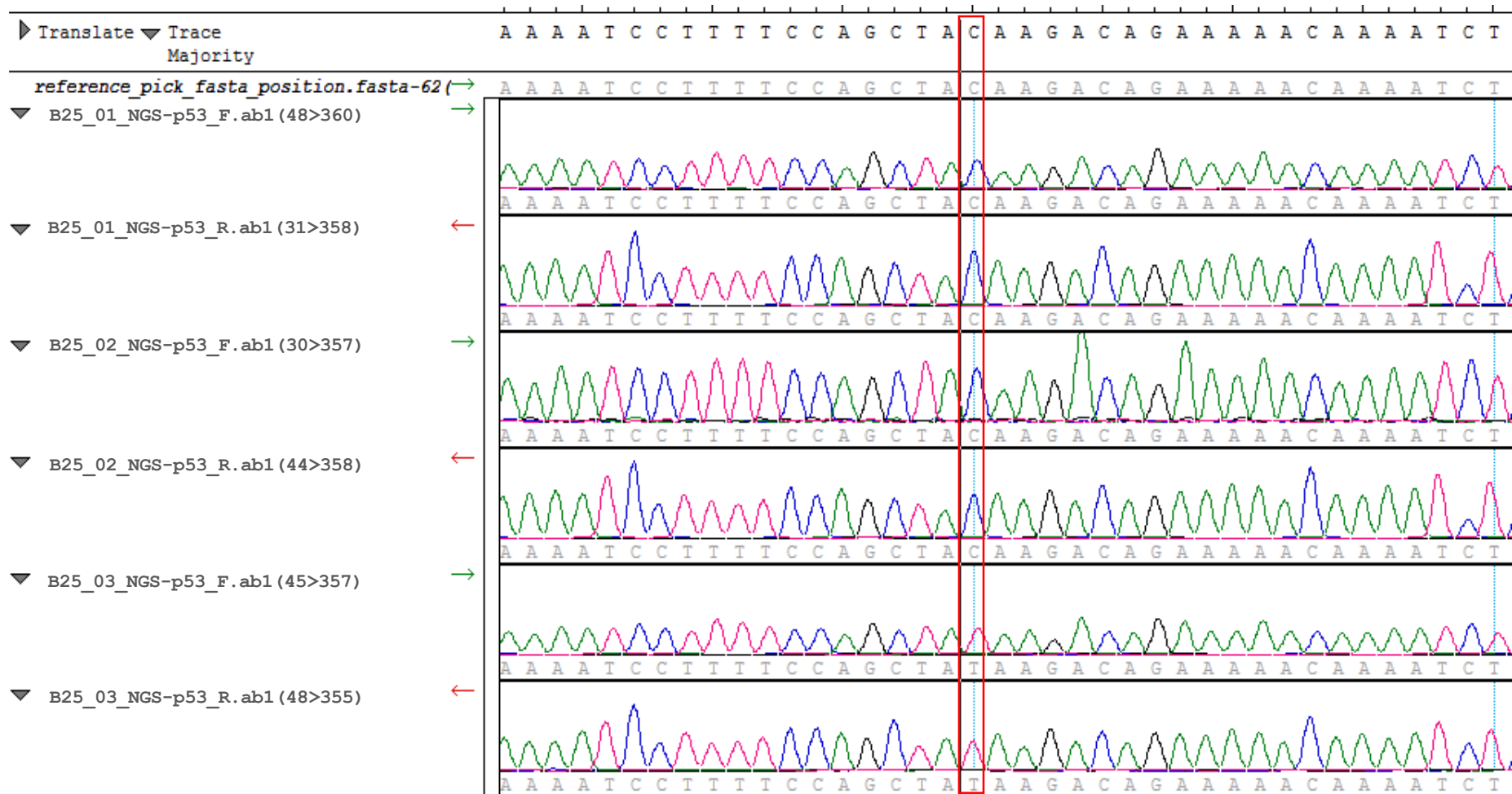
Comment

2_C2108-03 : heterozygosity (del GCCGTGACTCGGAAACGAGCGGCCA)

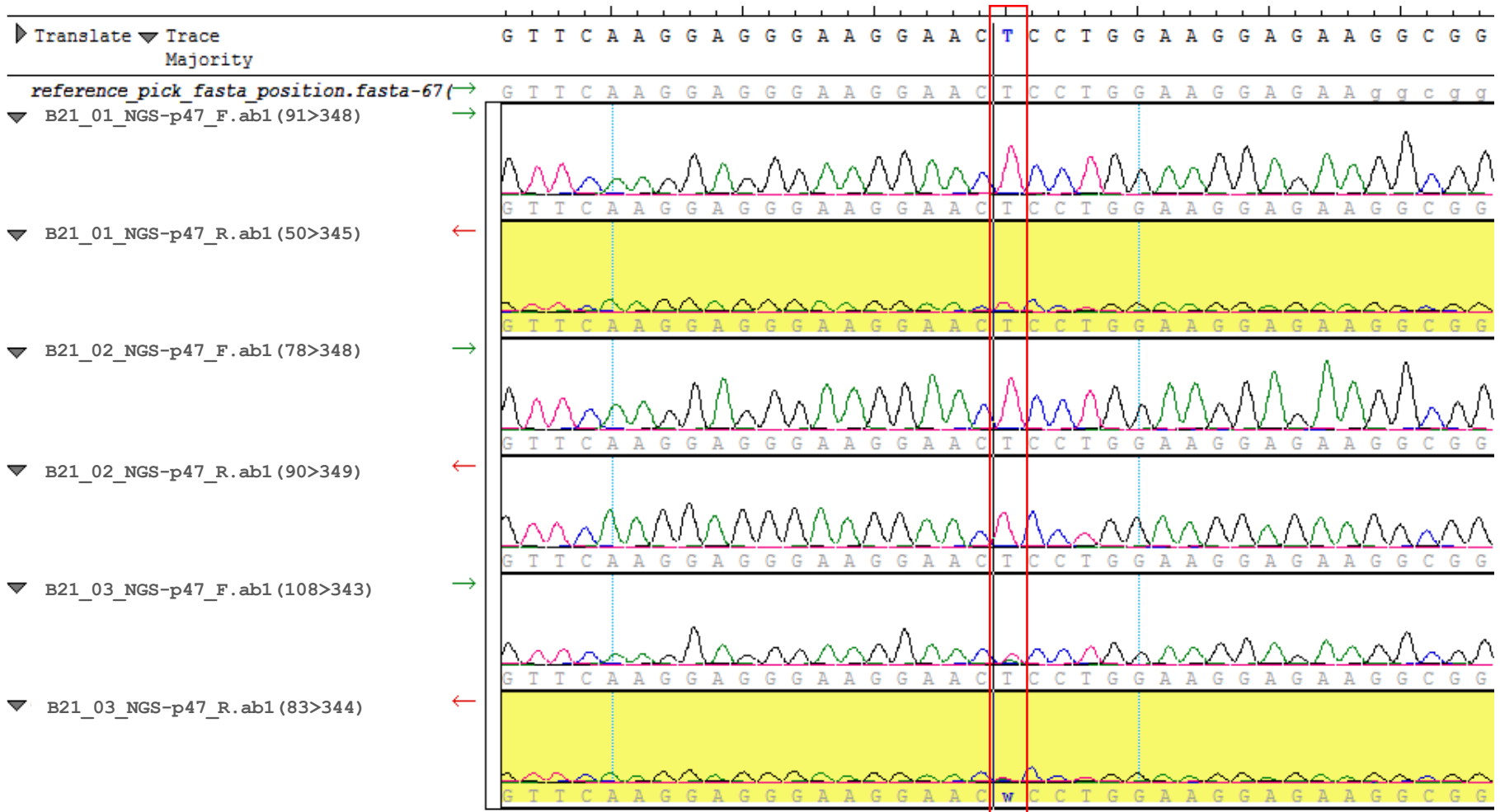
2_C2108-01 : chr16:88804927 : del C (other variant)



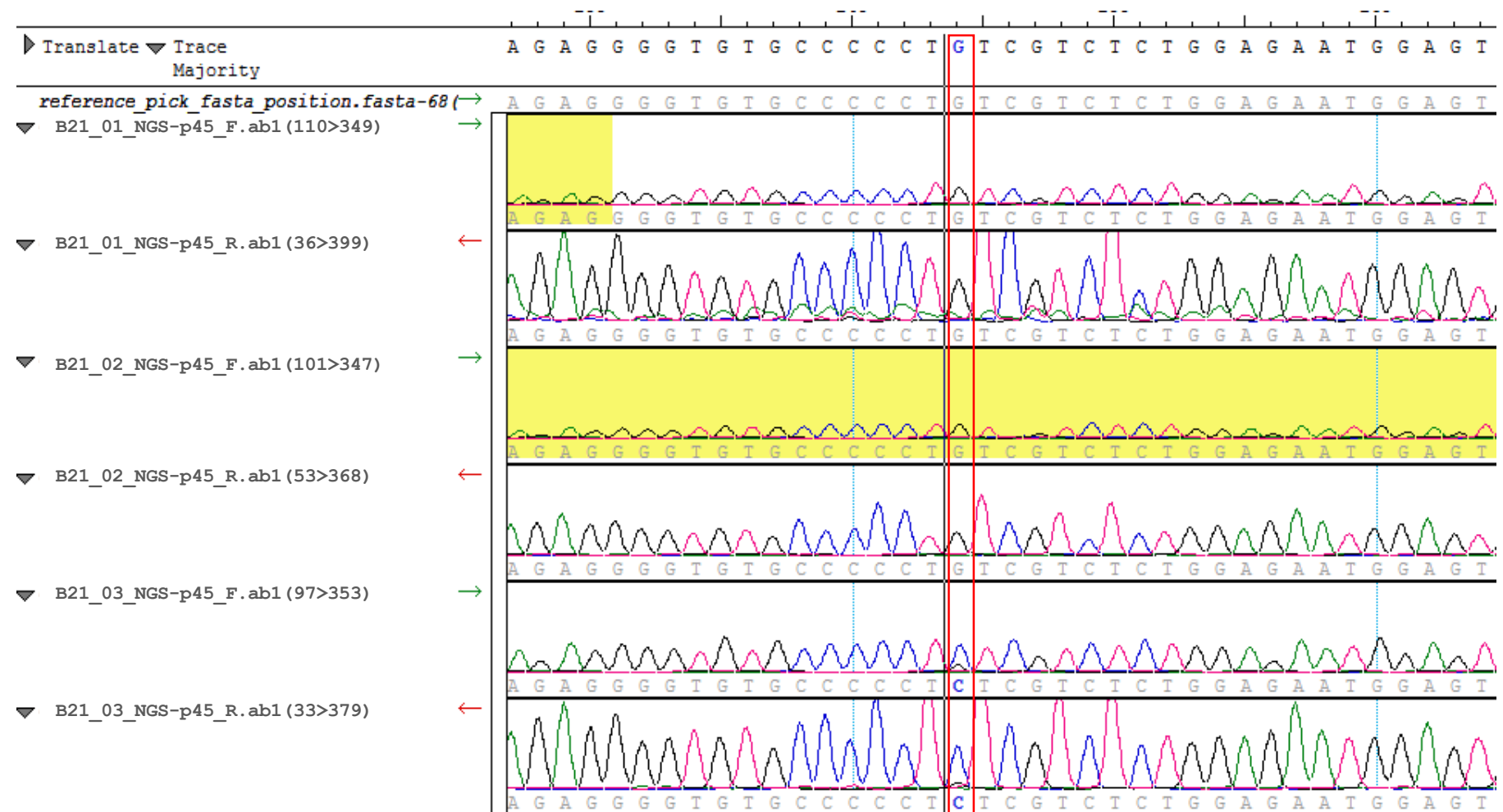
SNF8



GMIP

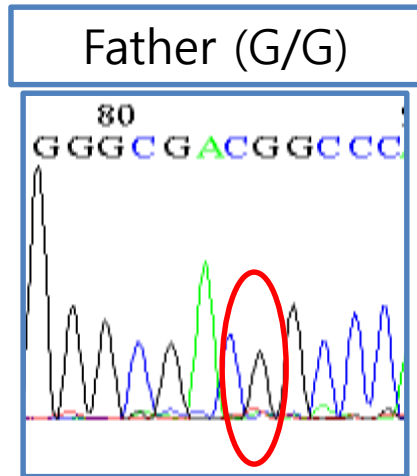


ANKRD27

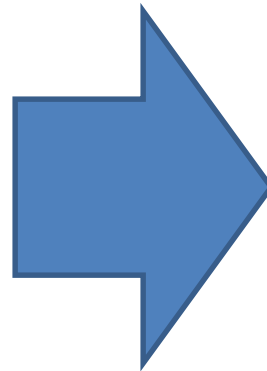
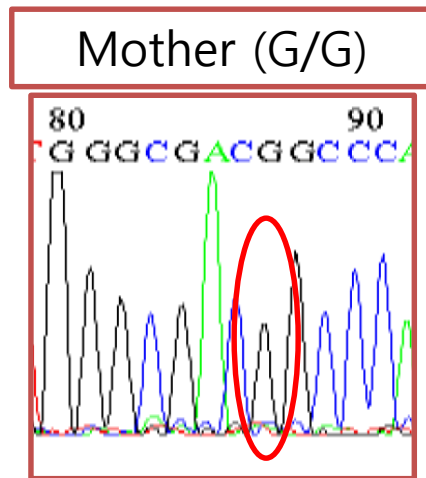


MYH14

A3_01

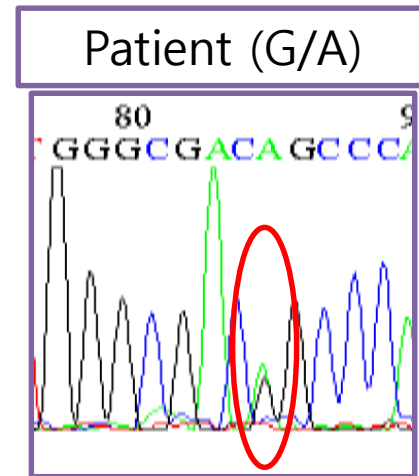


A3_02



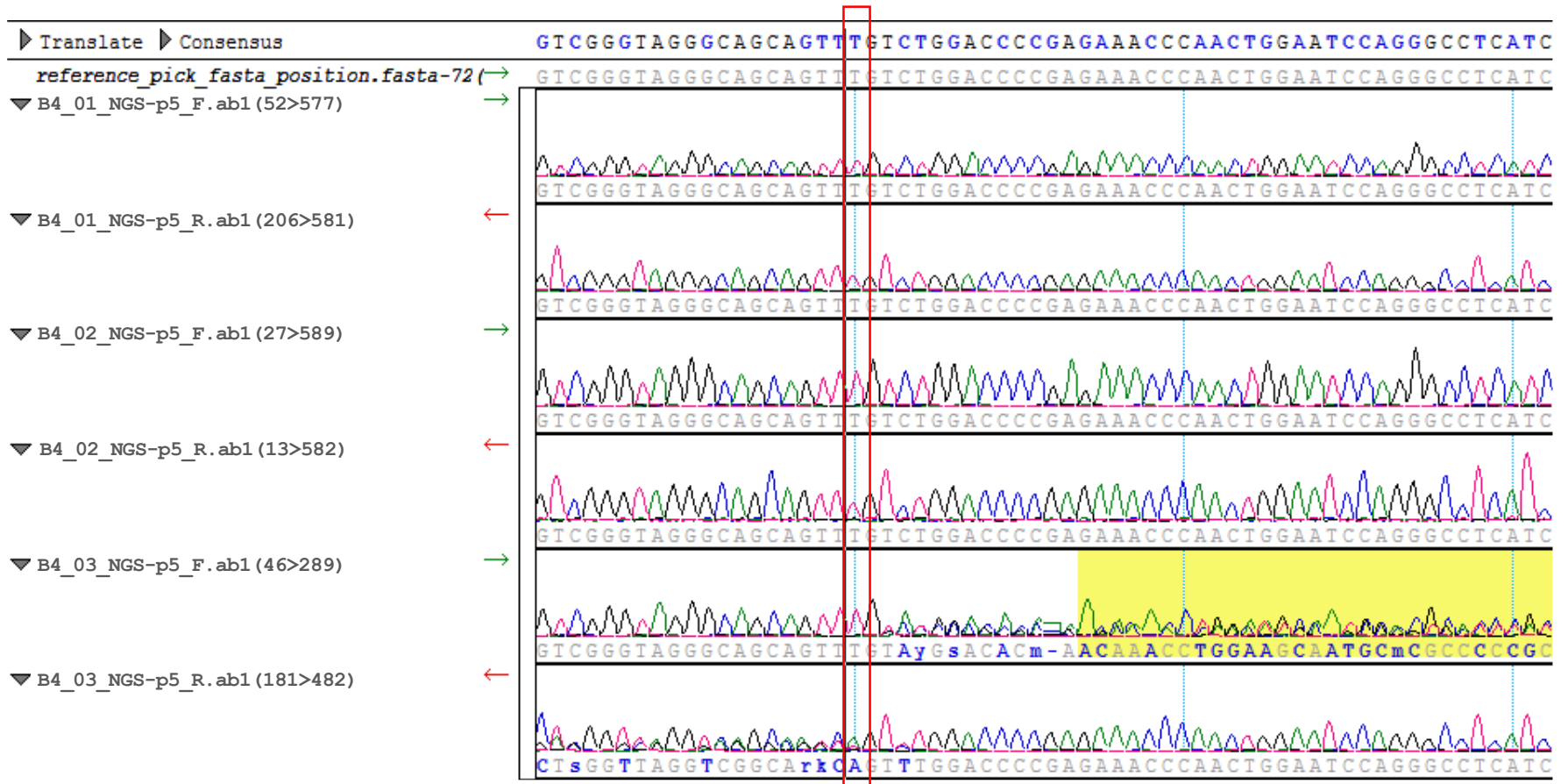
Patient (G/A)

A3_03

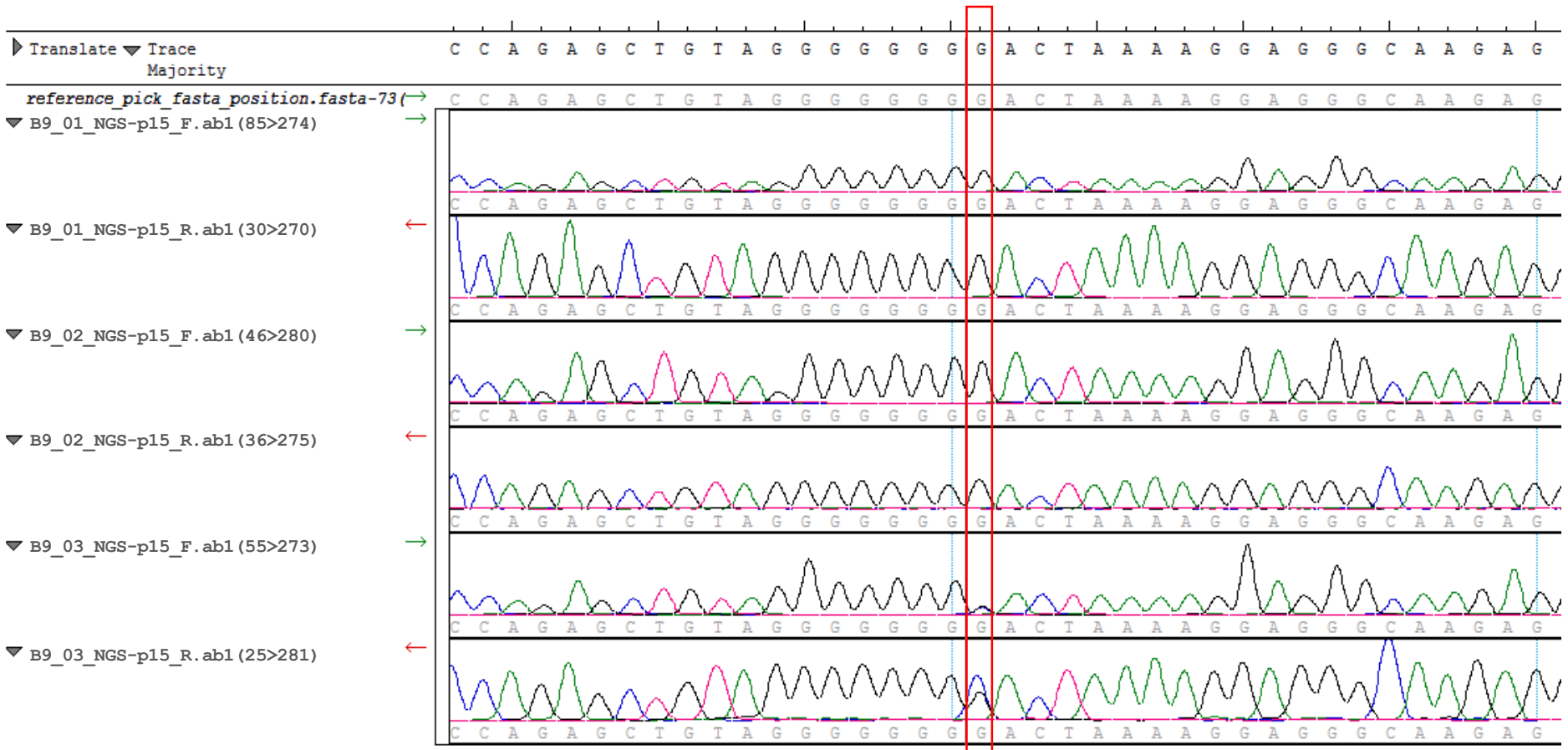


De novo mutation in MYH14 gene

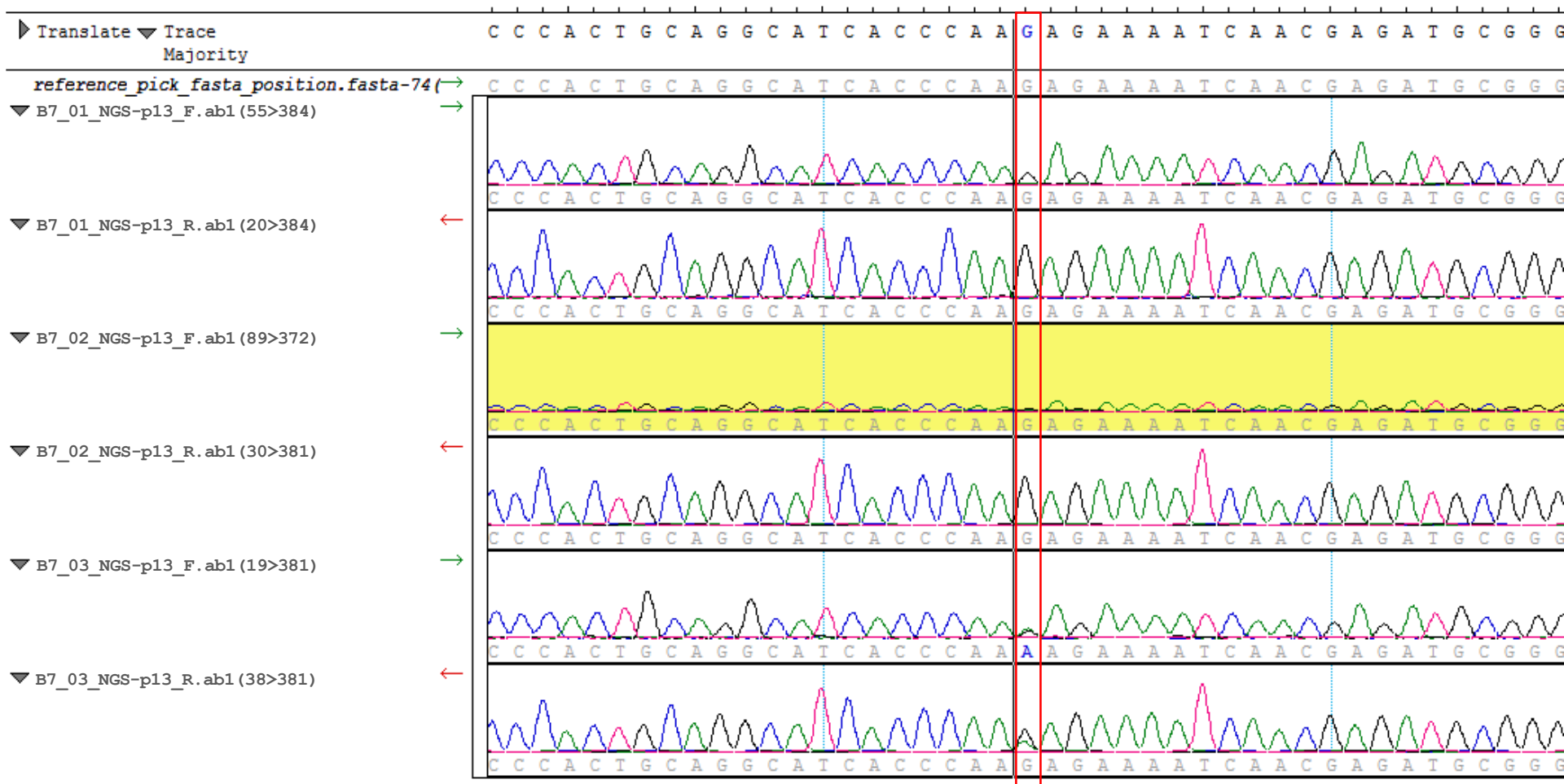
TFPT



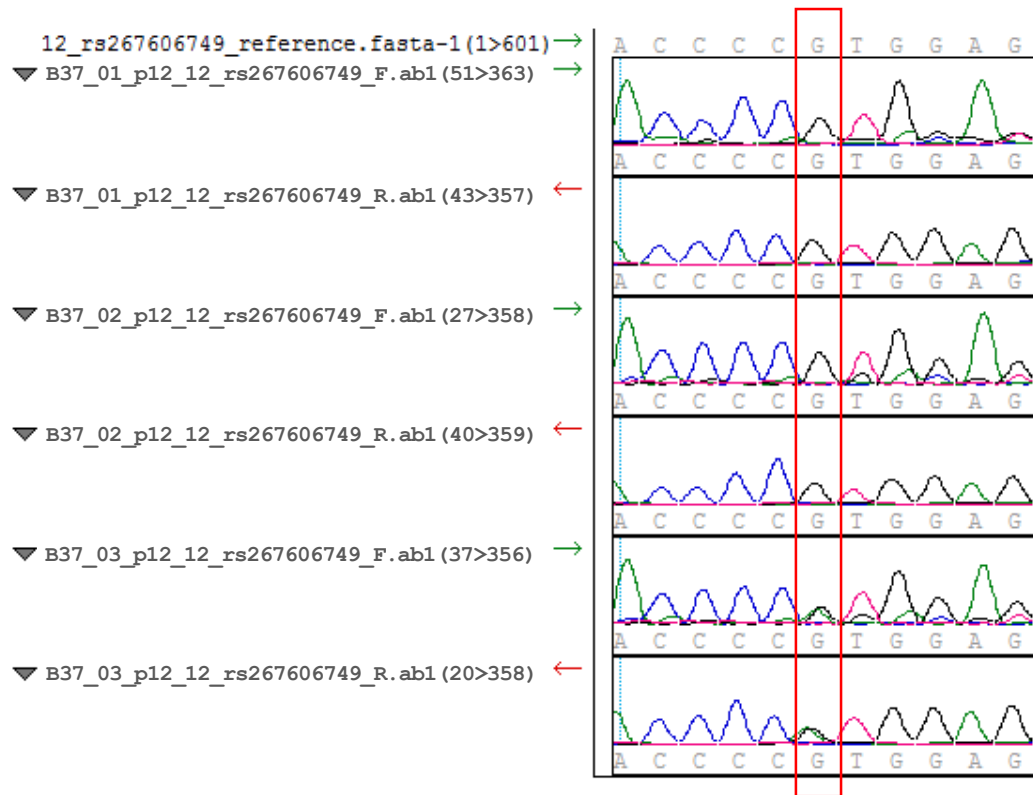
NOL4L



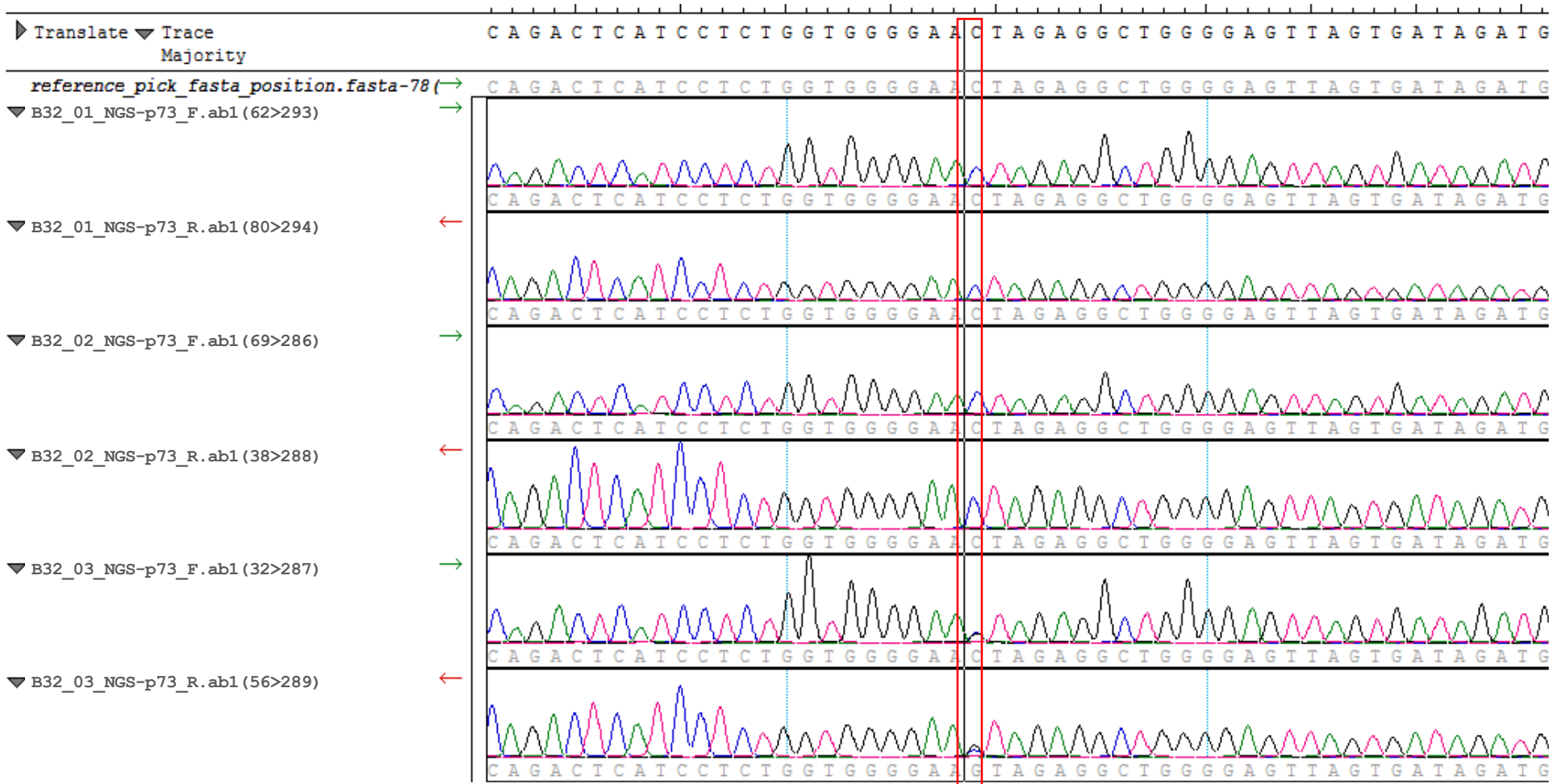
PPP1R16B



COL6A2



HMGXB4



MT-ND2

