

SnackVar Report

ID : CF8754900; Met1Thr

Description : Confirmation of Met1Thr in patient CF8754900

Reference sequence : NM_000492.4(CFTR).fasta

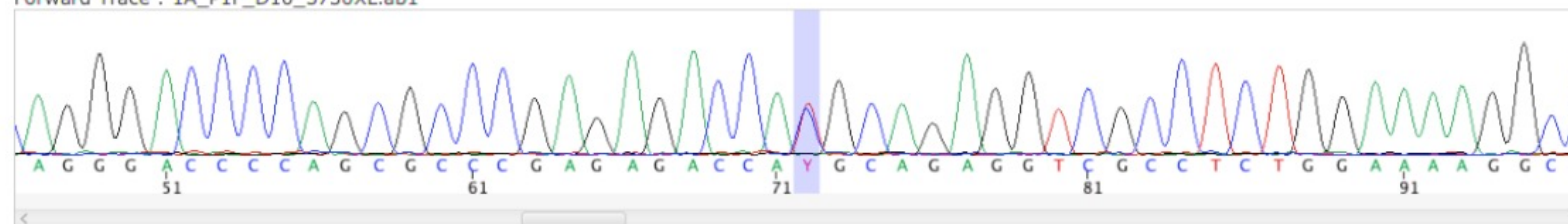
Variant 1

Description : c.2T>C, p.(Met1Thr), heterozygous

Alignment

11 21 31 41 51 61 71 81 91 101 111 121
gtctt tggcatt aggagctt gagcccagacggccct agcagggacccccagcgccccgagagacca TGCAGAGGT CGCCTCTGGAAAAGGCCAGCGTTGTCTCCAAACTTTTTTTCAGgt gaga
gtcttttggcatttaggagctt gagcccagacggccct agcagggacccccagcgccccgagagacca TGCAGAGGT CGCCTCTGGAAAAGGCCAGCGTTGTCTCCAAACTTTTTTTCAGGT GAGA
gtcttttggcatttaggagctt gagcccagacggccct agcagggacccccagcgccccgagagacca TGCAGAGGT CGCCTCTGGAAAAGGCCAGCGTTGTCTCCAAACTTTTTTTCAGGT GAGA

Forward Trace : 1A_P1F_D10_3730XL.ab1



Reverse Trace : 1A_P1R_H10_3730XL.ab1



Figure 1: SnackVar report for Met1Thr variant in CF8754900

SnackVar Report

ID : CF1782680; Met1Thr

Description : Confirmation of Met1Thr variant in patient CF1782680

Reference sequence : NM_000492.4(CFTR).fasta

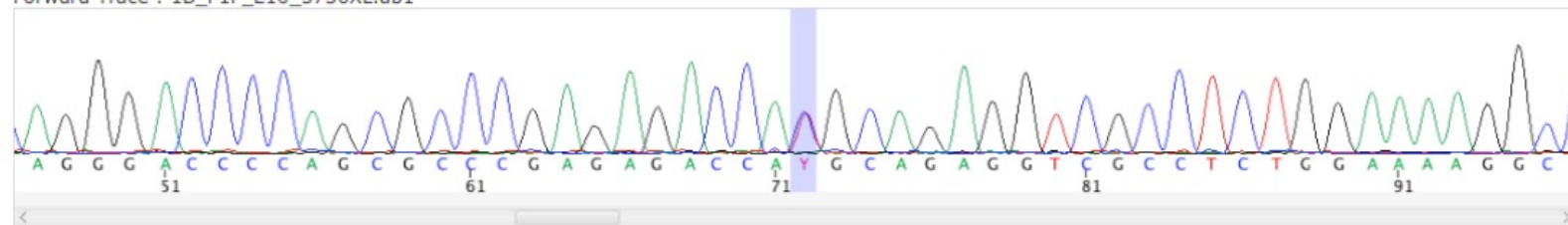
Variant 1

Description : c.2T>C, p.(Met1Thr), hetero

Alignment



Forward Trace : 1B_P1F_E10_3730XL.ab1



Reverse Trace : 1B_P1R_A11_3730XL.ab1



Figure 2: SnackVar report for Met1Thr variant in CF1782680

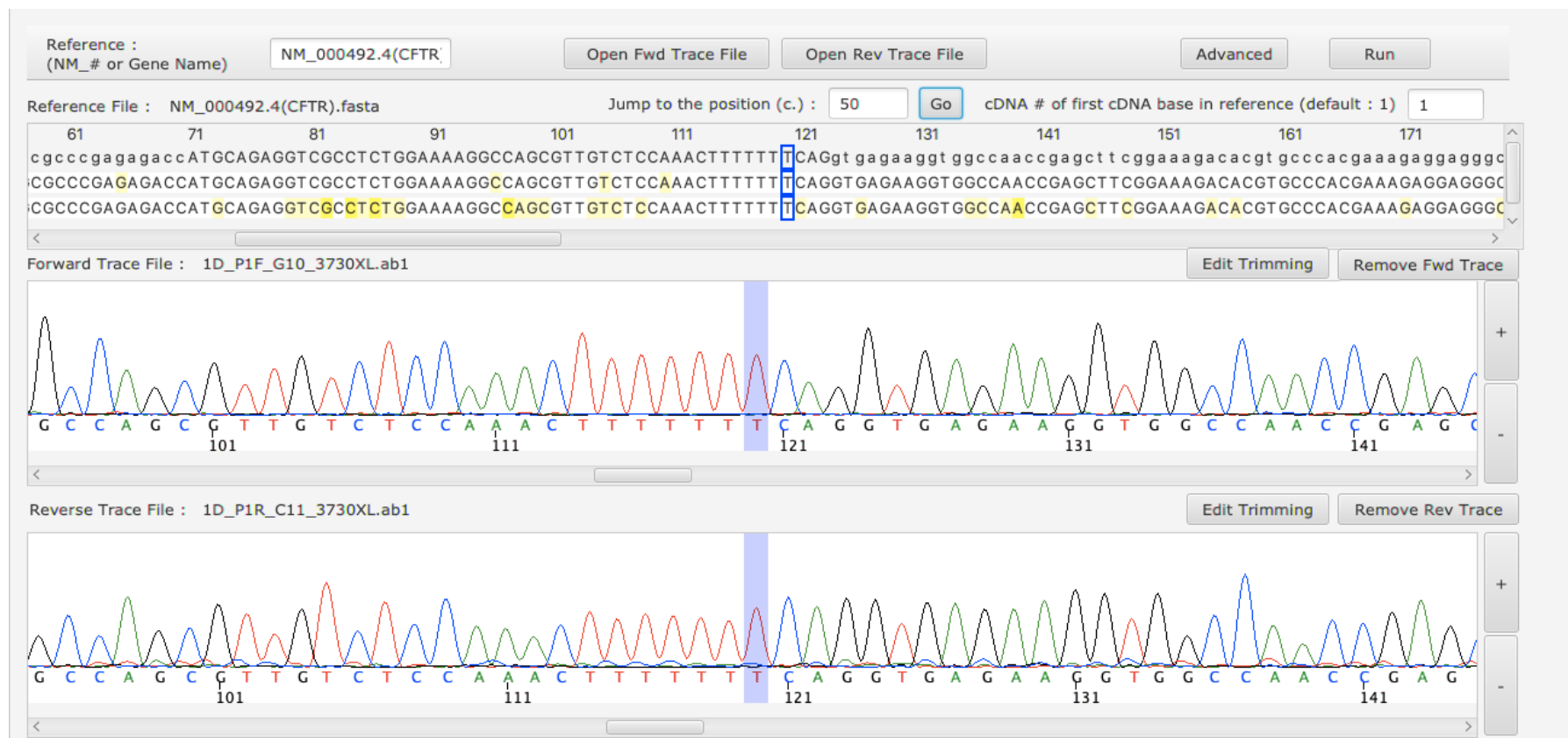


Figure 3: SnackVar report for CF3594271 at p.Phe17SerfsX10 position



Figure 4: SnackVar report for CF7930867 at p.Phe17SerfsX10 position

SnackVar Report

ID : CF4062212; p.Leu218Ter

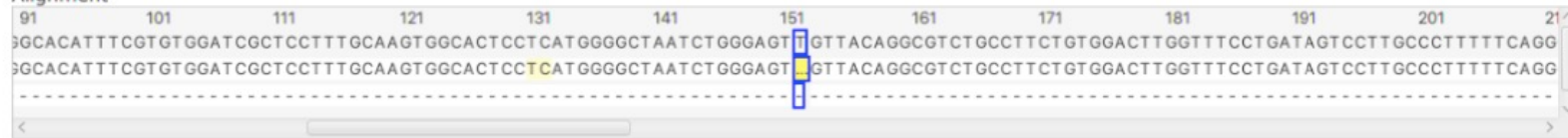
Description : Confirmation of p.Leu218Ter in sample CF4062212

Reference sequence : NM_000492.4(CFTR).fasta

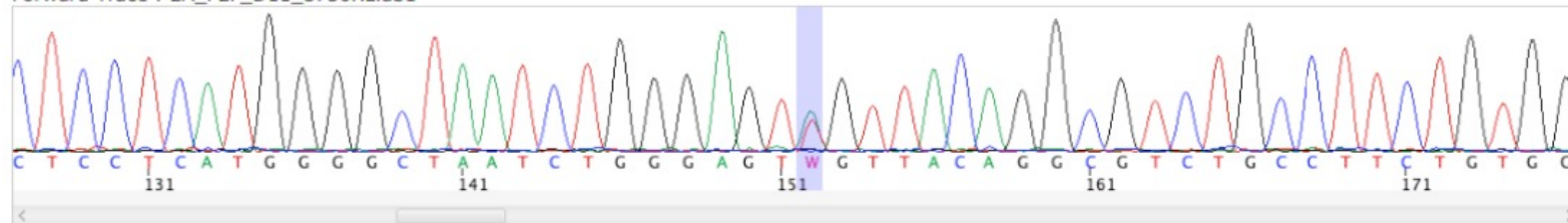
Variant 1

Description : c.653T>A, p.(Leu218*), hetero

Alignment



Forward Trace : 2A_P2F_D11_3730XL.ab1



Reverse Trace : 2A_P2R_E11_3730XL.ab1

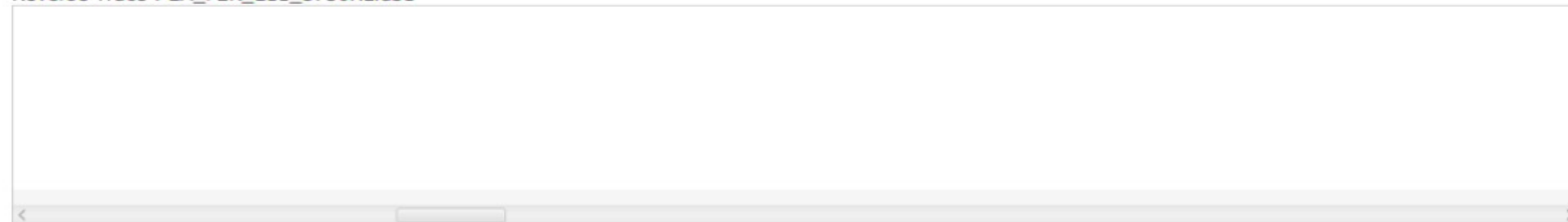


Figure 5: SnackVar report for p.Leu218Ter variant in CF4062212



Figure 6: SnackVar report for p.Arg303AlafsTer16 position in CF4062212



Figure 7: SnackVar report for p.Arg104_Ala107del position in CF1323468

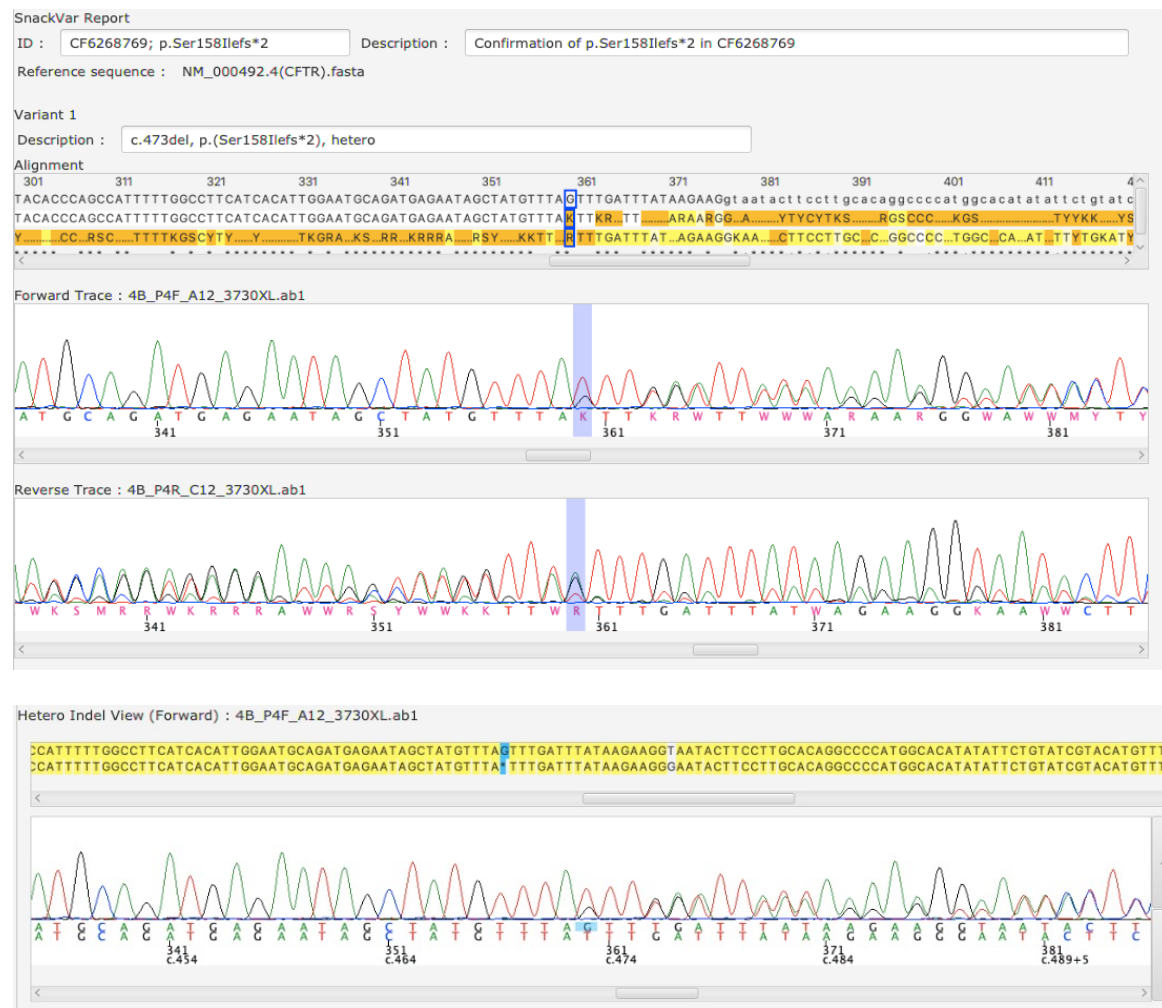


Figure 8: SnackVar report for p.Ser158IlefsTer2 variant in CF6268769



Figure 9: SnackVar report for p.His856SerfsTer5 position in CF4062212



Figure 10: SnackVar report for p.His856SerfsTer5 position in CF3239825



Figure 11: SnackVar report for p.Ser1297LeufsTer31 position in CF6188367

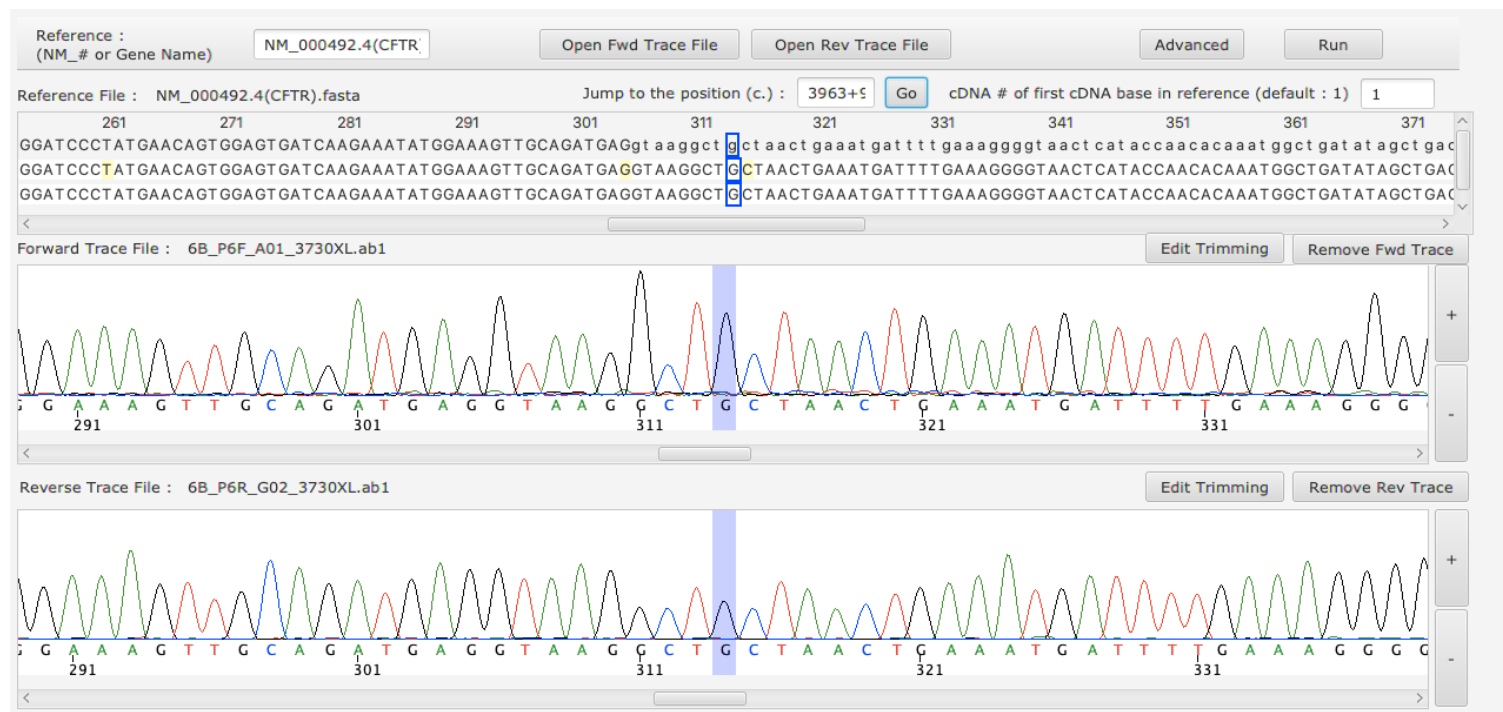


Figure 12: SnackVar report for c.3963+9G>C position in CF3512286



Figure 13: SnackVar report for the c.3963+9G>C position in CF3594271

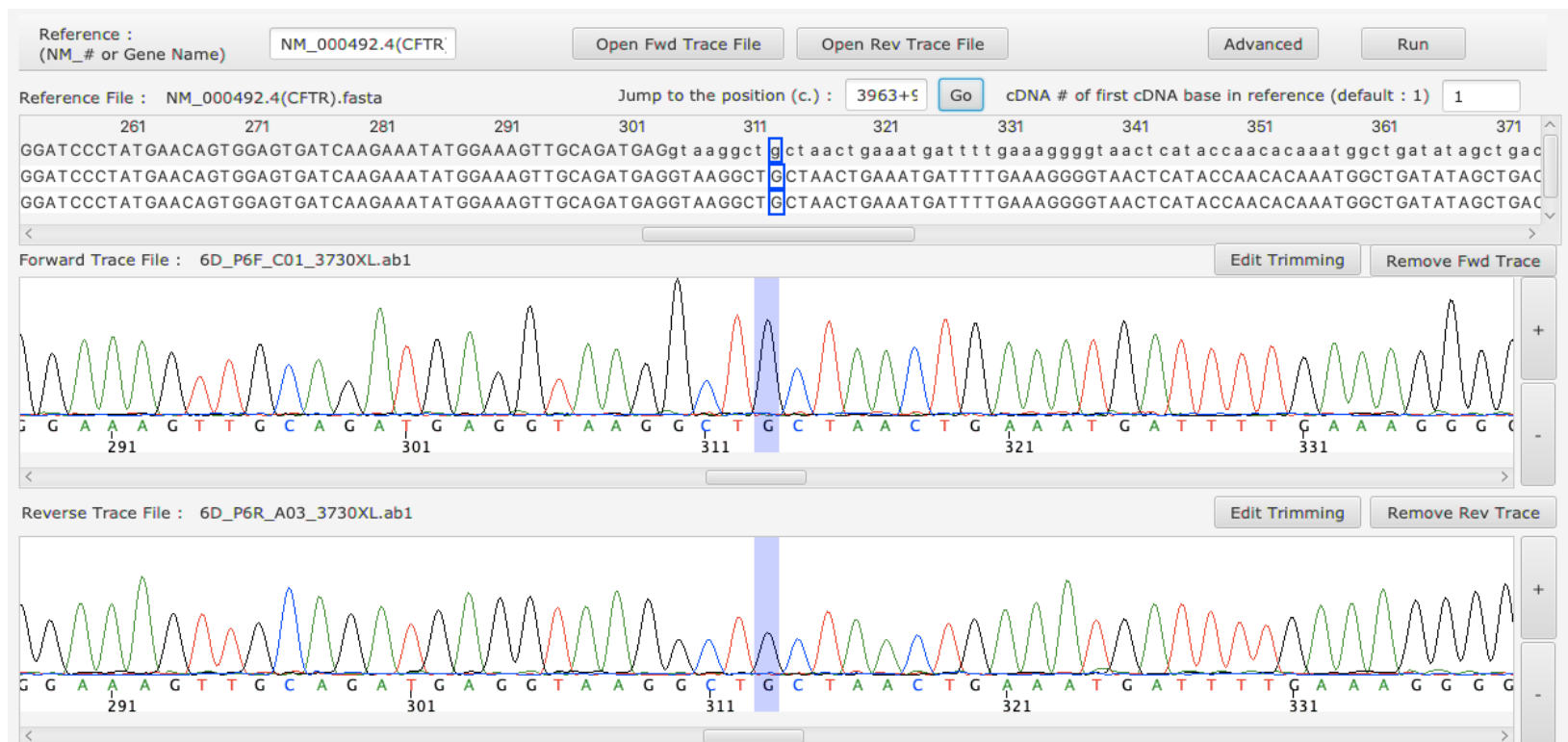


Figure 14: SnackVar report for the c.3963+9G>C position in CF1697504

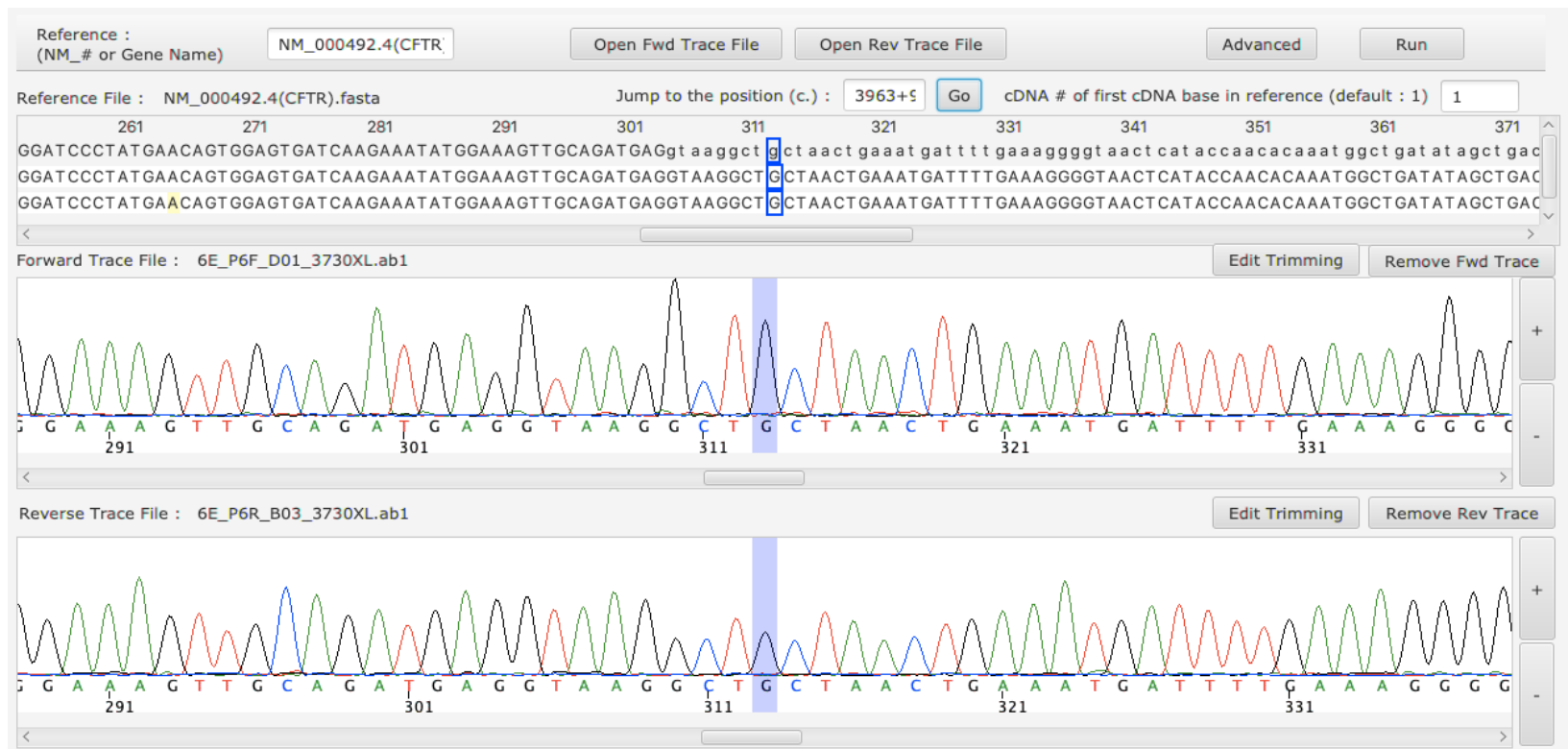


Figure 15: SnackVar report for the c.3963+9G>C position in CA0144930

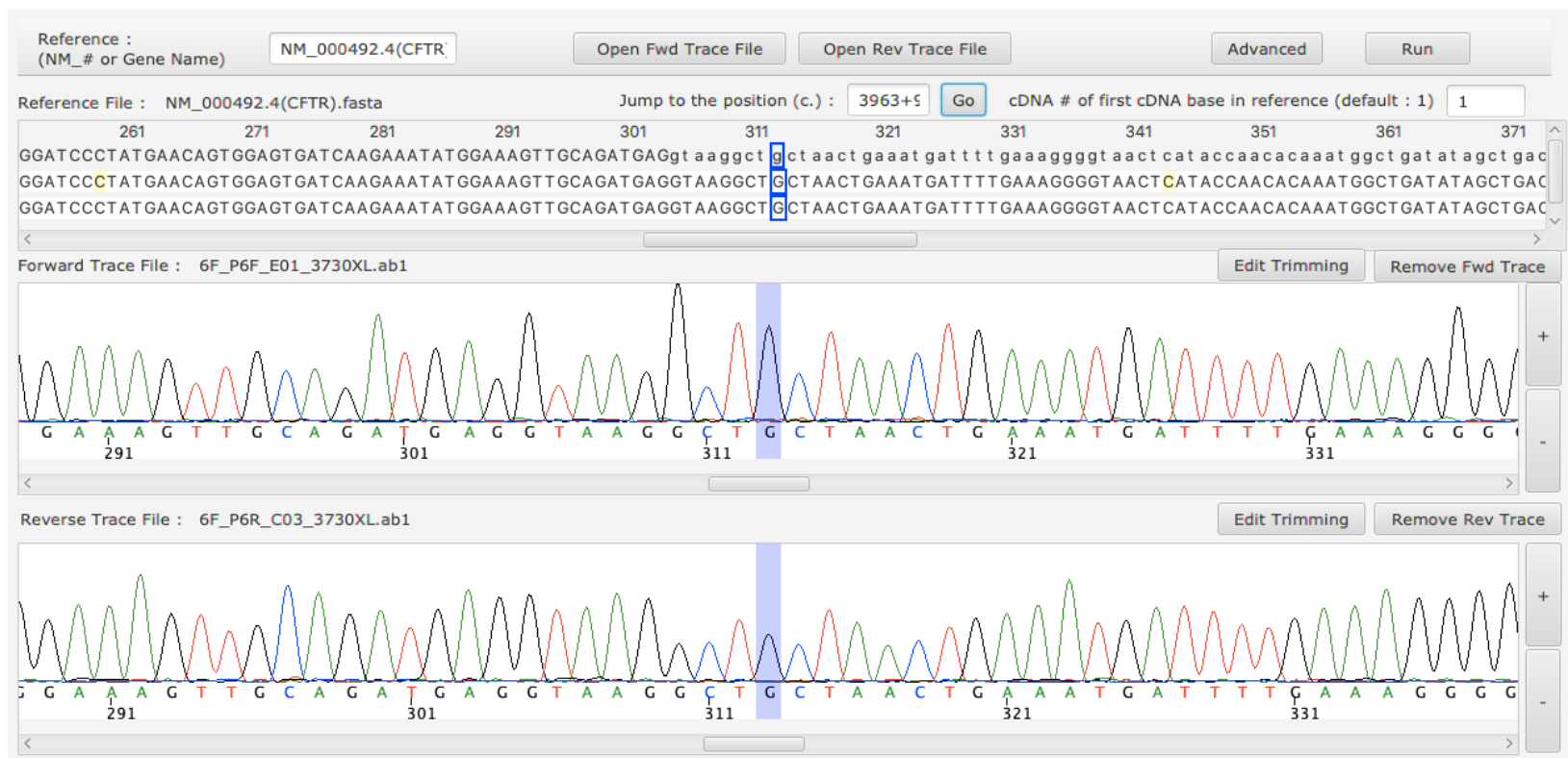


Figure 16: SnackVar report for the c.3963+9G>C position in CA1615190

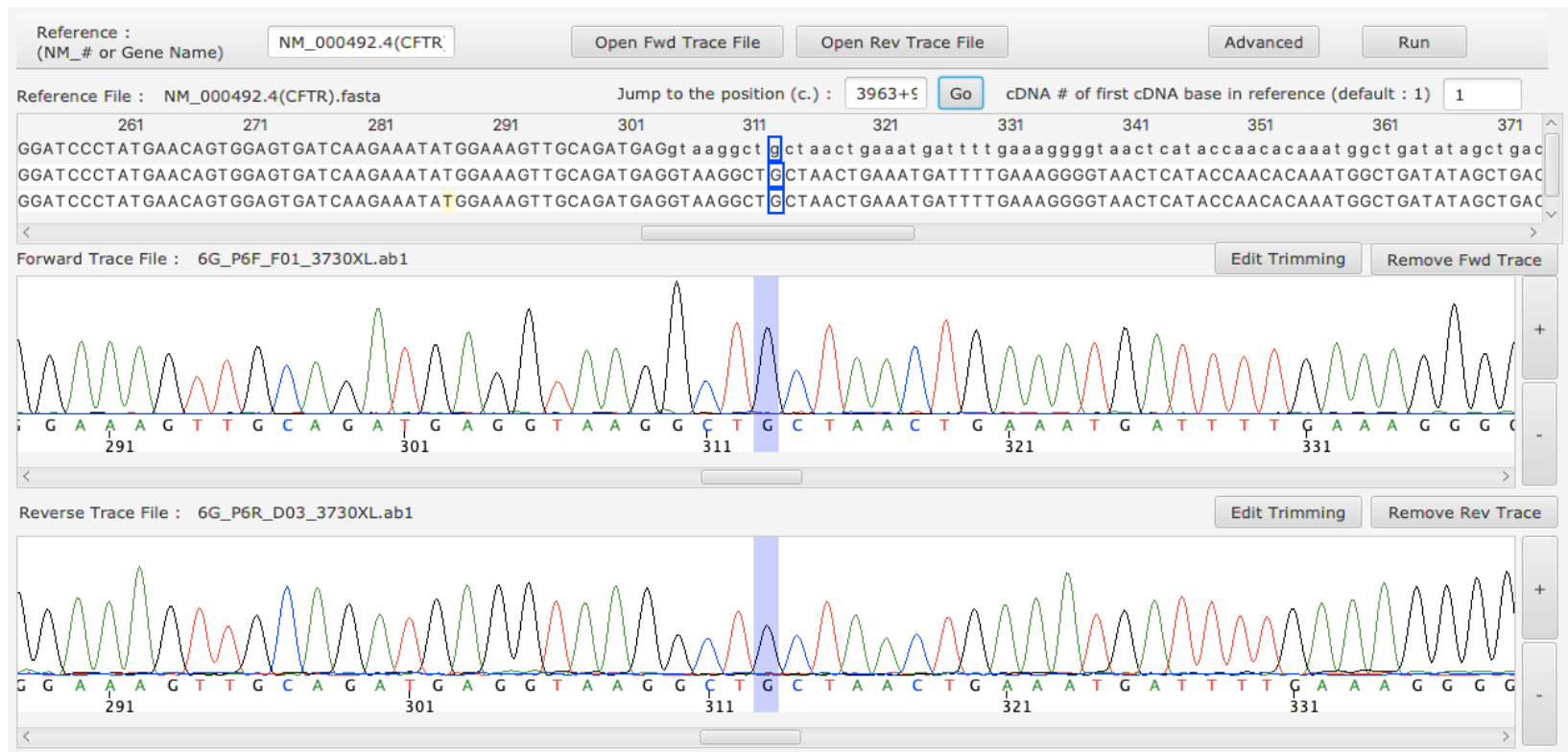


Figure 17: SnackVar report for the c.3963+9G>C position in CF9830825

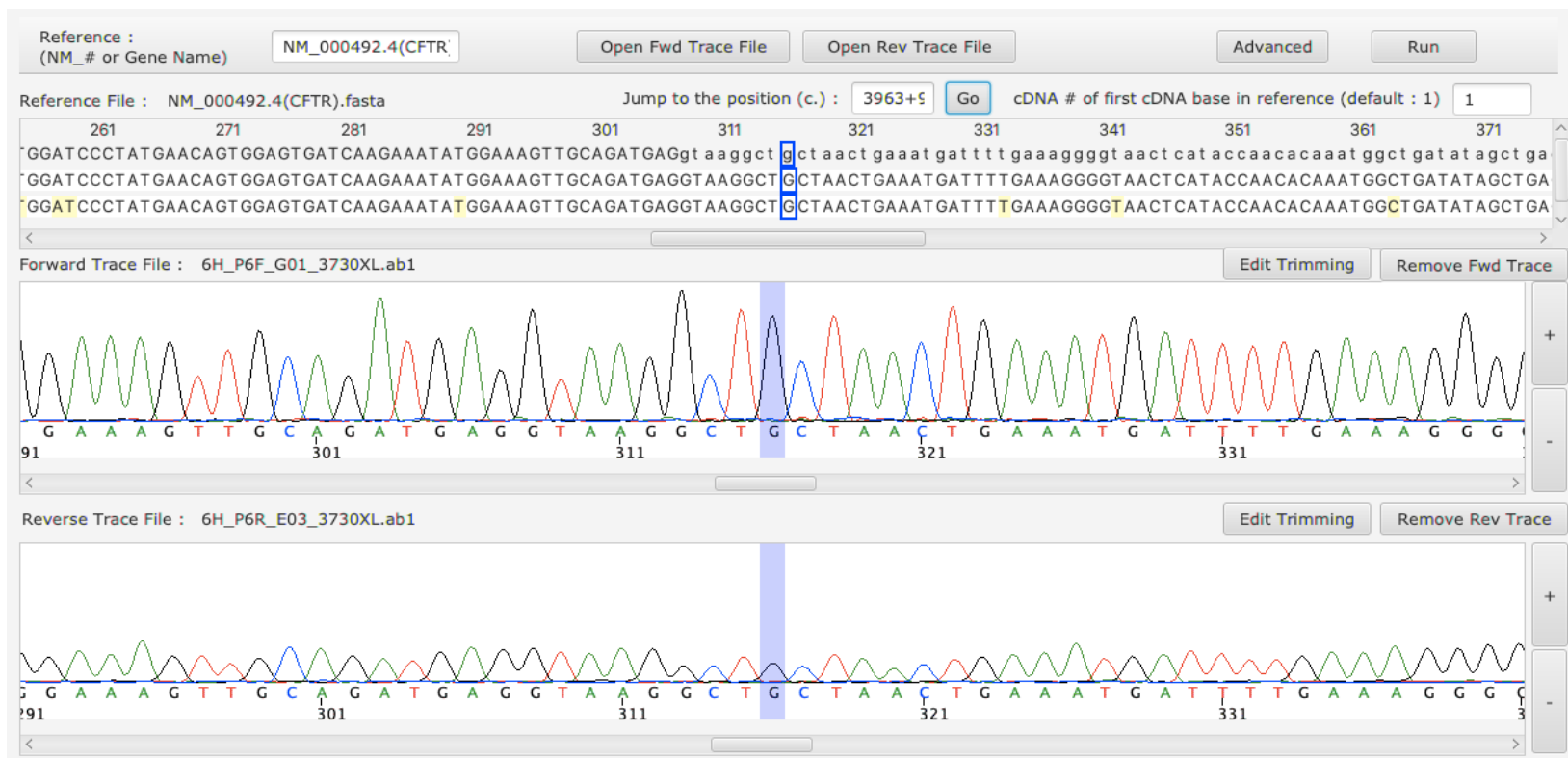


Figure 18: SnackVar report for the c.3963+9G>C position in CF7527369



Figure 19: SnackVar report for the c.3963+9G>C position in CF8213552



Figure 20: SnackVar report for the c.3963+9G>C position in CF5158167

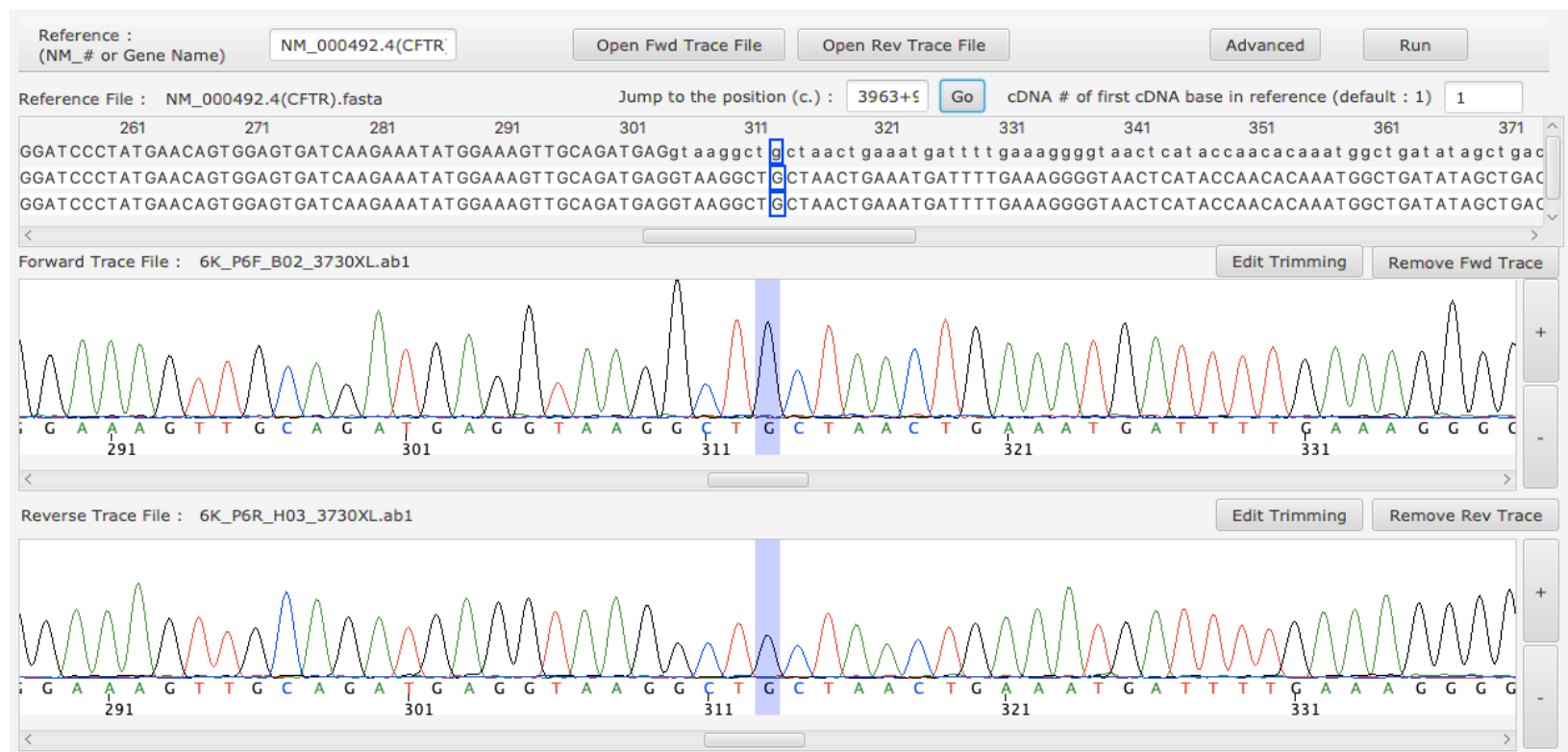


Figure 21: SnackVar report for the c.3963+9G>C position in CF4833948

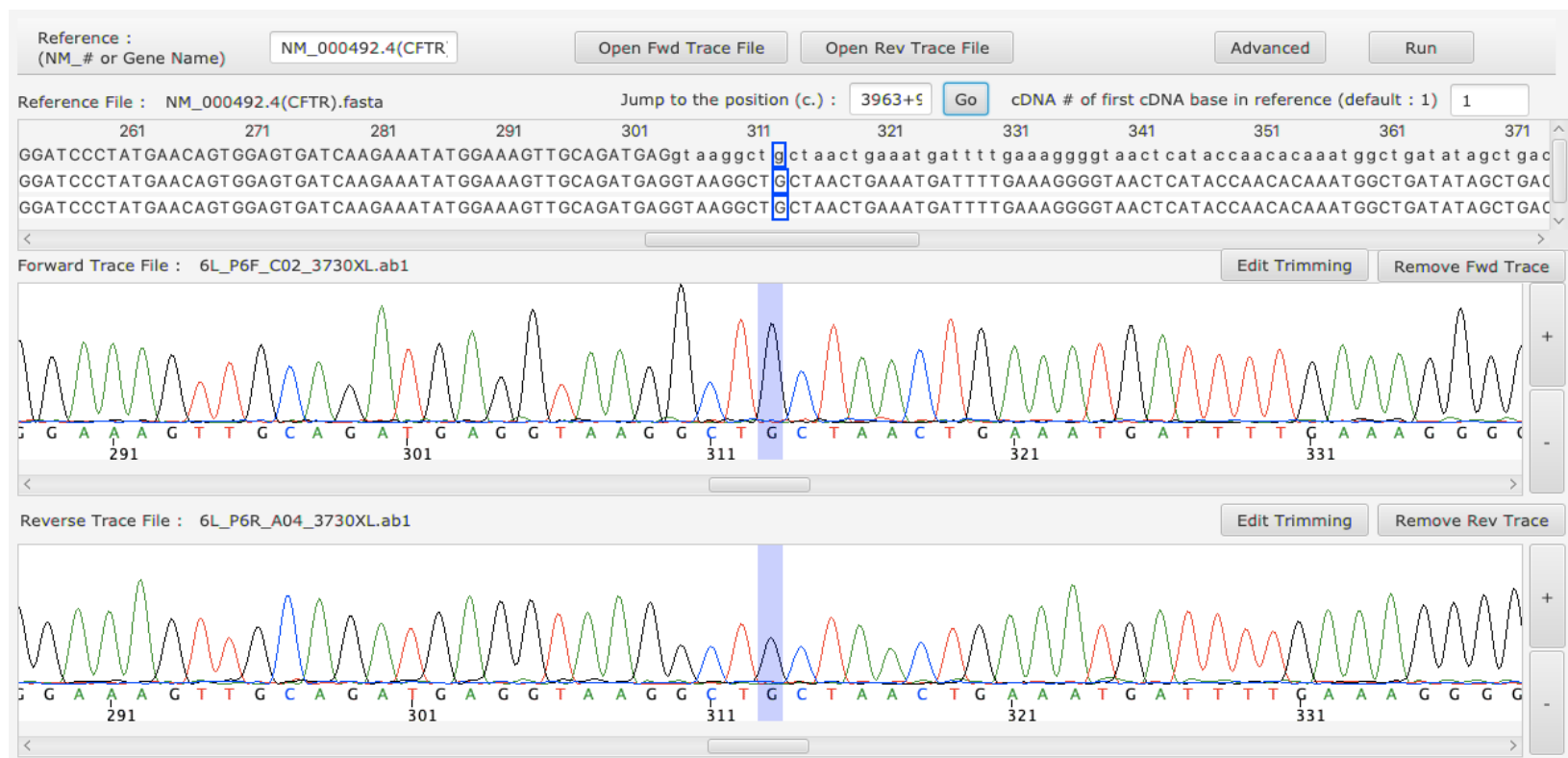


Figure 22: SnackVar report for the c.3963+9G>C position in CF4471587

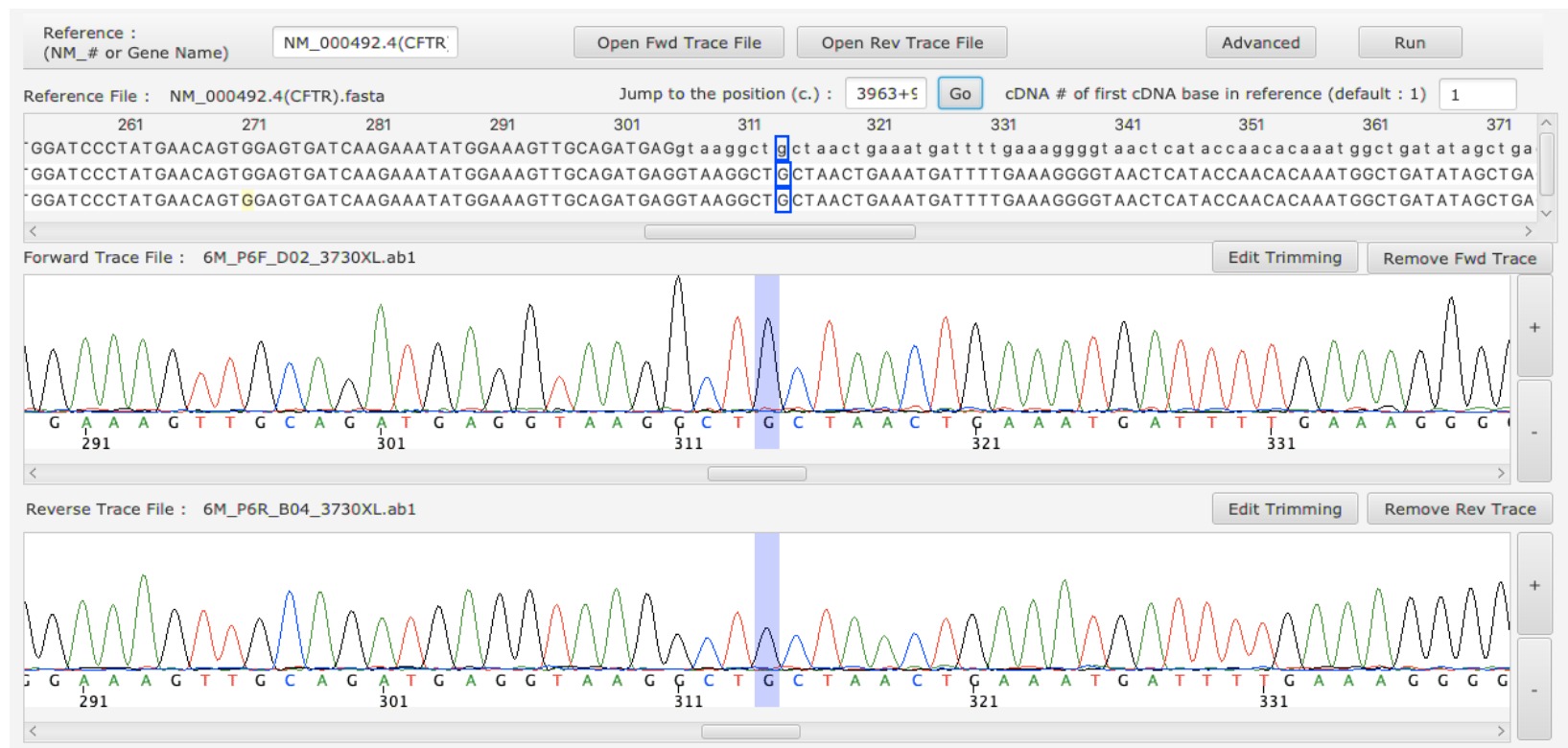


Figure 23: SnackVar report for the c.3963+9G>C position in CF5181003



Figure 24: SnackVar report for the c.3963+9G>C position in CF0018616



Figure 25: SnackVar report for the p.Gly451Ter position in CA4932026



Figure 26: SnackVar report for the p.Gly451Ter position in CF1697504



Figure 28: SnackVar report for the p.Gly451Ter position in CF4833948



Figure 29: SnackVar report for the p.Gly451Ter position in CF5158167



Figure 30: SnackVar report for the p.Gly451Ter position in CF4283433

ID : CF0014912; p.Leu1258PhefsX7 Description : Confirmation of pLeu1258PhefsX7 variant in CF0014912
Reference sequence : NM_000492.4(CFTR).fasta

Description : c.3773dup, p.(Leu1258Phefs*7), hetero

111 121 131 141 151 161 171 181 191 201 211 221
 ttttaccttatagGTGGGCCTCTTGGGAAGAACTGGATCAGGGAAGAGTACTTTGTTATCAGCTTTTTTGTAGACTACTGAACACTGAAGGAGAAATCCAGATCGATGGTGTGTCTTGGGATT
 TTTTACCTTATAGGTGGGCCTCTTGGGAAGAACTGGATCAGGGAAGAGTACTTTGTTATCAGCTTTTTTKRRR...Y...YKRA...YKRRRRRRRA...YC_RA_YSR_KKGGKKYYTKGGR...T
 TTT...CCYT...RGKKGGSCYYTYTKGRRARRA_YKGR_Y_RGRRARRRK...YTKKT...Y_RSYTTTTTGTAGACTACTGAACACTGAAGGAGAAATCCAGATCGATGGTGTGTCTTGGGATT

Figure 31: SnackVar report for the p.Leu1258PhefsX7 variant in CF0014912

Hetero Indel View (Forward) : 8A_P8F_H05_3730XL.ab1



Hetero Indel View (Reverse) : 8A_P8R_A06_3730XL.ab1



Figure 32: SnackVar report for the p.Leu1258PhefsX7 variant in CF0014912



Figure 33: SnackVar report for the p.Ser549Asn position in CF0014912

SnackVar Report

ID : CF3796568; p.Ser1118Phe

Description : Confirmation of homozygous p.Ser1118Phe in CF3796568;

Reference sequence : NM_000492.4(CFTR).fasta

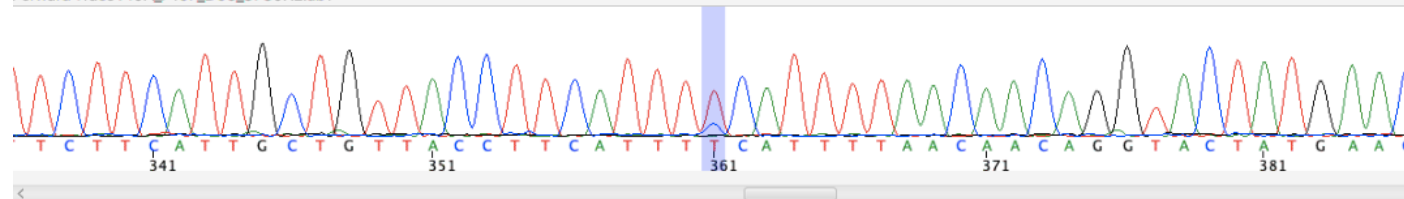
Variant 1

Description : c.3353C>T, p.(Ser1118Phe), homo

Alignment

311 321 331 341 351 361 371 381 391 401 411 421
TCCAAATGAGAATAGAAATGATTTTGTGTCATCTTCTTCATTGCTGTTACCTTCATTTTCATTTTAACAACAGGt act at gaact cat t aact t t agct aagcat t t aagt aaaaaatt t t c
TCCAAATGAGAATAGAAATGATTTTGTGTCATCTTCTTCATTGCTGTTACCTTCATTTTCATTTTAACAACAGGTACTATGAACCTCATTAACTTTAGCTAAGCATTTAAGTAAAAAATTTTC
TCCAAATGAGAATAGAAATGATTTTGTGTCATCTTCTTCATTGCTGTT...CCYTCATTTTCATTTTAACA...CAGGTRCTATRAACTCATT...CTTTAGSTAAGCATTTAAGTAAAAAATTTTC

Forward Trace: 10A_P10F_D06_3730XLab1



Reverse Trace: 10A_P10R_E06_3730XLab1

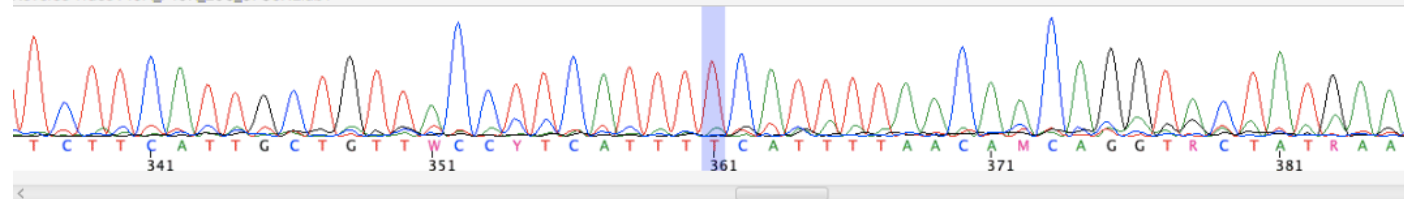


Figure 34: SnackVar report for the p.Ser1118Phe variant in CF3796568

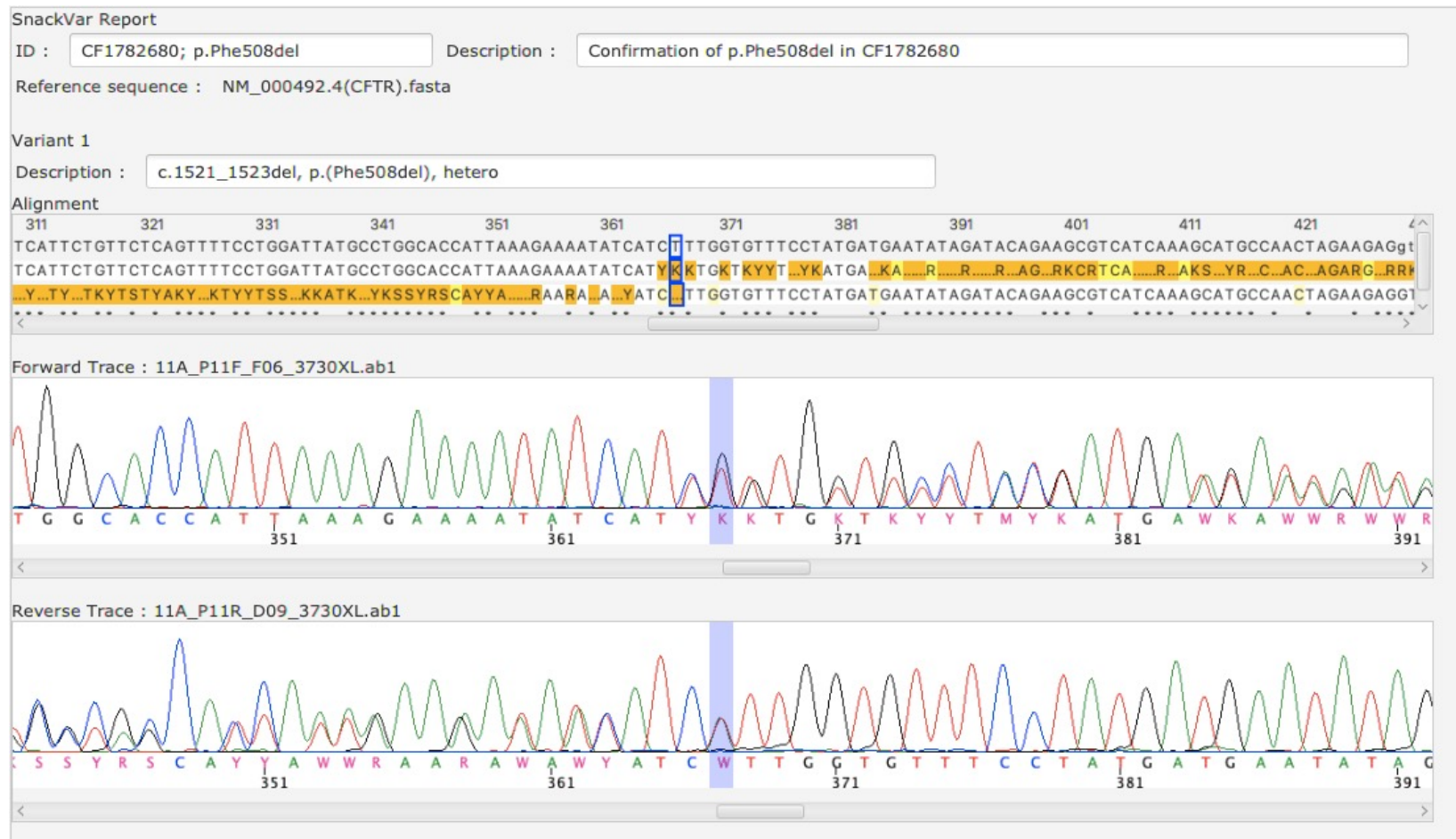
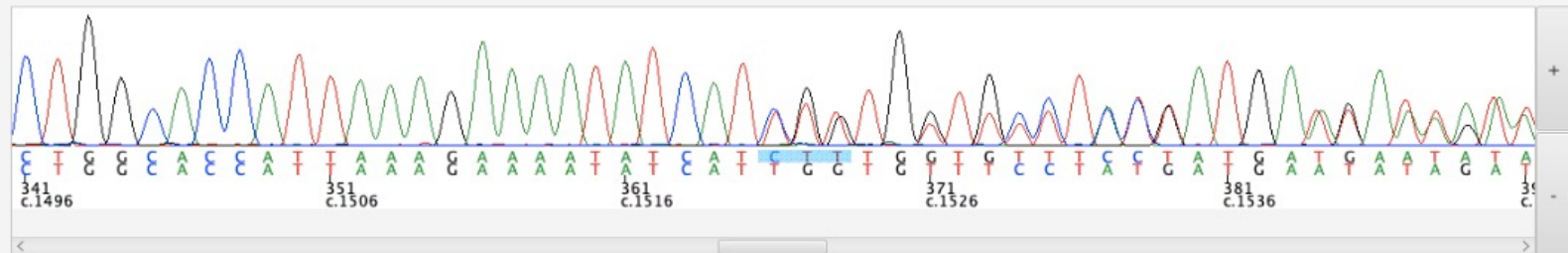


Figure 35: SnackVar report for the p.Phe508del variant in CF1782680

Hetero Indel View (Forward) : 11A_P11F_F06_3730XL.ab1

```
CTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATCTTTGGTGTTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAACTA  
CTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATTTTGGTGTTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTACAAGAGGTAAGAACTA
```



Hetero Indel View (Reverse) : 11A_P11R_D09_3730XL.ab1

```
TCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATCTTTGGTGTTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAA  
TCTGTTATCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATTTTGGTGTTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAA
```

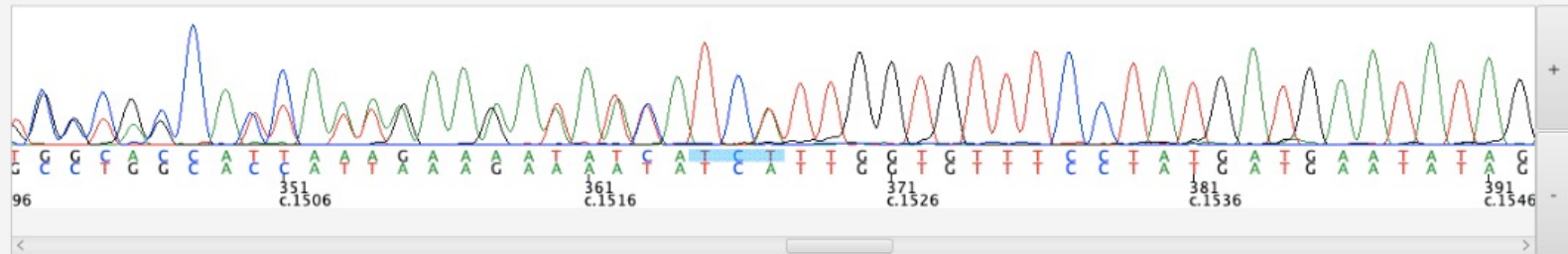


Figure 36: SnackVar report for the p.Phe508del variant in CF1782680

SnackVar Report

ID : CF7930867; p.Phe508del

Description : Confirmation of p.Phe508del in CF7930867

Reference sequence : NM_000492.4(CFTR).fasta

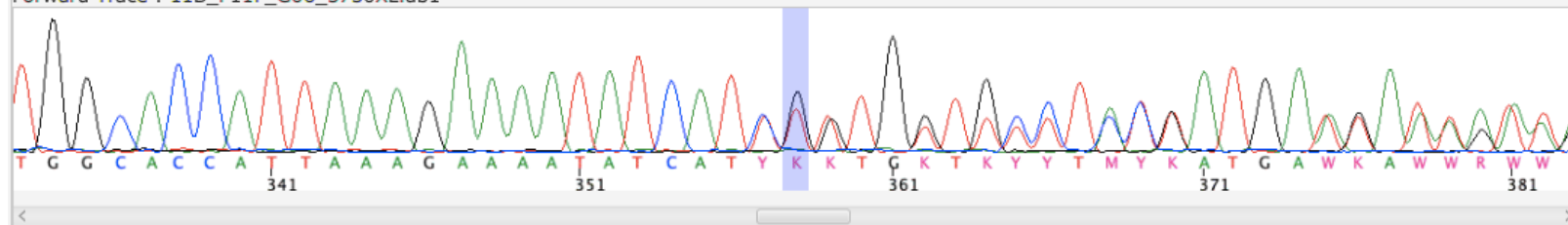
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment

301 311 321 331 341 351 361 371 381 391 401 411
AATTTCAATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAG.
AATTTCAATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAG.
AAK...Y...TY...TKYTSTYAKY...KTYTSS...KKATK...YKSSYRSCAYYA...RAARA...A...YATC...TTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAG.
<>

Forward Trace : 11B_P11F_G06_3730XL.ab1



Reverse Trace : 11B_P11R_E09_3730XL.ab1

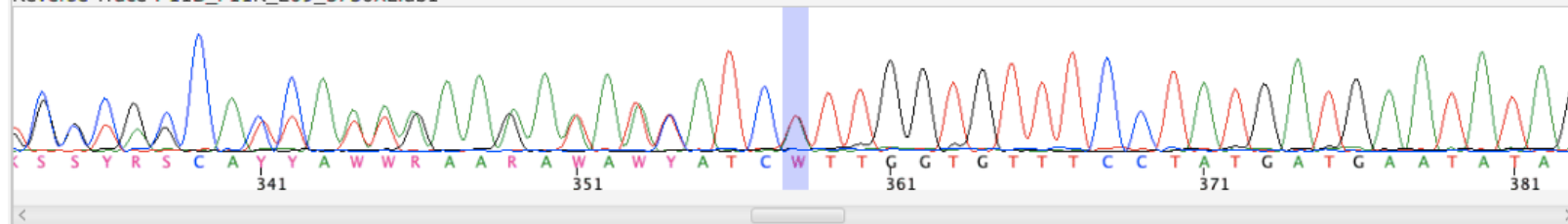


Figure 37: SnackVar report for the p.Phe508del variant in CF7930867

Hetero Indel View (Forward) : 11B_P11F_G06_3730XL.ab1

```
CTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAACTAT  
CTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAACTAT
```



Hetero Indel View (Reverse) : 11B_P11R_E09_3730XL.ab1

```
TCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAA  
TCTGTTATCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAAATATCATTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAA
```



Figure 38: SnackVar report for the p.Phe508del variant in CF7930867

SnackVar Report

ID : CF4062212; c.1393-1G>A

Description : Confirmation of c.1393-1G>A in CF4062212

Reference sequence : NM_000492.4(CFTR).fasta

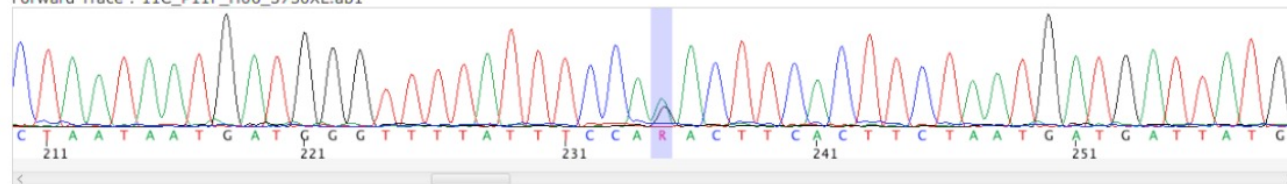
Variant 1

Description : c.1393-1G>A, p.?, hetero

Alignment

181 191 201 211 221 231 241 251 261 271 281 291
jaggcaagt gaat cct gagcgt gat t t gataaat gacct aat aat gat ggggt t t t t t cca g a c t t c a c t t c t a a t g g t g a t t a t g g g a g a a c t g g a g c c t t c a g a g g g t a a a t t a a g c a c /
A G G C A A G T G A A T C C T G A G C G T G A T T T G A T A A T G A C C T A A T A A T G A T G G G T T T T A T T T C C A g a c t t c a c t t c t a a t g a t g a t t a t g g g a g a a c t g g a g c c t t c a g a g g t a a a t t a a g c a c /
A G G C A A G T G A A T C C T G A G C G T G A T T T G A T A A T G A C C T A A T A A T G A T G G G T T T T A T T T C C A g a c t t c a c t t c t a a t g a t g a t t a t g g g a g a a c t g g a g c c t t c a g a g g t a a a t t a a g c a c /

Forward Trace : 11C_P11F_H06_3730XL.ab1



Reverse Trace : 11C_P11R_F09_3730XL.ab1

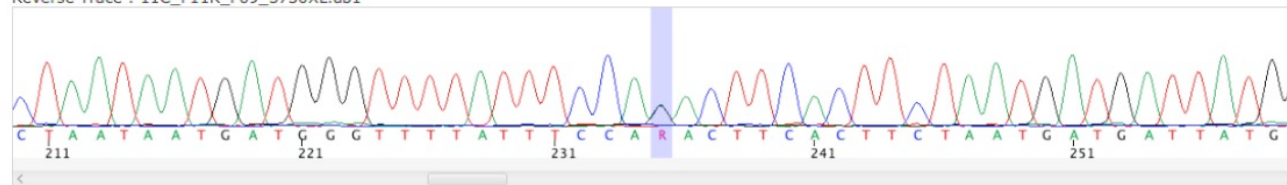


Figure 39: SnackVar report for the c.1393-1G>A variant in CF4062212

SnackVar Report

ID : CF1697504; p.Phe508del

Description : Confirmation of p.Phe508del in CF1697504

Reference sequence : NM_000492.4(CFTR).fasta

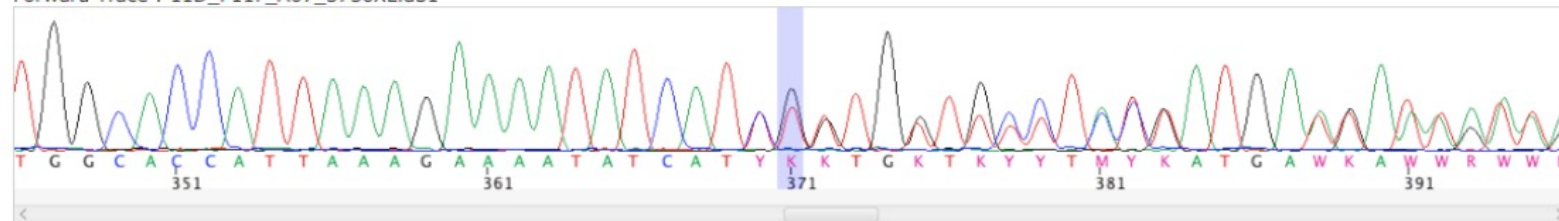
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment

```
1      321      331      341      351      361      371      381      391      401      411      421      431
TTTCATTCTGTTCTCAGTTCCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAG
TTTCATTCTGTTCTCAGTTCCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAG
<...Y...TY...TKYTSTYAKY...KTYTSS...KKATK...YKSSYRSCAYYA...RAARA...A...YATC...TTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAG
```

Forward Trace : 11D_P11F_A07_3730XL.ab1



Reverse Trace : 11D_P11R_G09_3730XL.ab1



Figure 40: SnackVar report for the p.Phe508del variant in CF1697504

Hetero Indel View (Forward) : 11D_P11F_A07_3730XL.ab1



Hetero Indel View (Reverse) : 11D_P11R_G09_3730XL.ab1



Figure 41: SnackVar report for the p.Phe508del variant in CF1697504

SnackVar Report

ID : CF4544212; p.Phe508del

Description : Confirmation of homozygous p.Phe508del in CF4544212

Reference sequence : NM_000492.4(CFTR).fasta

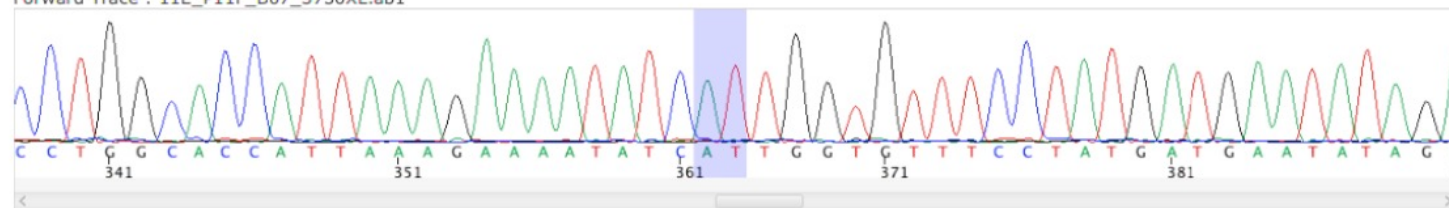
Variant 1

Description : c.1521_1523del, p.(Phe508del), homo

Alignment

311 321 331 341 351 361 371 381 391 401 411 421
AGAATTTCAATCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCACTTTTGGTGTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAG
AGAATTTCAATCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCACTTTTGGTGTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAG
AGAATTTCAATCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCACTTTTGGTGTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAG

Forward Trace : 11E_P11F_B07_3730XL.ab1



Reverse Trace : 11E_P11R_H09_3730XL.ab1

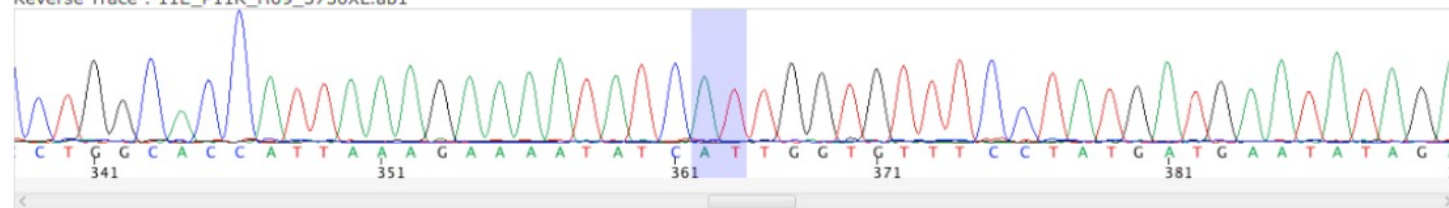


Figure 42: SnackVar report for the p.Phe508del variant in CF4544212

SnackVar Report

ID : CF7527369; p.Phe508del

Description : Confirmation of p.Phe508del in CF7527369

Reference sequence : NM_000492.4(CFTR).fasta

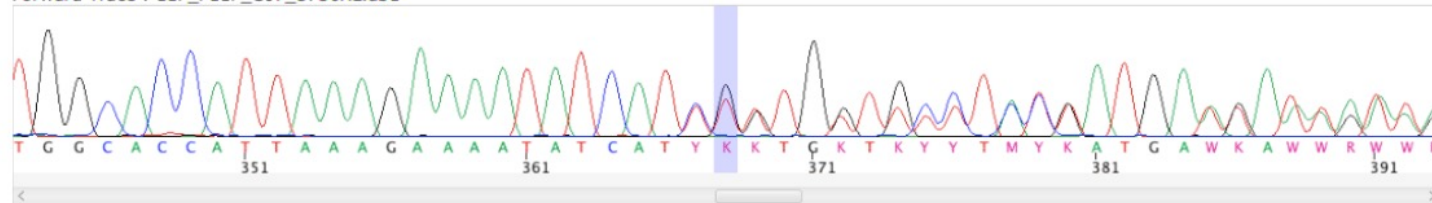
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment

311 321 331 341 351 361 371 381 391 401 411 421
TTTCATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAG
TTTCATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAG
K...Y...TY...TKYTSTY_KY_KIYYTSS_KKATK_YKSSYRSCAYYA...RAARA_A_YATC...TTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAG

Forward Trace : 11F_P11F_C07_3730XL.ab1



Reverse Trace : 11F_P11R_A10_3730XL.ab1



Figure 43: SnackVar report for the p.Phe508del variant in CF7527369

Hetero Indel View (Forward) : 11F_P11F_C07_3730XL.ab1



Hetero Indel View (Reverse) : 11F_P11R_A10_3730XL.ab1



Figure 44: SnackVar report for the p.Phe508del variant in CF7527369

SnackVar Report

ID : CF4602380; p.Phe508del

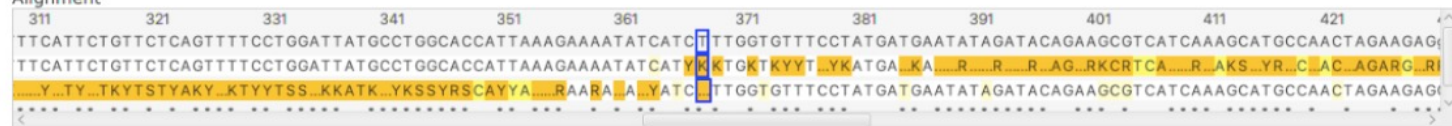
Description : Confirmation of p.Phe508del in CF4602380

Reference sequence : NM_000492.4(CFTR).fasta

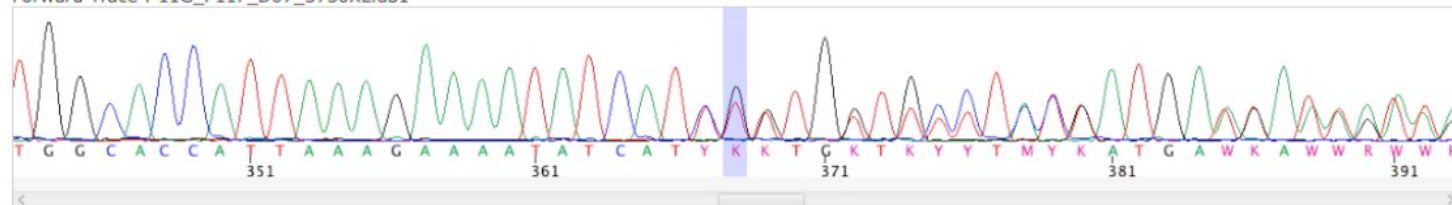
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11G_P11F_D07_3730XL.ab1



Reverse Trace : 11G_P11R_B10_3730XL.ab1

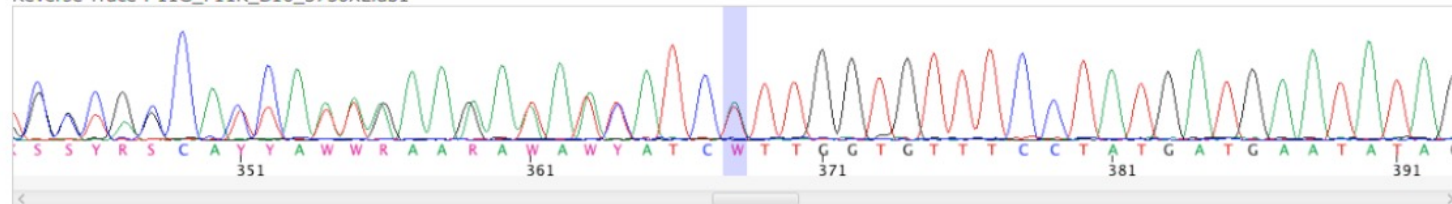
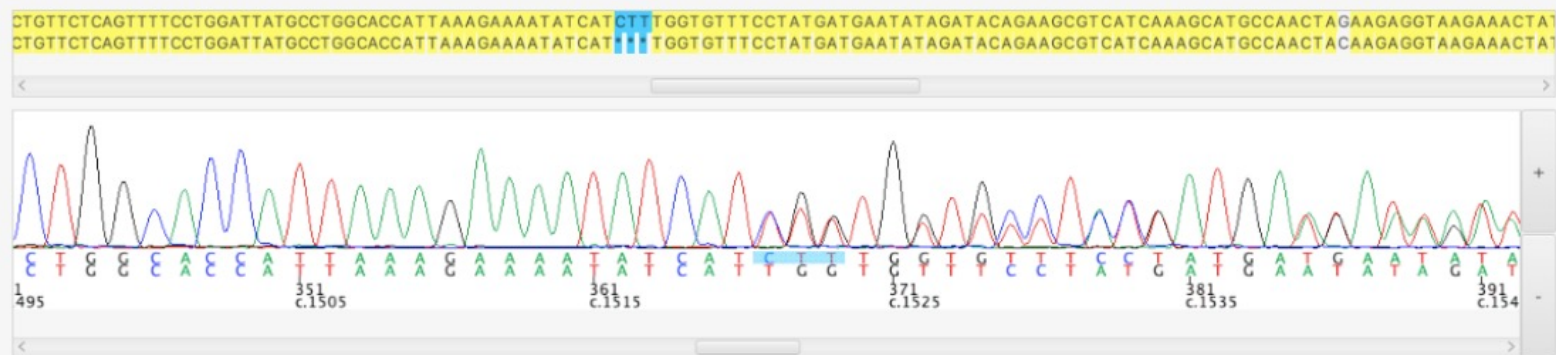


Figure 45: SnackVar report for the p.Phe508del variant in CF4602380

Hetero Indel View (Forward) : 11G_P11F_D07_3730XL.ab1



Hetero Indel View (Reverse) : 11G_P11R_B10_3730XL.ab1



Figure 46: SnackVar report for the p.Phe508del variant in CF4602380

SnackVar Report

ID : CF5181003; p.Phe508del

Description : Confirmation of homozygous p.Phe508del in CF5181003

Reference sequence : NM_000492.4(CFTR).fasta

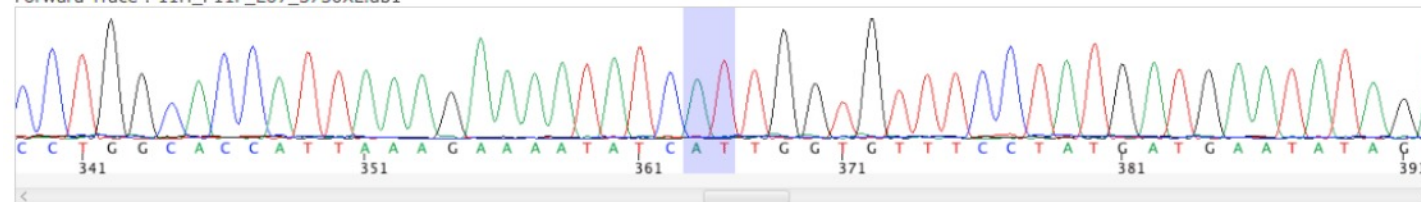
Variant 1

Description : c.1521_1523del, p.(Phe508del), homo

Alignment



Forward Trace : 11H_P11F_E07_3730XL.ab1



Reverse Trace : 11H_P11R_C10_3730XL.ab1

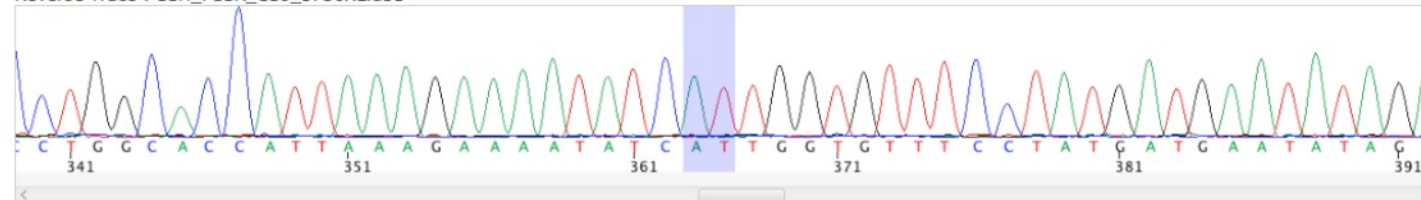


Figure 47: SnackVar report for the p.Phe508del variant in CF5181003

SnackVar Report

ID : CF3115703; p.Phe508del

Description : Confirmation of homozygous p.Phe508del in CF3115703

Reference sequence : NM_000492.4(CFTR).fasta

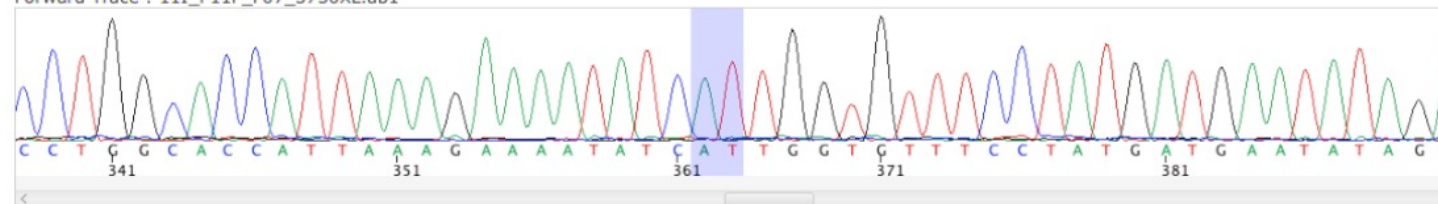
Variant 1

Description : c.1521_1523del, p.(Phe508del), homo

Alignment

311 321 331 341 351 361 371 381 391 401 411 421
AATTTTCATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCAATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAA
AATTTTCATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCAATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAA
AATTTTCATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCAATCTTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAA
< * * *

Forward Trace : 11I_P11F_F07_3730XL.ab1



Reverse Trace : 11I_P11R_D10_3730XL.ab1

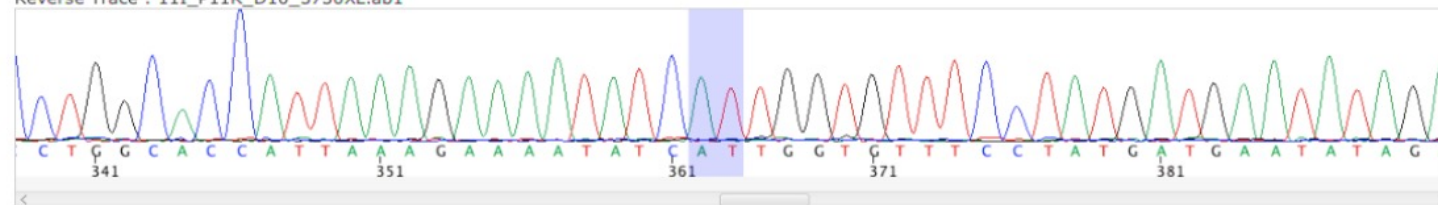


Figure 48: SnackVar report for the p.Phe508del variant in CF3115703

SnackVar Report

ID : CF3512286; p.Phe508del

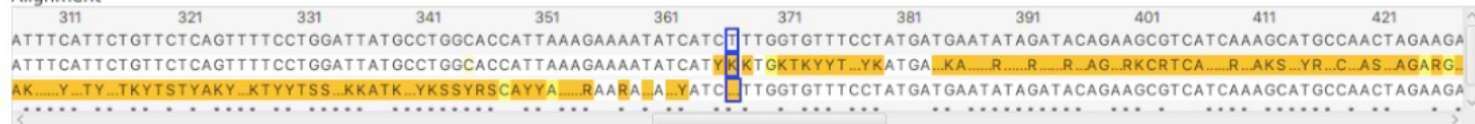
Description : Confirmation of p.Phe508del in CF3512286

Reference sequence : NM_000492.4(CFTR).fasta

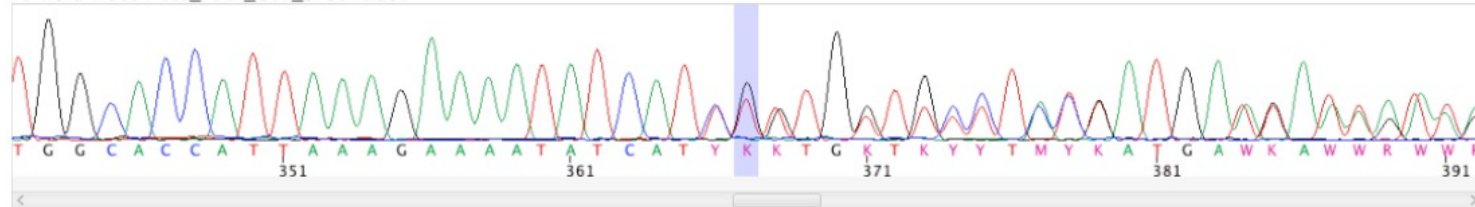
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11J_P11F_G07_3730XL.ab1



Reverse Trace : 11J_P11R_E10_3730XL.ab1

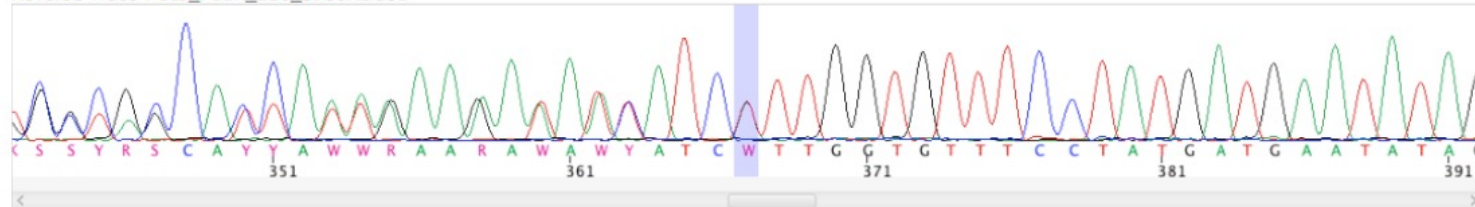


Figure 49: SnackVar report for the p.Phe508del variant in CF3512286

Hetero Indel View (Forward) : 11J_P11F_G07_3730XL.ab1



Hetero Indel View (Reverse) : 11J_P11R_E10_3730XL.ab1



Figure 50: SnackVar report for the p.Phe508del variant in CF3512286

SnackVar Report

ID : CF2843425; p.Phe508del

Description :	Confirmation of p.Phe508del in CF2843425
---------------	--

Reference sequence : NM_000492.4(CFTR).fasta

Variant 1

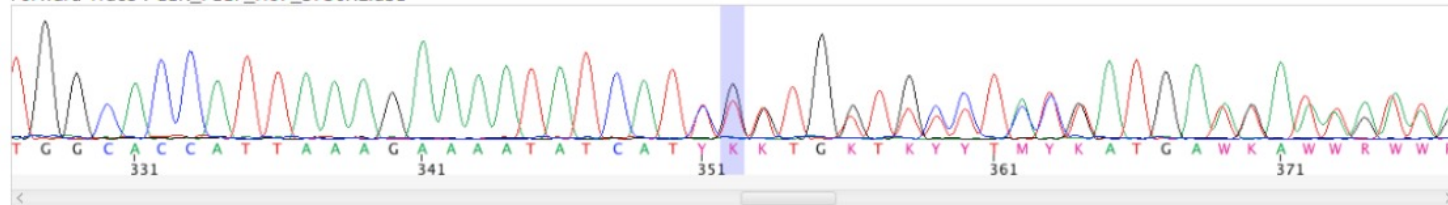
Description : c.1521_1523del, p.(Phe508del), hetero

Alignment

TCATTCTGTTCTCAGTTTTCTCGGATTATGCCTGGACCATTAAAGAAAATATCATCTTGGTGTTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGgt
TCATTCTGTTCTCAGTTTTCTCGGATTATGCCTGGACCATTAAAGAAAATATCATYKKTGTkTKYYT_YKATGA_KA_R_R_R_AG_RKRTCA_R_AKS_YR_C_AS_AGARGARRH
Y_TY_TKYTSTYAKY_KTYITSS_KKATK_YKSSYSRCAYYA_RAARA_A_YATCTGGTGTTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGgt

< * * * * *

Forward Trace : 11K_P11F_H07_3730XL.ab1



Reverse Trace : 11K_P11R_F10_3730XL.ab1

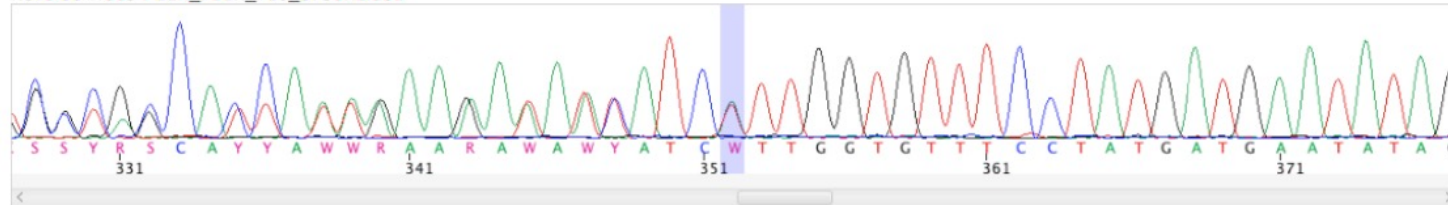


Figure 51: SnackVar report for the p.Phe508del variant in CF2843425

Hetero Indel View (Forward) : 11K_P11F_H07_3730XL.ab1



Hetero Indel View (Reverse) : 11K_P11R_F10_3730XL.ab1



Figure 52: SnackVar report for the p.Phe508del variant in CF2843425

SnackVar Report

ID : CF8213552; p.Phe508del

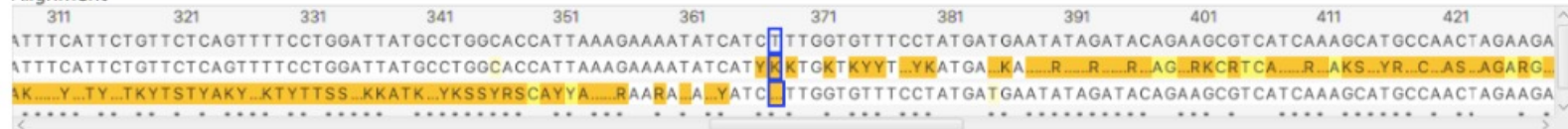
Description : Confirmation of p.Phe508del in CF8213552

Reference sequence : NM_000492.4(CFTR).fasta

Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11L_P11F_A08_3730XL.ab1

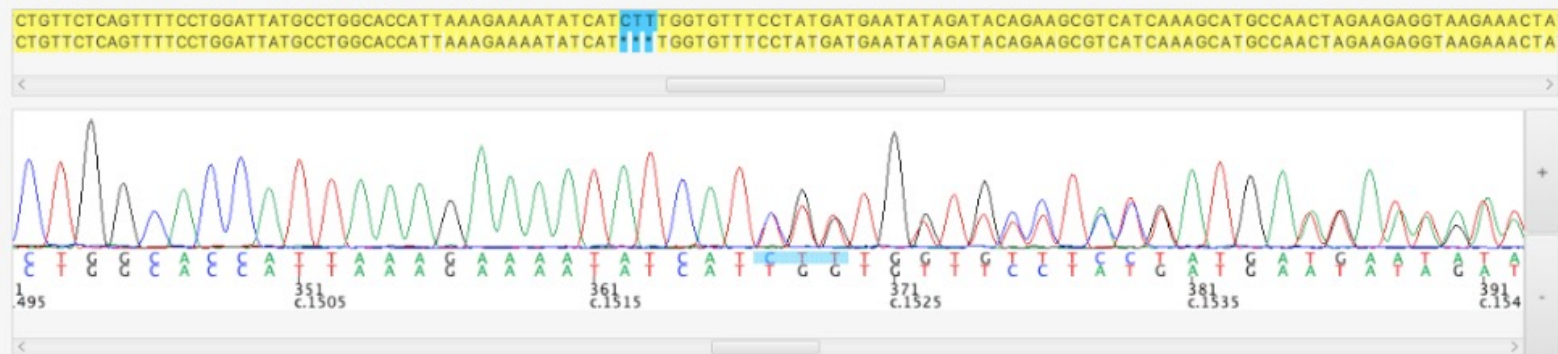


Reverse Trace : 11L_P11R_G10_3730XL.ab1



Figure 53: SnackVar report for the p.Phe508del variant in CF8213552

Hetero Indel View (Forward) : 11L_P11F_A08_3730XL.ab1



Hetero Indel View (Reverse) : 11L_P11R_G10_3730XL.ab1

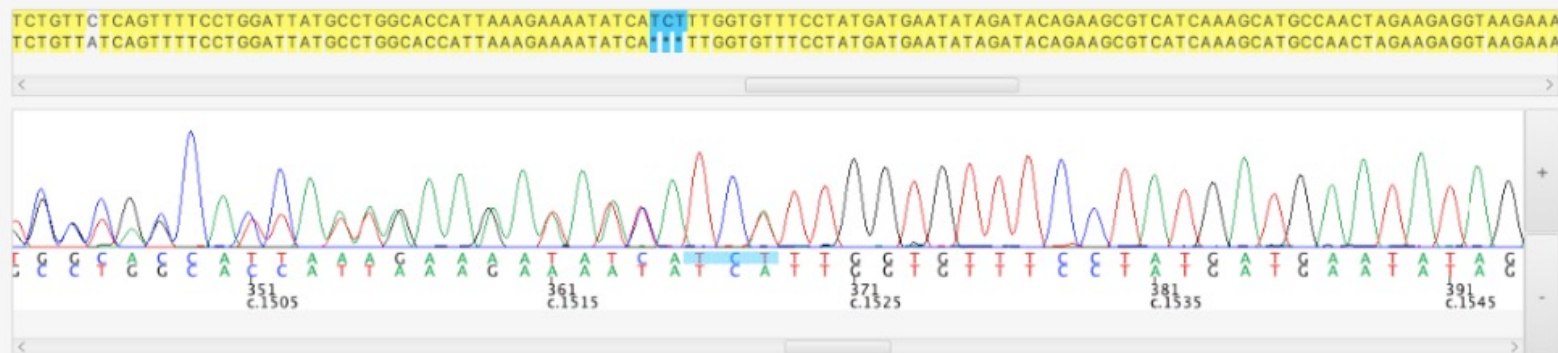


Figure 54: SnackVar report for the p.Phe508del variant in CF8213552

SnackVar Report

ID : CF3803349; p.Phe508del

Description : Confirmation of p.Phe508del in CF3803349

Reference sequence : NM_000492.4(CFTR).fasta

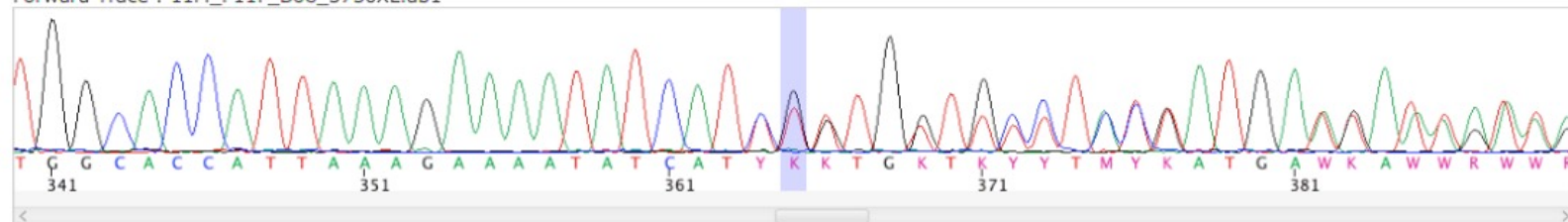
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11M_P11F_B08_3730XL.ab1



Reverse Trace : 11M_P11R_H10_3730XL.ab1

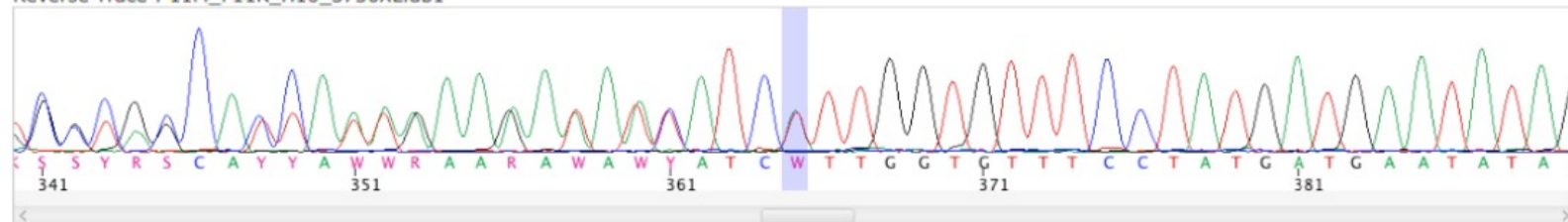
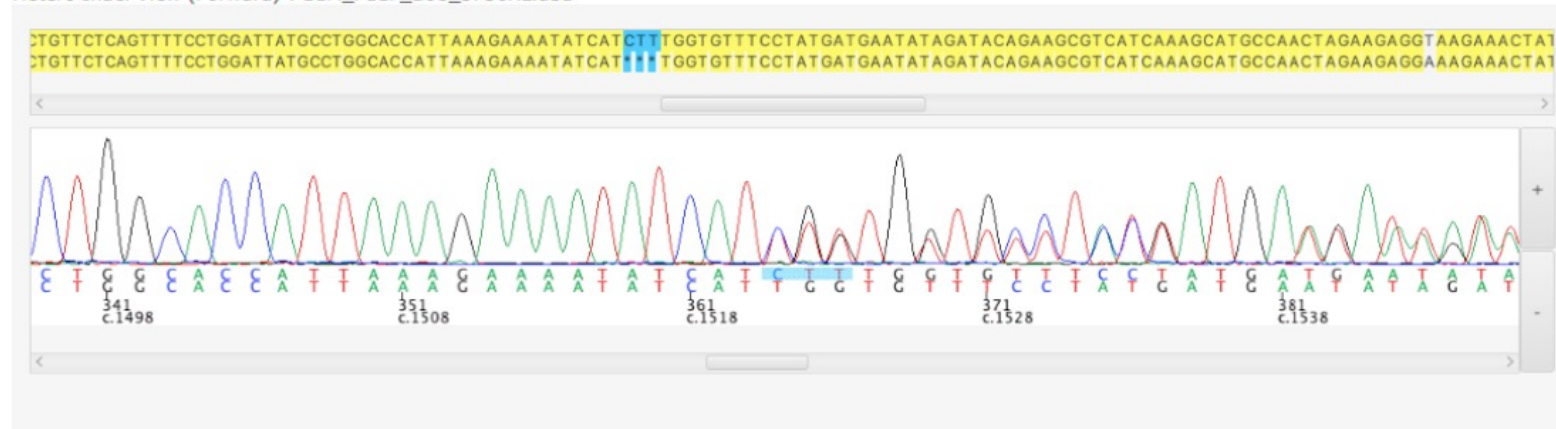


Figure 55: SnackVar report for the p.Phe508del variant in CF3803349

Hetero Indel View (Forward) : 11M_P11F_B08_3730XL.ab1



Hetero Indel View (Reverse) : 11M_P11R_H10_3730XL.ab1



Figure 56: SnackVar report for the p.Phe508del variant in CF3803349

SnackVar Report

ID : CF9830825; p.Phe508del

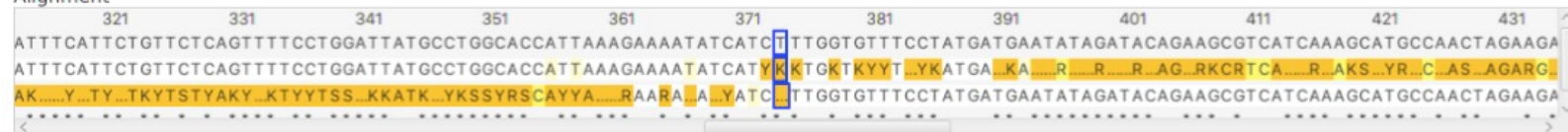
Description : Confirmation of p.Phe508del in CF9830825

Reference sequence : NM_000492.4(CFTR).fasta

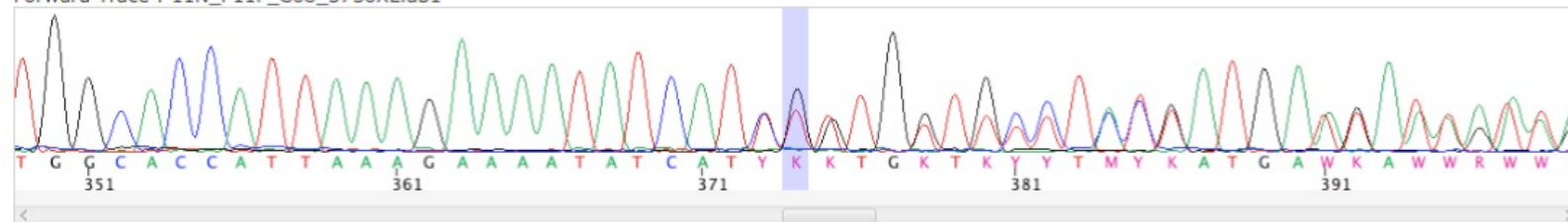
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11N_P11F_C08_3730XL.ab1



Reverse Trace : 11N_P11R_A11_3730XL.ab1

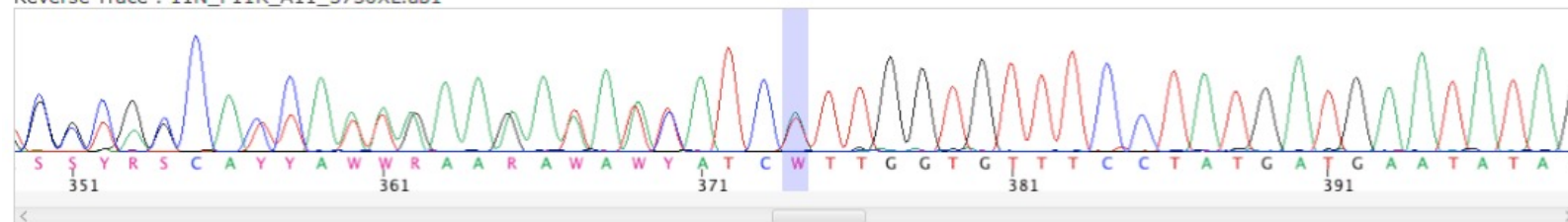


Figure 57: SnackVar report for the p.Phe508del variant in CF9830825

Hetero Indel View (Forward) : 11N_P11F_C08_3730XL.ab1



Hetero Indel View (Reverse) : 11N_P11R_A11_3730XL.ab1



Figure 58: SnackVar report for the p.Phe508del variant in CF9830825

SnackVar Report

ID : CF5384911; p.Phe508del

Description : Confirmation of p.Phe508del in CF5384911

Reference sequence : NM_000492.4(CFTR).fasta

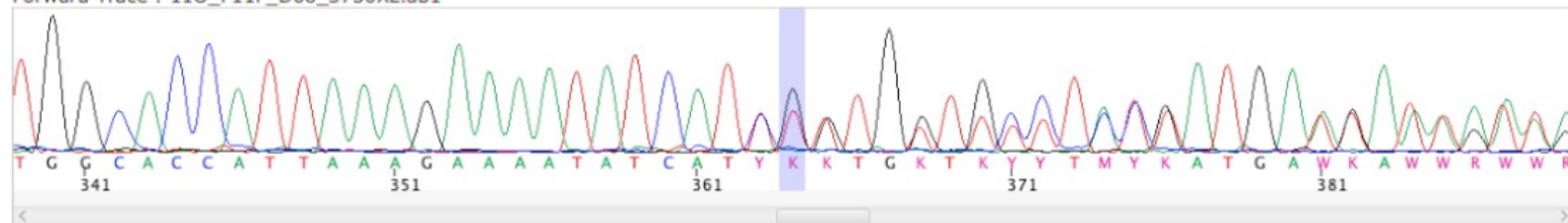
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment

Alignment view showing the reference sequence (NM_000492.4(CFTR).fasta) and the variant sequence (c.1521_1523del, p.(Phe508del), hetero). The variant is highlighted in yellow. The alignment shows a deletion of three nucleotides (CTG) at position 1521, resulting in a premature stop codon (TGA) at position 1523.

Forward Trace : 11O_P11F_D08_3730XL.ab1



Reverse Trace : 11O_P11R_B11_3730XL.ab1

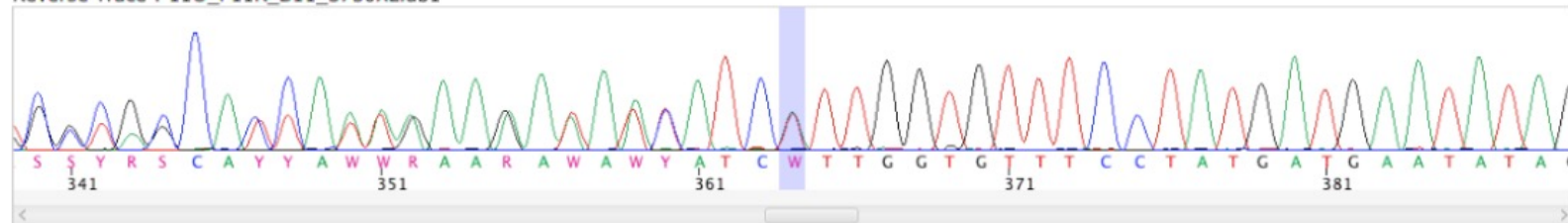


Figure 59: SnackVar report for the p.Phe508del variant in CF5384911

Hetero Indel View (Forward) : 11O_P11F_D08_3730XL.ab1



Hetero Indel View (Reverse) : 11O_P11R_B11_3730XL.ab1

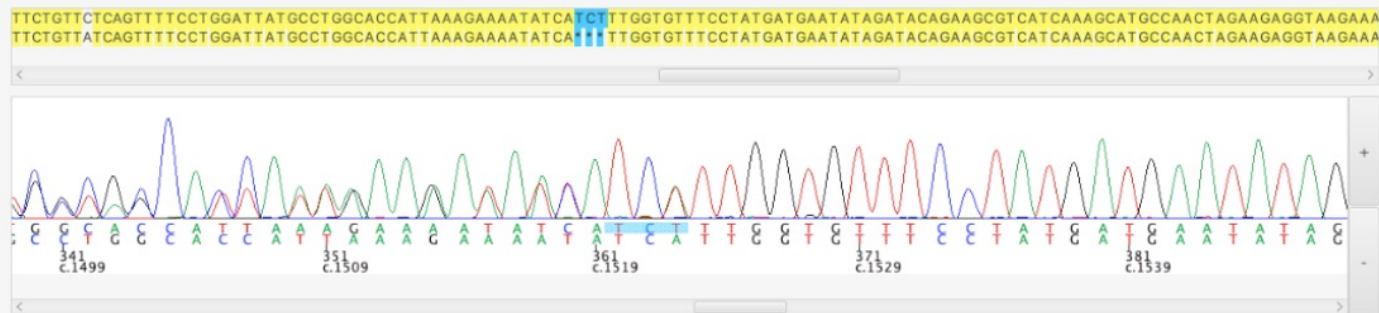


Figure 60: SnackVar report for the p.Phe508del variant in CF5384911

SnackVar Report

ID : CF9862557; p.Phe508del

Description : Confirmation of p.Phe508del in CF9862557

Reference sequence : NM_000492.4(CFTR).fasta

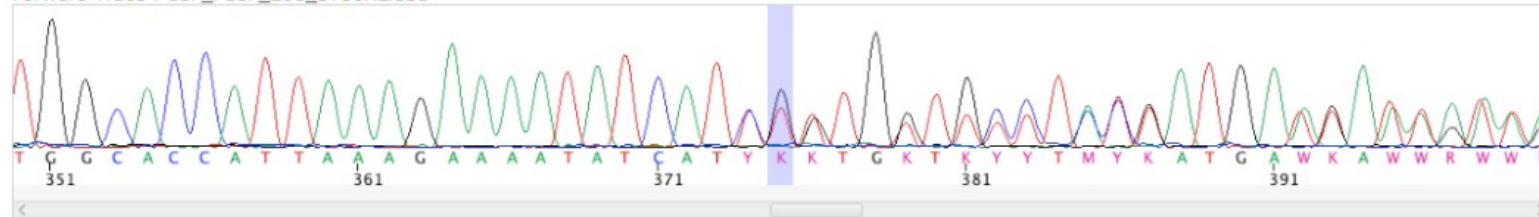
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11P_P11F_E08_3730XL.ab1



Reverse Trace : 11P_P11R_C11_3730XL.ab1

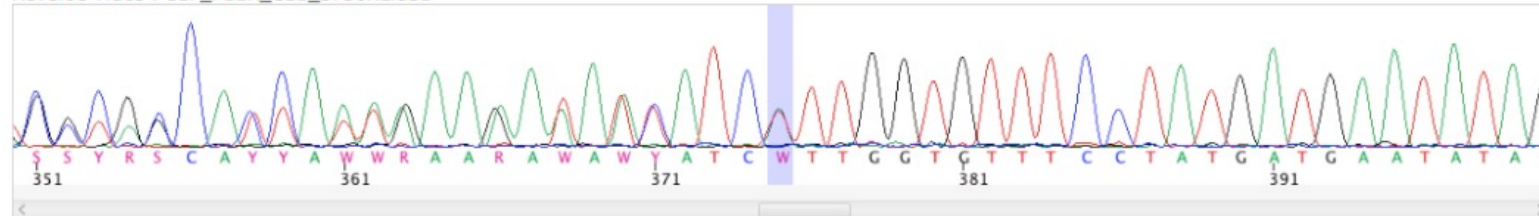


Figure 61: SnackVar report for the p.Phe508del variant in CF9862557

Hetero Indel View (Forward) : 11P_P11F_E08_3730XL.ab1



Hetero Indel View (Reverse) : 11P_P11R_C11_3730XL.ab1



Figure 62: SnackVar report for the p.Phe508del variant in CF9862557

SnackVar Report

ID : CF3796568; p.Phe508del

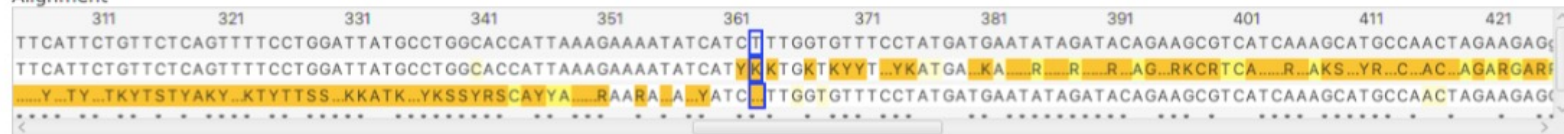
Description : Confirmation of p.Phe508del in CF3796568

Reference sequence : NM_000492.4(CFTR).fasta

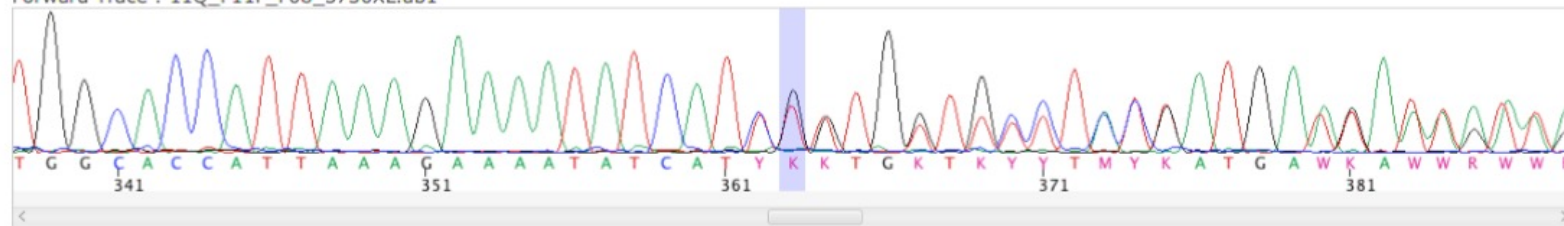
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11Q_P11F_F08_3730XL.ab1



Reverse Trace : 11Q_P11R_D11_3730XL.ab1

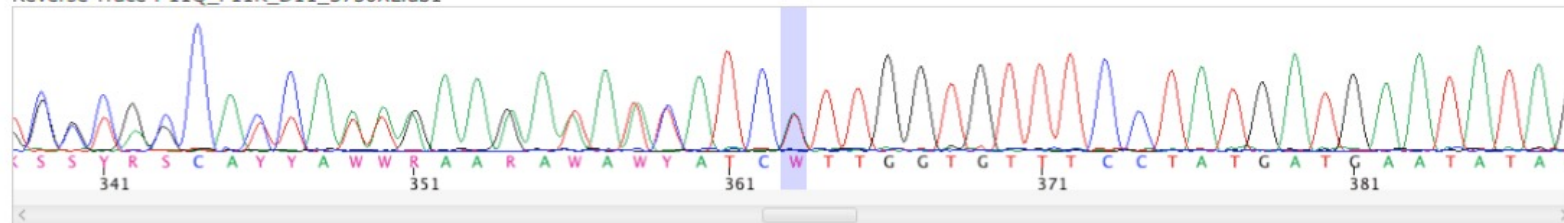
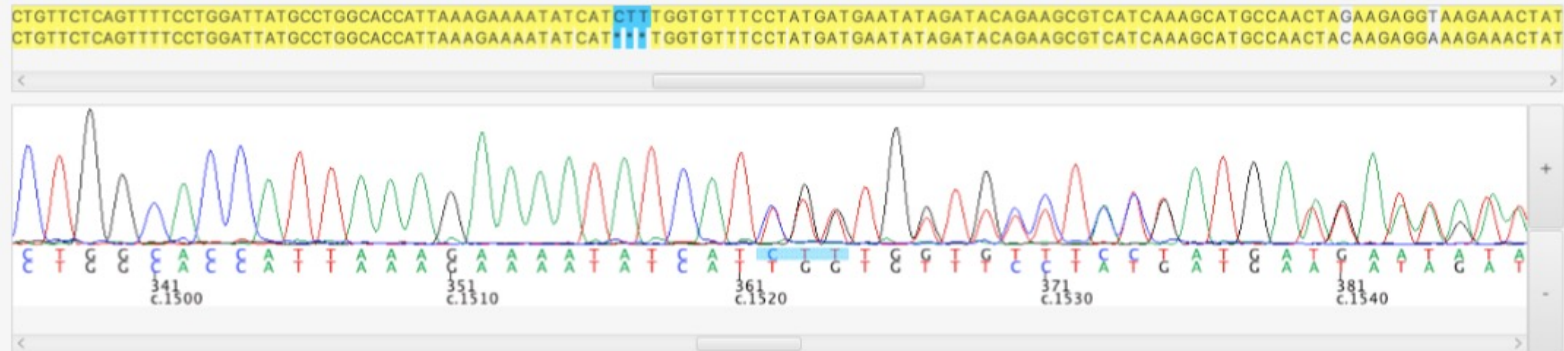


Figure 63: SnackVar report for the p.Phe508del variant in CF3796568

Hetero Indel View (Forward) : 11Q_P11F_F08_3730XL.ab1



Hetero Indel View (Reverse) : 11Q_P11R_D11_3730XL.ab1



Figure 64: SnackVar report for the p.Phe508del variant in CF3796568

SnackVar Report

ID : CF1133987; p.Phe508del

Description : Sanger confirmation of p.Phe508del CF1133987;

Reference sequence : NM_000492.4(CFTR).fasta

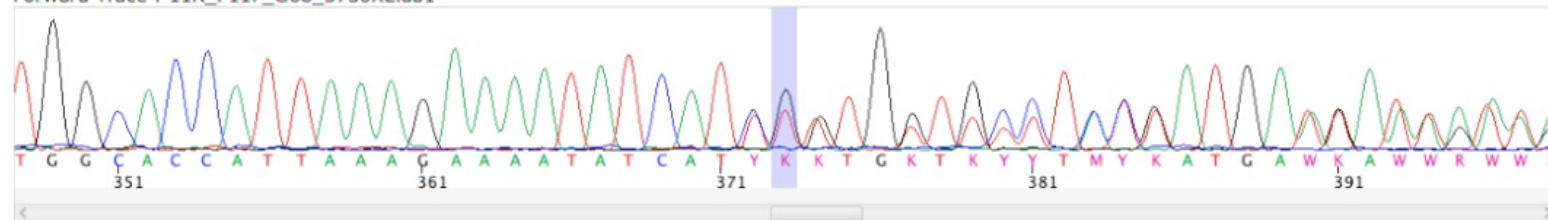
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment

Alignment view showing the reference sequence (NM_000492.4(CFTR).fasta) and the variant sequence (c.1521_1523del, p.(Phe508del), hetero). The variant is highlighted in yellow. The alignment shows a deletion of three nucleotides (1521-1523) in the variant sequence, which corresponds to the deletion of the Phe508 codon (TTC) in the protein sequence.

Forward Trace : 11R_P11F_G08_3730XL.ab1



Reverse Trace : 11R_P11R_E11_3730XL.ab1



Figure 65: SnackVar report for the p.Phe508del variant in CF1133987

Hetero Indel View (Forward) : 11R_P11F_G08_3730XL.ab1



Hetero Indel View (Reverse) : 11R_P11R_E11_3730XL.ab1



Figure 66: SnackVar report for the p.Phe508del variant in CF1133987

SnackVar Report

ID : CF5107567; p.Phe508del

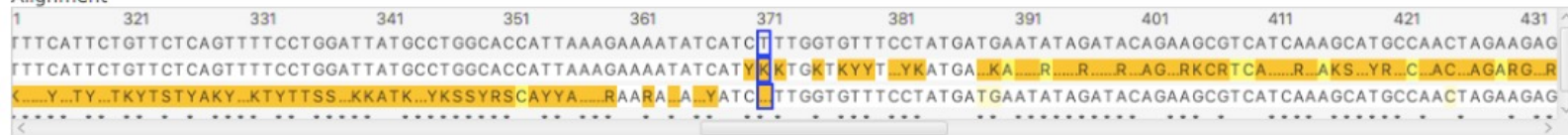
Description : Confirmation of p.Phe508del CF5107567

Reference sequence : NM_000492.4(CFTR).fasta

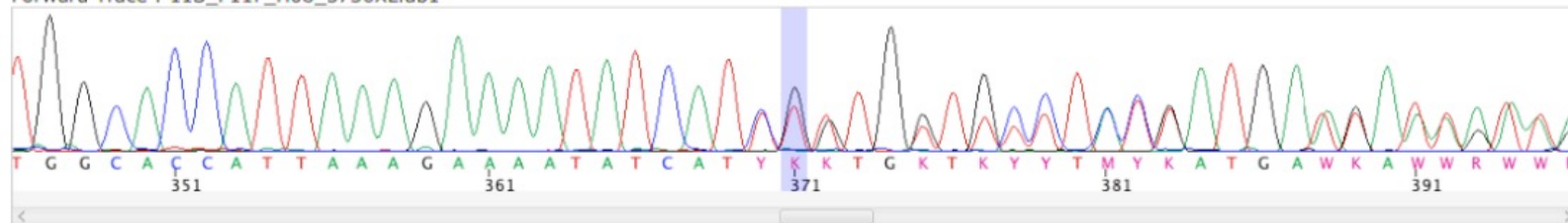
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11S_P11F_H08_3730XL.ab1



Reverse Trace : 11S_P11R_F11_3730XL.ab1

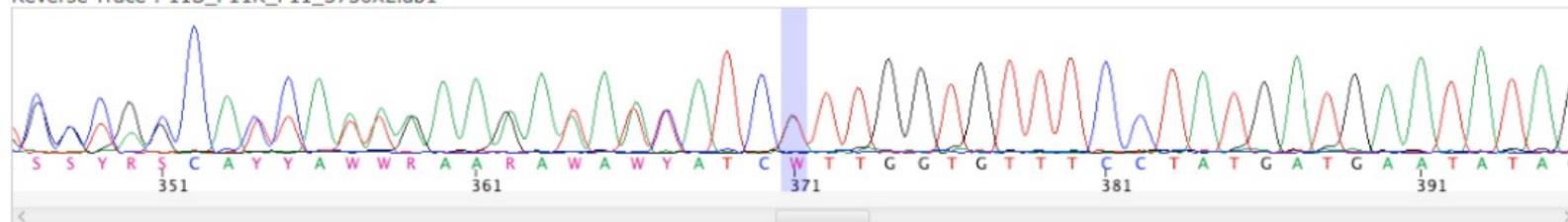
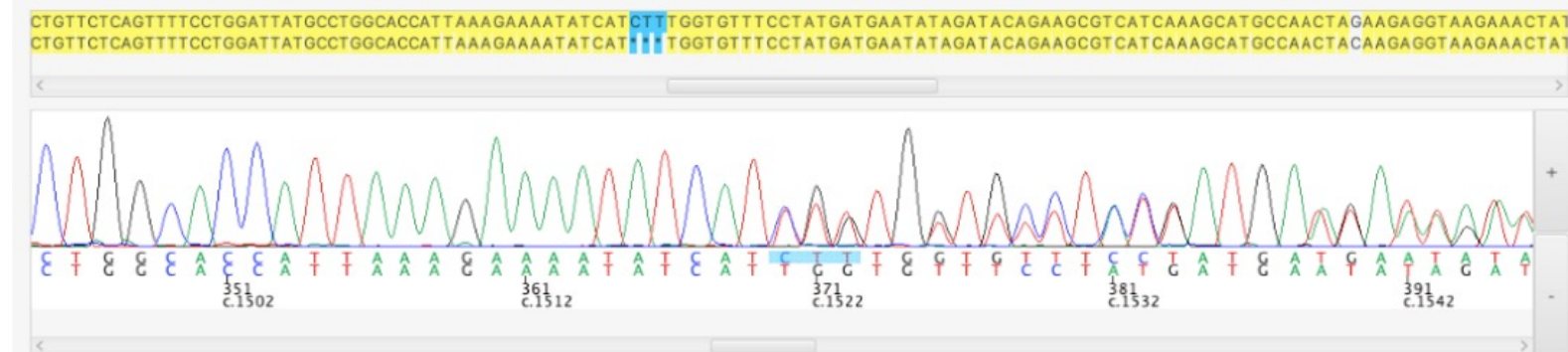


Figure 67: SnackVar report for the p.Phe508del variant in CF5107567

Hetero Indel View (Forward) : 11S_P11F_H08_3730XL.ab1



Hetero Indel View (Reverse) : 11S_P11R_F11_3730XL.ab1



Figure 68: SnackVar report for the p.Phe508del variant in CF5107567

SnackVar Report

ID : CF4495056; p.Phe508del

Description : Confirmation of p.Phe508del in CF4495056

Reference sequence : NM_000492.4(CFTR)..fasta

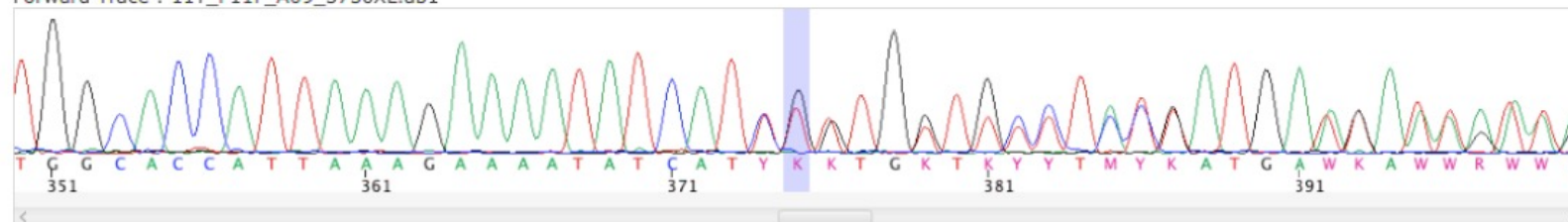
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment

321 331 341 351 361 371 381 391 401 411 421 431
TTCATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGC
TTCATTCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATYKKTGKTKYYT_YKATGA_KA_R_R_R_AG_RKCRTCA_R_AKS_YR_C_AS_AGARGARF
.....Y_TY...TKYTSTYAKY...KTYTSS_KKATK...YKSSYRSCAYYA.....RAARA_A_YATC...TTGGTGTTCCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGC

Forward Trace : 11T_P11F_A09_3730XL.ab1



Reverse Trace : 11T_P11R_G11_3730XL.ab1

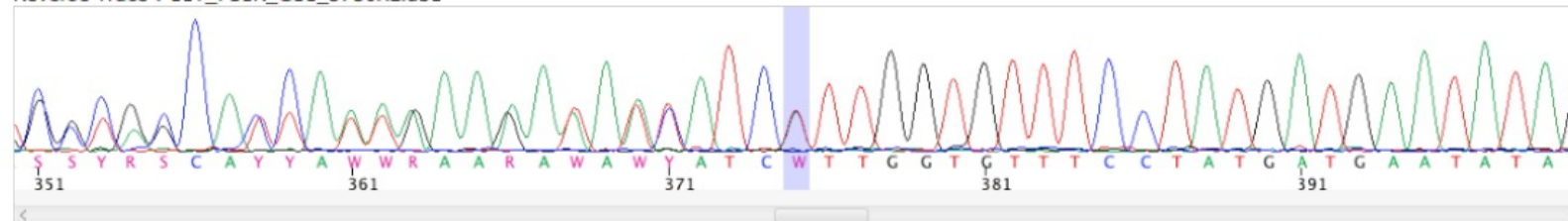
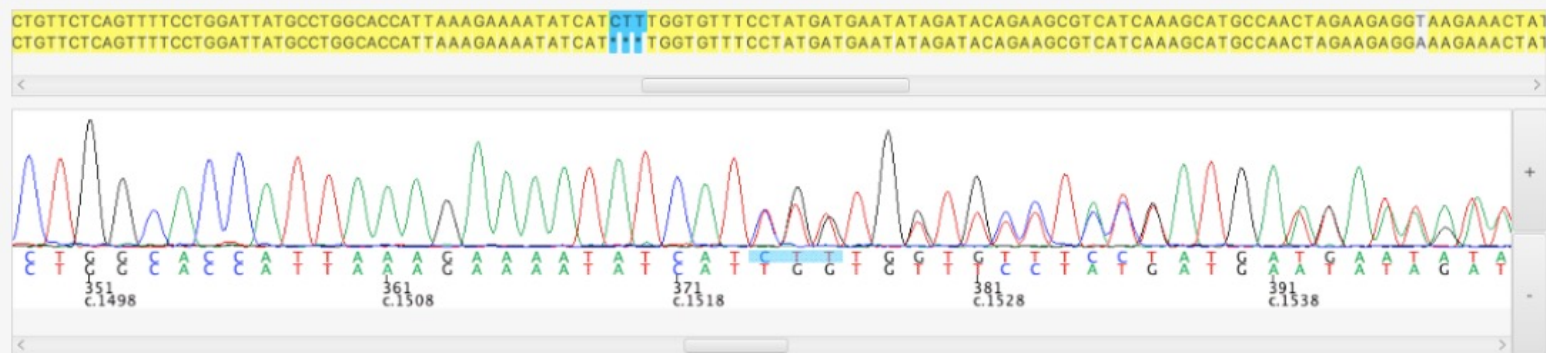


Figure 69: SnackVar report for the p.Phe508del variant in CF4495056

Hetero Indel View (Forward) : 11T_P11F_A09_3730XL.ab1



Hetero Indel View (Reverse) : 11T_P11R_G11_3730XL.ab1

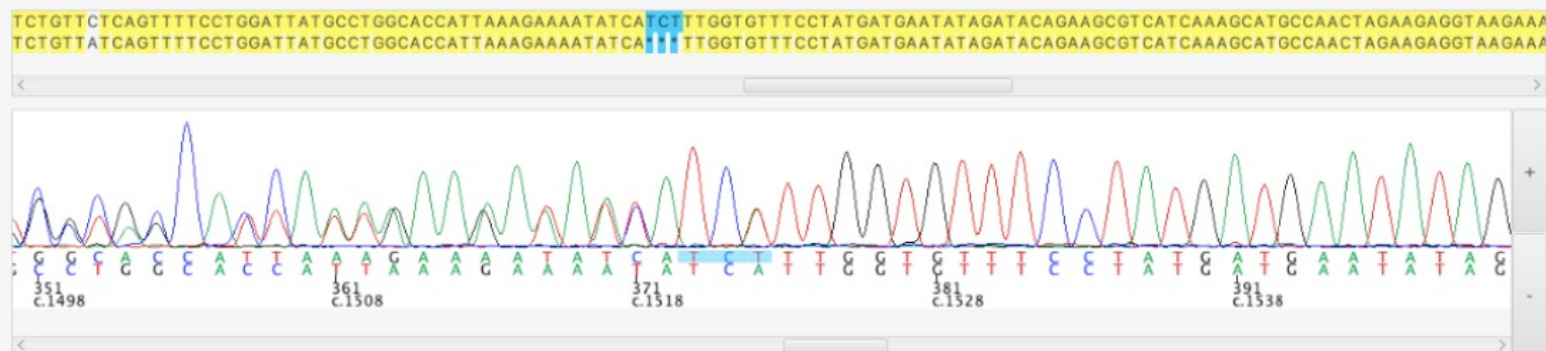


Figure 70: SnackVar report for the p.Phe508del variant in CF4495056

SnackVar Report

ID : CF5980227; p.Phe508del

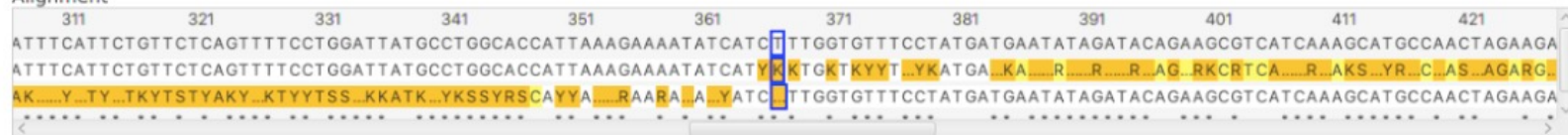
Description : Confirmation of p.Phe508del in CF5980227

Reference sequence : NM_000492.4(CFTR).fasta

Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11U_P11F_B09_3730XL.ab1



Reverse Trace : 11U_P11R_H11_3730XL.ab1

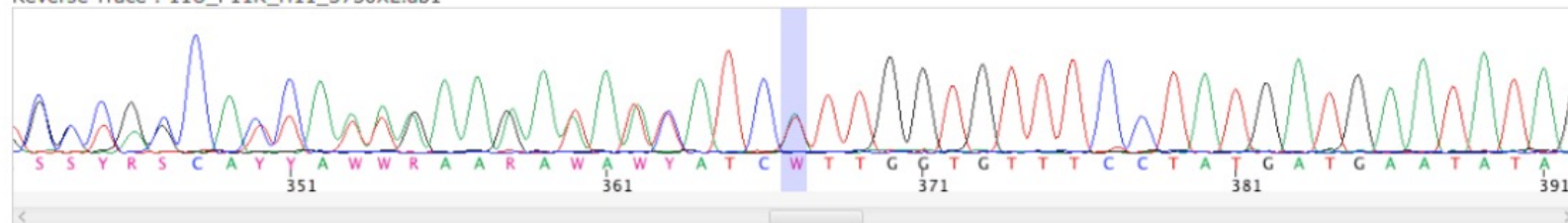
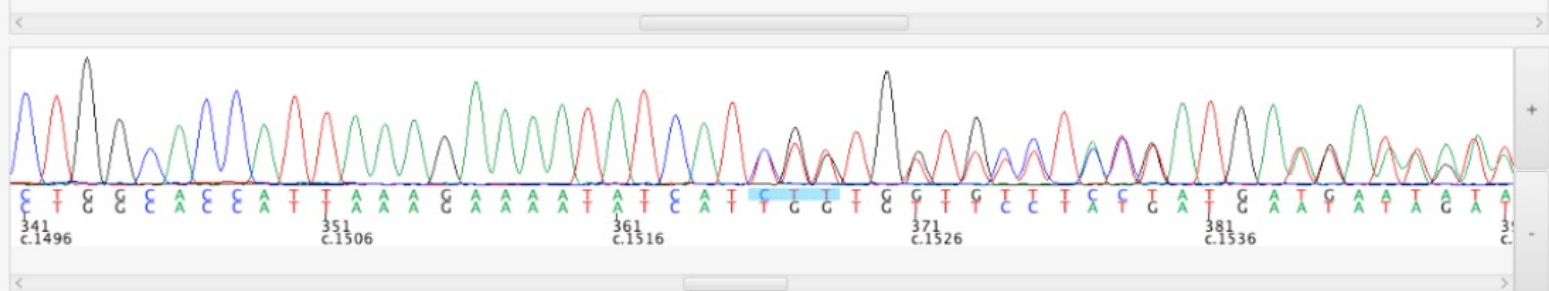


Figure 71: SnackVar report for the p.Phe508del variant in CF5980227

CTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTTCTTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAAGAACTA
CTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTTCTTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAAGAACTA



TCTGTTCTCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATCTTTGGTGTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAATCTGTTATCAGTTTTCTGGATTATGCCTGGCACCATTAAAGAAAATATCATTTTGGTGTTTCTATGATGAATATAGATACAGAAGCGTCATCAAAGCATGCCAACTAGAAGAGGTAAGAAAT

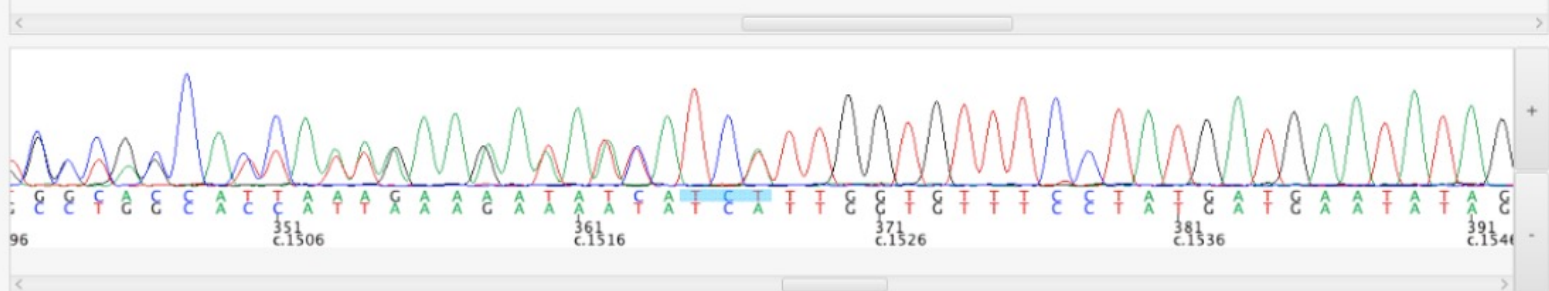


Figure 72: SnackVar report for the p.Phe508del variant in CF5980227

SnackVar Report

ID : CF6268769; p.Phe508del

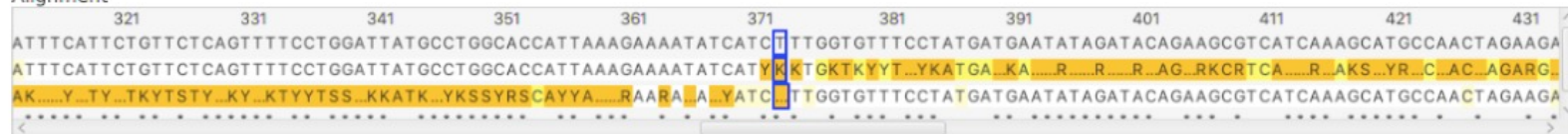
Description : Confirmation of p.Phe508del in CF6268769

Reference sequence : NM_000492.4(CFTR).fasta

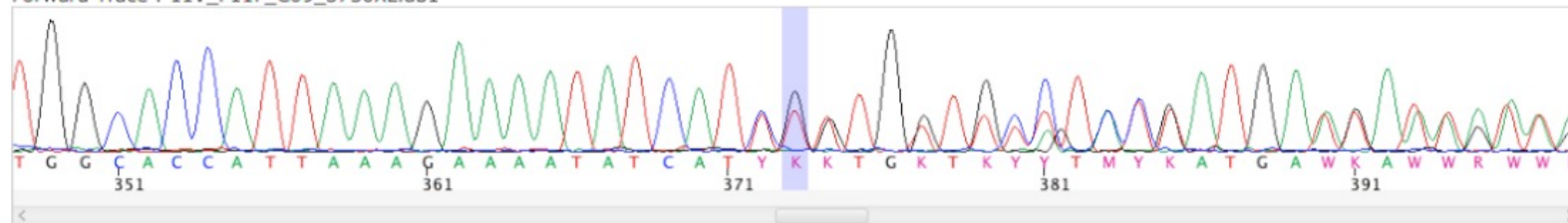
Variant 1

Description : c.1521_1523del, p.(Phe508del), hetero

Alignment



Forward Trace : 11V_P11F_C09_3730XL.ab1



Reverse Trace : 11V_P11R_A12_3730XL.ab1

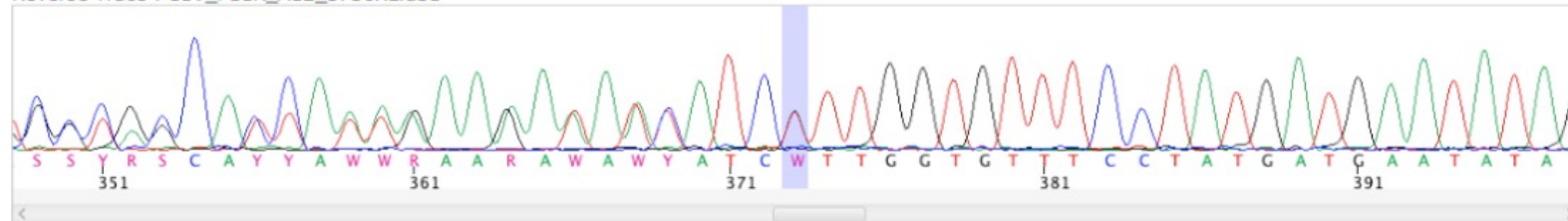


Figure 73: SnackVar report for the p.Phe508del variant in CF6268769

Hetero Indel View (Forward) : 11V_P11F_C09_3730XL.ab1



Hetero Indel View (Reverse) : 11V_P11R_A12_3730XL.ab1

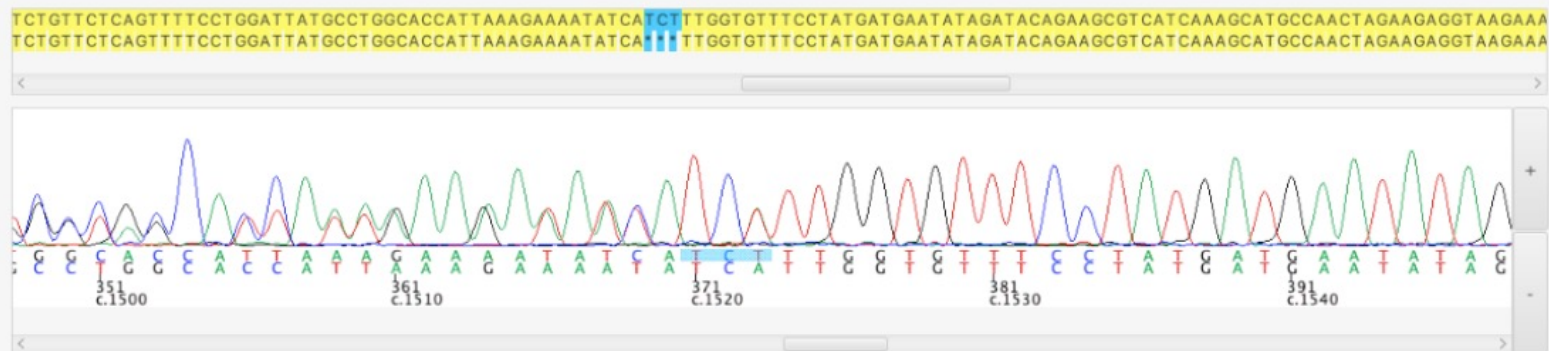


Figure 74: SnackVar report for the p.Phe508del variant in CF6268769

SnackVar Report

ID : CF2173052; p.Leu383*

Description : Confirmation of p.Leu383* in CF2173052

Reference sequence : NM_000492.4(CFTR).fasta

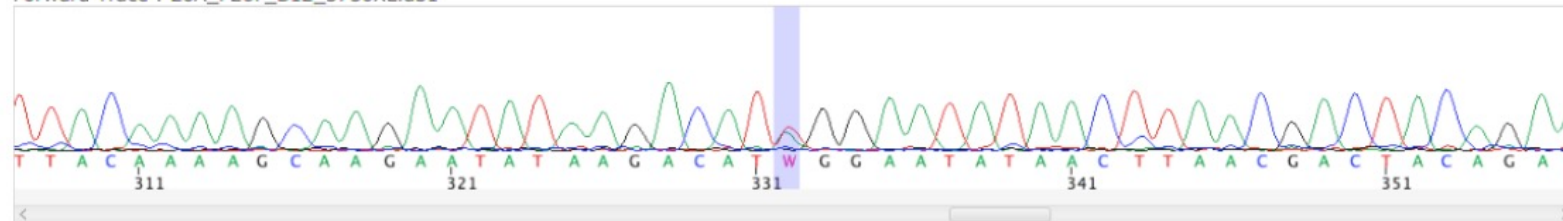
Variant 1

Description : c.1148T>A, p.(Leu383*), hetero

Alignment

```
281      291      301      311      321      331      341      351      361      371      381      391
t at gt t t t t g c t c t t t t a a a t a g G A T T T C T T A C A A A G C A A G A A T A T A A G A C A T T G G A A T A A C T T A A C G A C T A C A G A A G T A G T G A T G G A G A A T G T A A C A G C C T T C T G G G A G G A G g t
T A T G T T T T T G C T C T C T T T A T A A A T A G G A T T T C T T A C A A A G C A A G A A T A T A A G A C A T G G A A T A A C T T A A C G A C T A C A G A A G T A G T G A T G G A G A A T G T A A C A G C C T T C T G G G A G G A G G T
T A T G T T T T T G C T C T C T T T A T A A A T A G G A T T T C T T A C A A A G C A A G A A T A T A A G A C A T G G A A T A A C T T A A C G A C T A C A G A A G T A G T G A T G G A G A A T G T A A C A G C C T T C T G G G A G G A G G T
```

Forward Trace : 26A_P26F_B12_3730XL.ab1



Reverse Trace : 26A_P26R_C12_3730XL.ab1

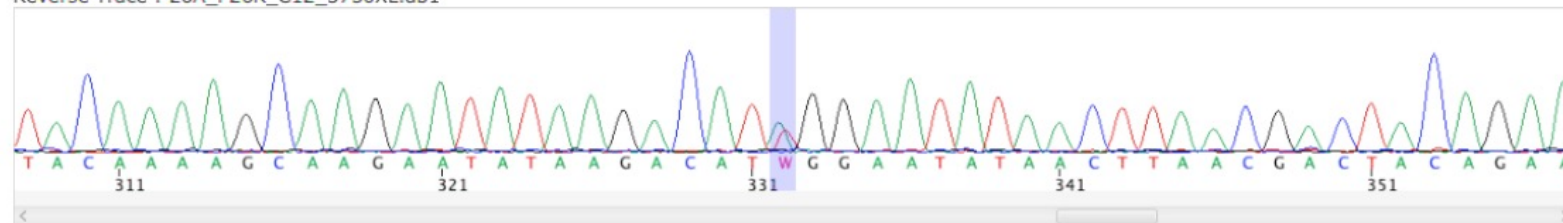


Figure 75: SnackVar report for the p.Leu383Ter variant in CF2173052



Figure 76: SnackVar report for the p.Trp57Leu position in CA1615190



Figure 77: SnackVar report for the p.Trp57Leu position in CF3115703

SnackVar Report

ID : CF4602380; p.Arg75Ter

Description : Confirmation of p.Arg75Ter in CF4602380

Reference sequence : NM_000492.4(CFTR).fasta

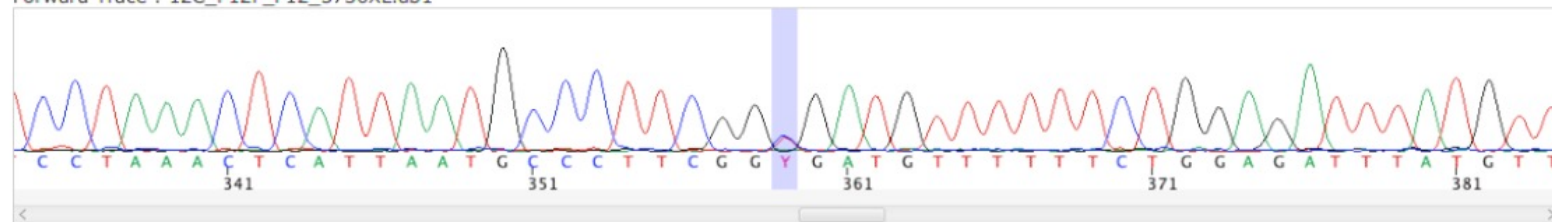
Variant 1

Description : c.223C>T, p.(Arg75*), hetero

Alignment

301	311	321	331	341	351	361	371	381	391	401	411	421
AGAATGGGATAGAGAGCTTGGCTTCAAAGAAAAATCCTAAACTCATTAATGCCCTTCGG	C	GATGTTTTTCTGGAGATTTATGTTCTATGGAATCTTTTATATTTAGGGgt aaggatctcat										
AGAATGGGATAGAGAGCTTGGCTTCAAAGAAAAATCCTAAACTCATTAATGCCCTTCGG	Y	GATGTTTTTCTGGAGATTTATGTTCTATGGAATCTTTTATATTTAGGGgt aaggatctcat										
AGAATGGGATAGAGAGCTTGGCTTCAAAGAAAAATCCTAAACTCATTAATGCCCTTCGG	Y	GATGTTTTTCTGGAGATTTATGTTCTATGGAATCTTTTATATTTAGGGgt aaggatctcat										

Forward Trace : 12C_P12F_F12_3730XL.ab1



Reverse Trace : 12C_P12R_C02_3730XL.ab1

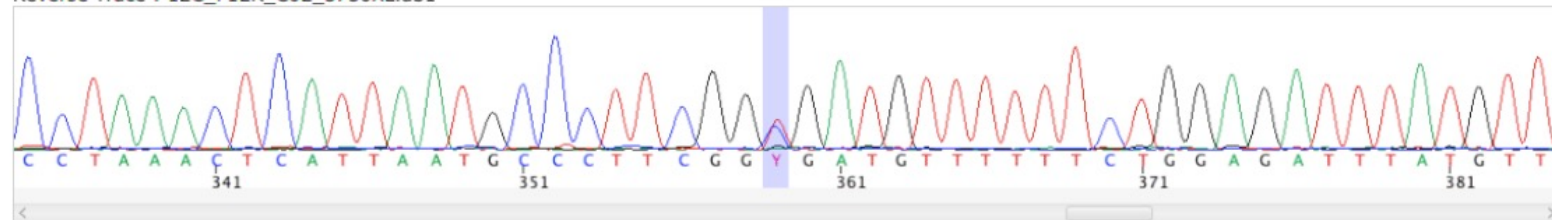


Figure 78: SnackVar report for the p.Arg75Ter variant in CF4602380



Figure 79: SnackVar report for the p.Trp57Leu position in CF0018616



Figure 80: SnackVar report for the p.Trp57Leu position in CF4544212



Figure 81: SnackVar report for the p.Trp57Leu position in CF2349244

SnackVar Report

ID : CF7527369; p.Leu88Ilefs*22

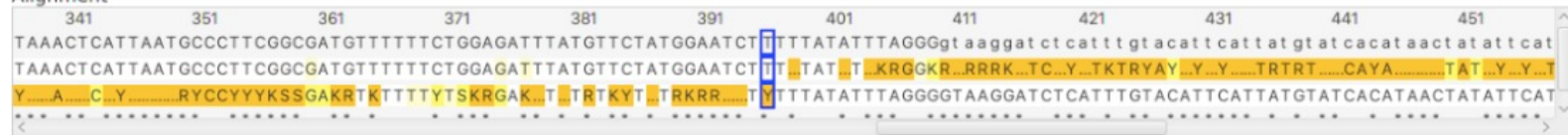
Description : Confirmation of p.Leu88Ilefs*22 in CF7527369

Reference sequence : NM_000492.4(CFTR).fasta

Variant 1

Description : c.262_263del, p.(Leu88Ilefs*22), hetero

Alignment



Forward Trace : 12G_P12F_B01_3730XL.ab1



Reverse Trace : 12G_P12R_F03_3730XL.ab1

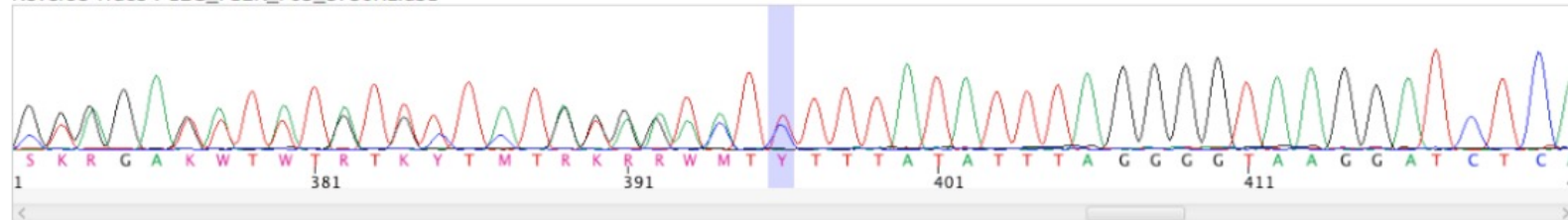


Figure 82: SnackVar report for the p.Leu88IlefsTer22 variant in CF7527369

Hetero Indel View (Forward) : 12G_P12F_B01_3730XL.ab1



Hetero Indel View (Reverse) : 12G_P12R_F03_3730XL.ab1



Figure 83: SnackVar report for the p.Leu88IlefsTer22 variant in CF7527369



Figure 84: SnackVar report for the p.Trp57Leu position in CF9830825

SnackVar Report

ID : CF3803349; p.Leu88Ilefs*22

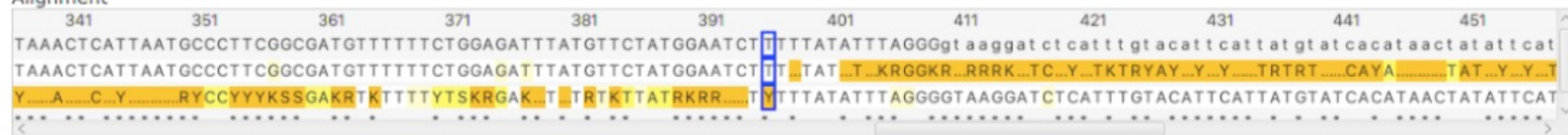
Description : Confirmation of p.Leu88Ilefs*22 in CF3803349

Reference sequence : NM_000492.4(CFTR).fasta

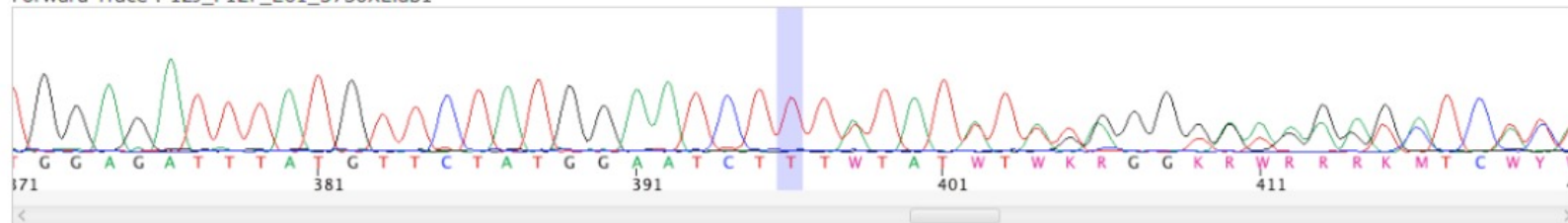
Variant 1

Description : c.262_263del, p.(Leu88Ilefs*22), hetero

Alignment



Forward Trace : 12J_P12F_E01_3730XL.ab1



Reverse Trace : 12J_P12R_B03_3730XL.ab1



Figure 86: SnackVar report for the p.Leu88IlefsTer22 variant in CF3803349

Hetero Indel View (Forward) : 12J_P12F_E01_3730XL.ab1



Hetero Indel View (Reverse) : 12J_P12R_B03_3730XL.ab1



Figure 87: SnackVar report for the p.Leu88IlefsTer22 variant in CF3803349

SnackVar Report

ID : CF1697504; p.Leu88Ilefs*22

Description : Confirmation of p.Leu88Ilefs*22 in CF1697504

Reference sequence : NM_000492.4(CFTR).fasta

Variant 1

Description : c.262_263del, p.(Leu88Ilefs*22), hetero

Alignment



Forward Trace : 12K_P12F_F01_3730XL.ab1



Reverse Trace : 12K_P12R_C03_3730XL.ab1

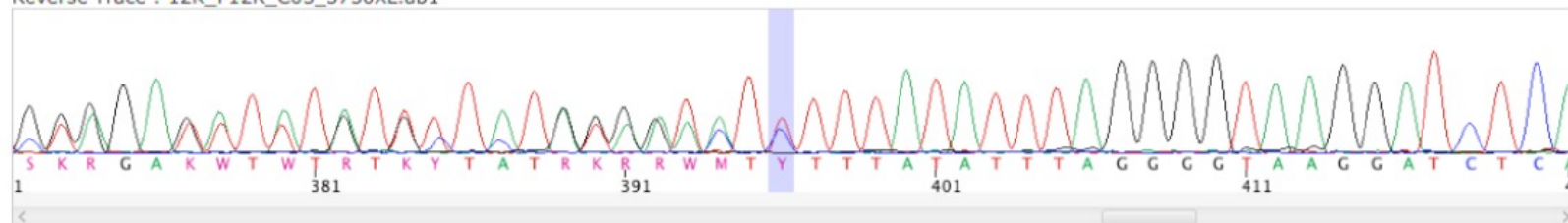


Figure 88: SnackVar report for the p.Leu88IlefsTer22 variant in CF1697504

Hetero Indel View (Forward) : 12K_P12F_F01_3730XL.ab1



Hetero Indel View (Reverse) : 12K_P12R_C03_3730XL.ab1



Figure 89: SnackVar report for the p.Leu88IlefsTer22 variant in CF1697504



Figure 90: SnackVar report for the p.Trp57Leu position in CF1697504



Figure 91: SnackVar report for the p.Trp57Leu position in CF5830853



Figure 92: SnackVar report for the p.Trp57Leu position in CA4932026



Figure 93: SnackVar report for the c.1680-1G>T position in CF1697504



Figure 94: SnackVar report for the c.1680-1G>T position in CF2349244



Figure 95: SnackVar report for the c.1680-1G>T position in CF5365245



Figure 96: SnackVar report for the c.1680-1G>T position in CF5830853

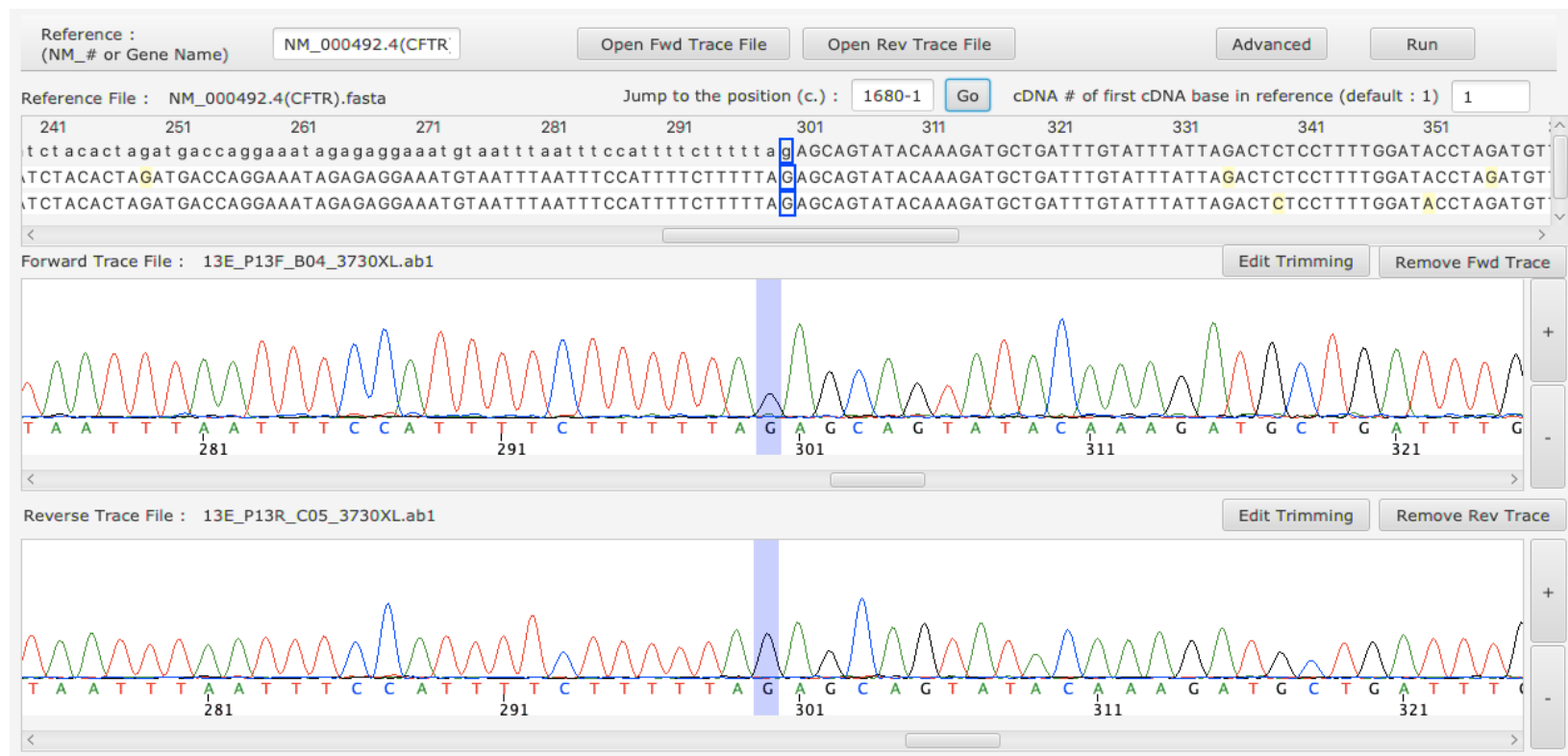


Figure 97: SnackVar report for the c.1680-1G>T position in CA4932026



Figure 98: SnackVar report for the c.1680-1G>T position in CF4833948



Figure 99: SnackVar report for the c.1680-1G>T position in CF4544212

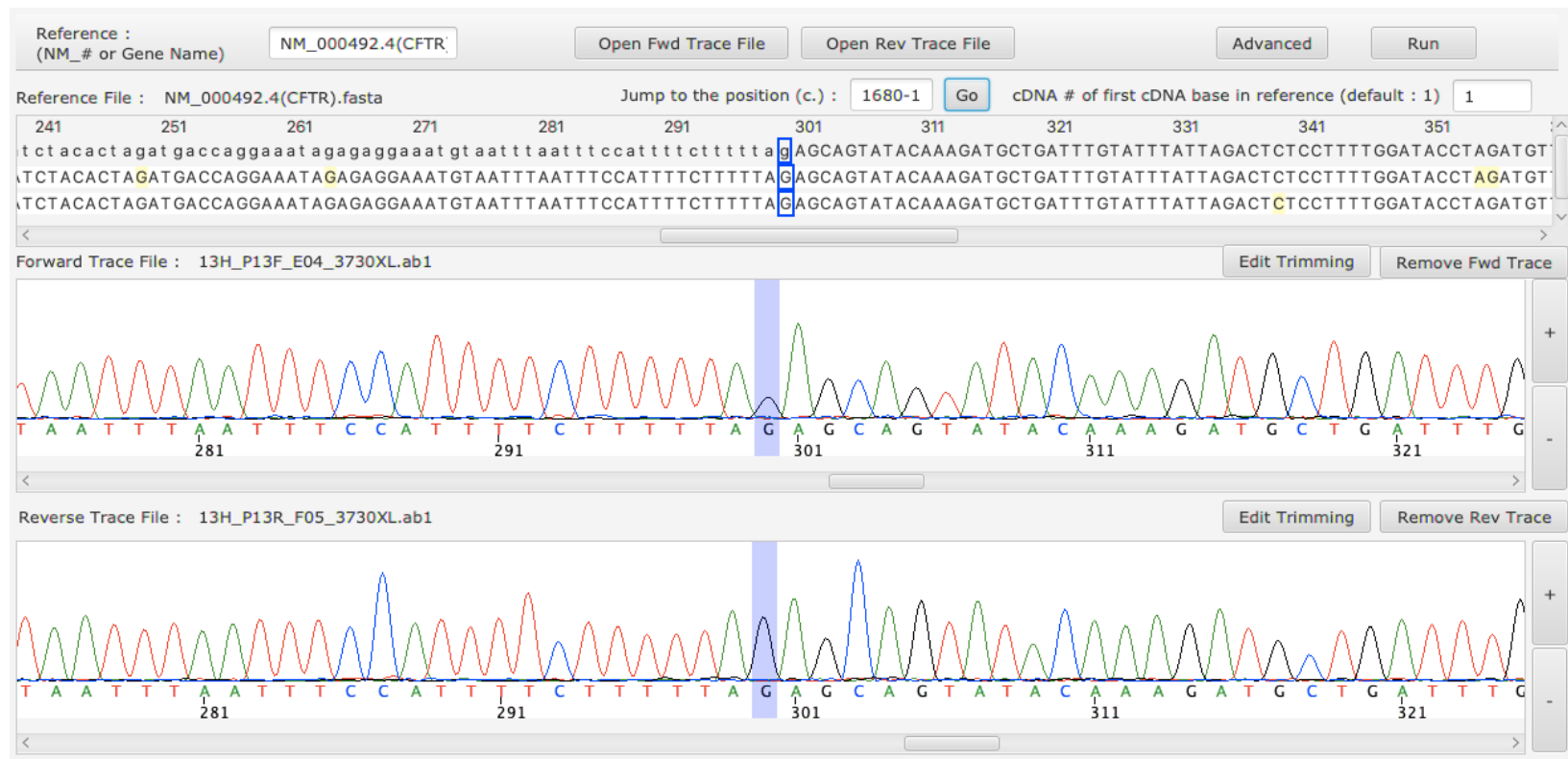


Figure 100: SnackVar report for the c.1680-1G>T position in CF3115703



Figure 101: SnackVar report for the c.1680-1G>T position in CF0018616

SnackVar Report

ID : CF3239825; p.Gln1382*

Description : Confirmation of p.Gln1382* in CF3239825

Reference sequence : NM_000492.4(CFTR).fasta

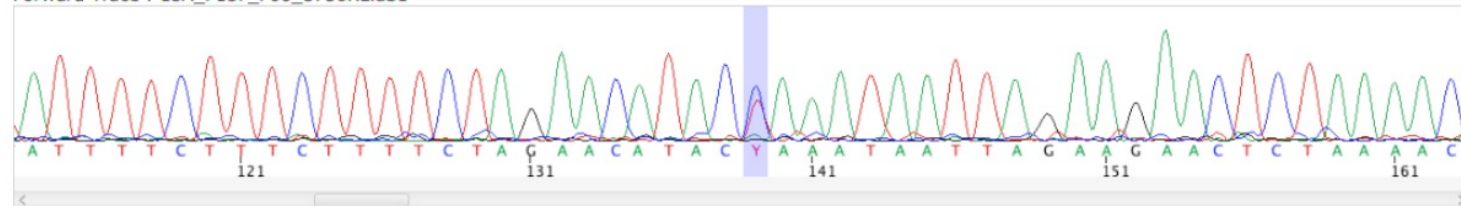
Variant 1

Description : c.4144C>T, p.(Gln1382*), hetero

Alignment

81 91 101 111 121 131 141 151 161 171 181 191
ica--gatcattactgttctgtgatattatgtgtggtatcttcttcttctagAACATACCAATAATTAGAAGAACTCTAAACAAGCATTGCTGATTGCACAGTAATTCTCTGTGAA
...C...TK...TCACTACTGTTCTGTGATATTATGTGGTATTTTCTTTCTTTCTAGAACATACYAAATAATTAGAAGAACTCTAAACAAGCATTGCTGATTGCACAG...AATTCTCTGTGAA
CA--GATCATTACTGTT...TGTGATA...TATGTGTGGTATTTTSTTTCTTTCTAGAACATA...YAAATAATTAGAAGAACTCTAAACAAGCATTGCTGATTGCACAGTAATT...TCTG...GAA

Forward Trace : 15A_P15F_F06_3730XL.ab1



Reverse Trace : 15A_P15R_C04_3730XL.ab1

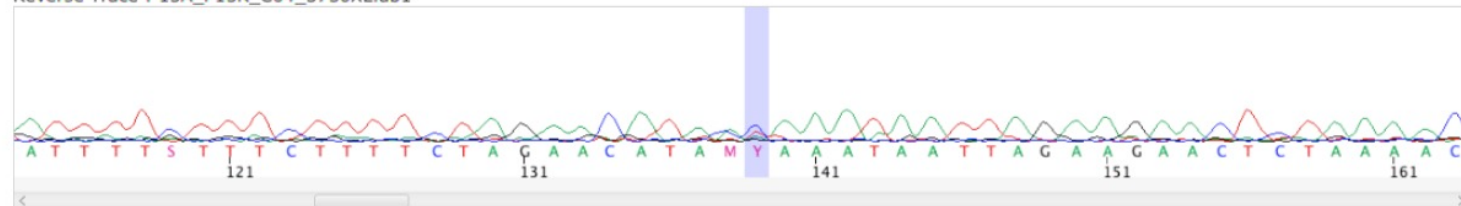


Figure 102: SnackVar report for the p.Gln1382X variant in CF3239825

SnackVar Report

ID : CF6757915; c.4242+1G>T

Description : Confirmation of c.4242+1G>T in CF6757915

Reference sequence : NM_000492.4(CFTR).fasta

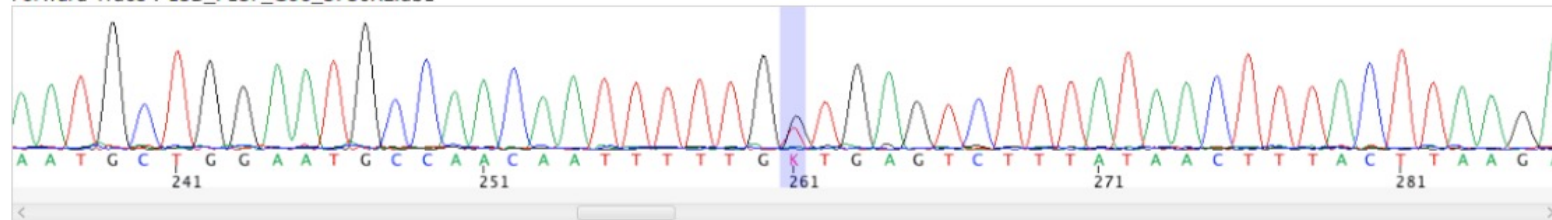
Variant 1

Description : c.4242+1G>T, p.?, hetero

Alignment

201 211 221 231 241 251 261 271 281 291 301 311 321
TTGCACAGTAATTCTCTGTGAACACAGGATAGAAGCAATGCTGGAATGCCAACAATTTTGGTgagtcctttataactttacttaagatctcattggcccttgtaattcttgataaacaatctca
TTGCACAGTAATTCTCTGTGAACACAGGATAGAAGCAATGCTGGAATGCCAACAATTTTGGTGTGAGTCTTTATAACTTTACTTAAGATCTCATTGCCCTTGTAAATTCTTGATAACAATCTCA
TTGCACAGTAATTCTCTGTGAACACAGGATAGAAGCAATGCTGGAATGCCAACAATTTTGGTGTGAGTCTTTATAACTTTACTTAAGATCTCATTGCCCTTGTAAATTCTTGATAACAATCTCA

Forward Trace : 15B_P15F_G06_3730XL.ab1



Reverse Trace : 15B_P15R_B07_3730XL.ab1

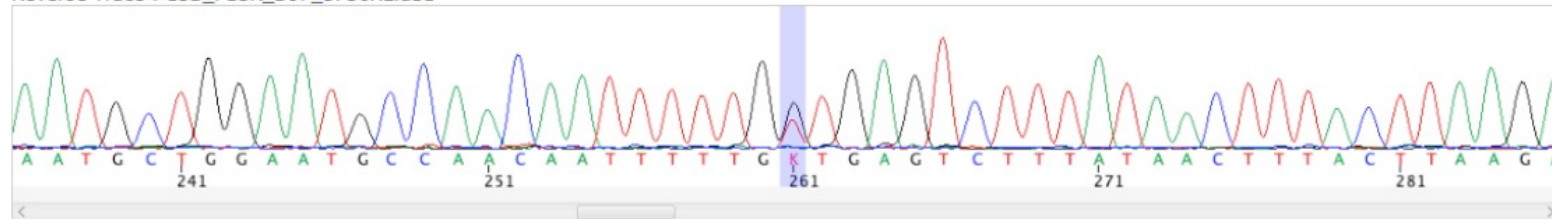


Figure 103: SnackVar report for the c.4242+1G>T variant in CF6757915

SnackVar Report

ID : CF2349244; p.Gln1411Pro

Description : Confirmation of p.Gln1411Pro in CF2349244

Reference sequence : NM_000492.4(CFTR).fasta

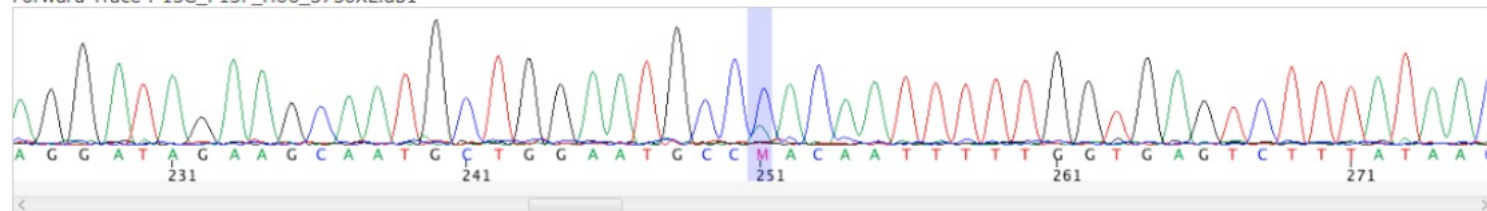
Variant 1

Description : c.4232A>C, p.(Gln1411Pro), hetero

Alignment

```
191      201      211      221      231      241      251      261      271      281      291      301      3
GCATTTGCTGATTGCACAGTAATTCTCTGTGAACACAGGATAGAAGCAATGCTGGAATGCCACAATTTTGGtgagtctttataactttacttaagatctcattgcccttgttaattcttga
GCATTTGCTGATTGCACAGTAATTCTCTGTGAACACAGGATAGAAGCAATGCTGGAATGCCACAATTTTGGTGAAGTCTTTATAACTTTACTTAAGATCTCATTGCCCTTGTAATTCTTGA
GCATTTGCTGATTGCACAGTAATTCTCTGTGAACACAGGATAGAAGCAATGCTGGAATGCCACAATTTTGGTGAAGTCTTTATAACTTTACTTAAGATCTCATTGCCCTTGTAATTCTTGA
```

Forward Trace : 15C_P15F_H06_3730XL.ab1



Reverse Trace : 15C_P15R_C07_3730XL.ab1

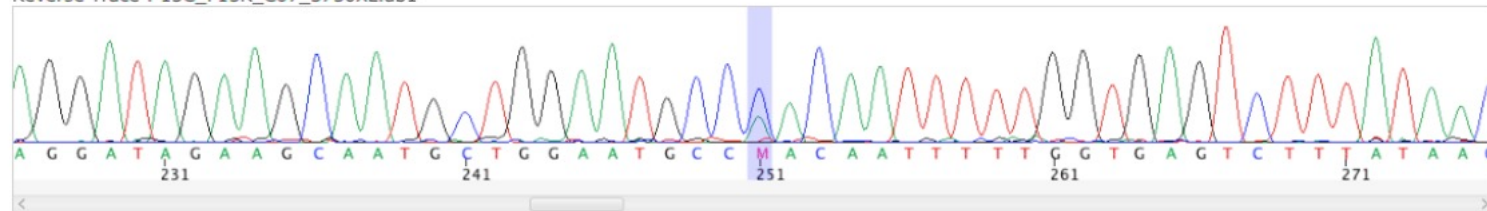


Figure 104: SnackVar report for the p.Gln1411Pro variant in CF2349244



Figure 105: SnackVar report for the p.Tyr627MetfsTer36 position in CF0235490



Figure 106: SnackVar report for the p.Tyr627MetfsTer36 position in CF3803349



Figure 107: SnackVar report for the p.Lys684AsnfsX38 position in CF7527369



Figure 108: SnackVar report for the p.Gly1125Ter position in CA4932026

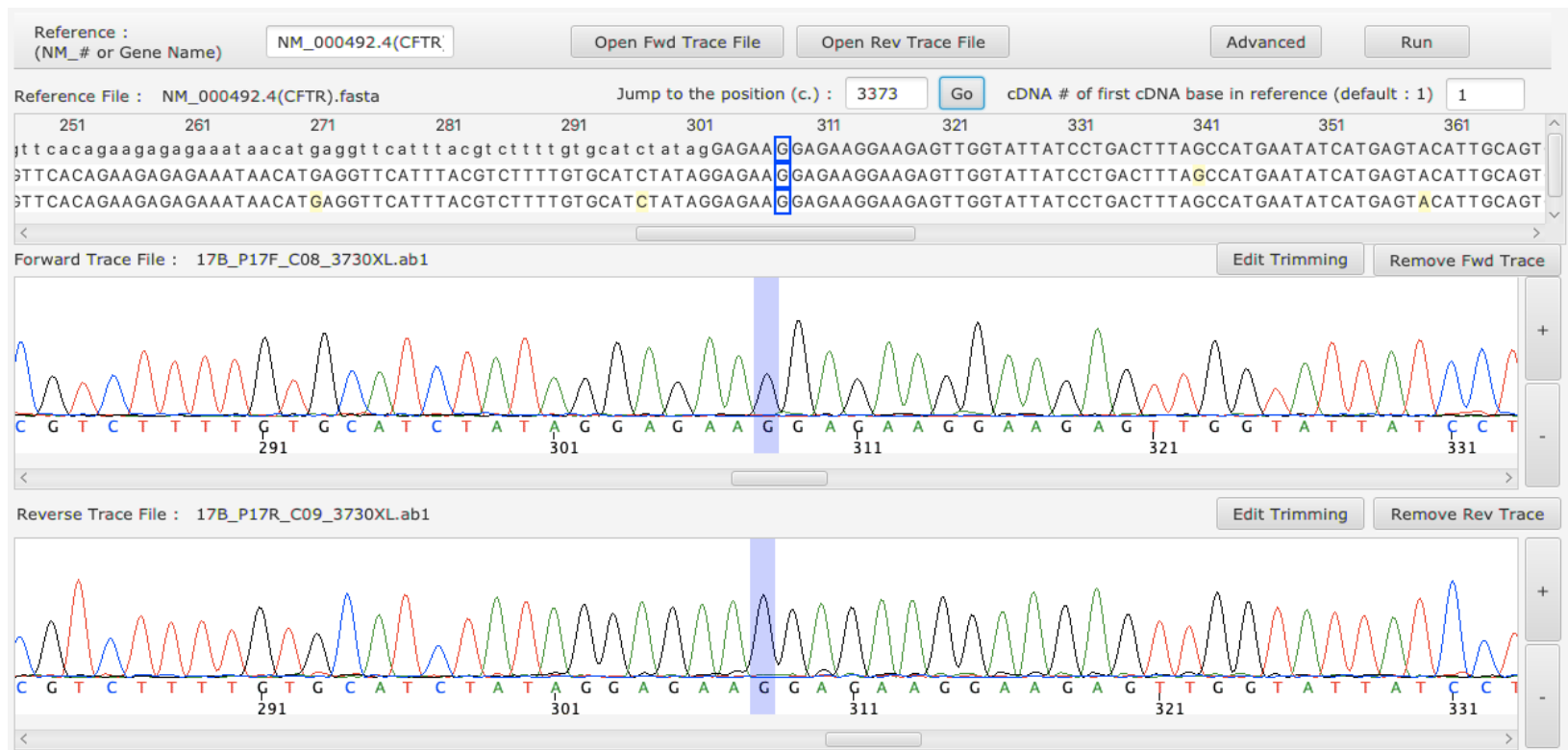


Figure 109: SnackVar report for the p.Gly1125Ter position in CF1697504

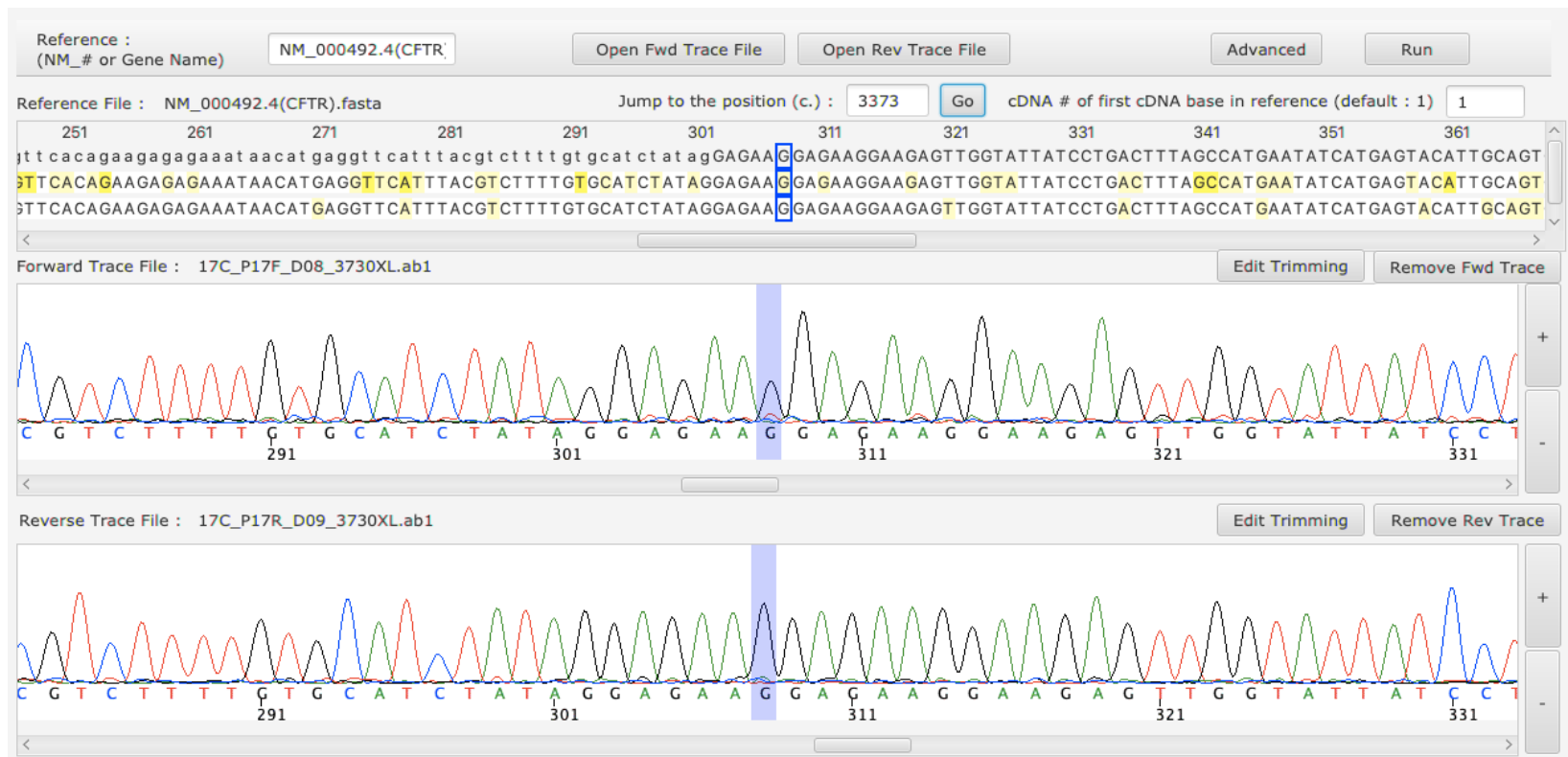


Figure 110: SnackVar report for the p.Gly1125Ter position in CF9830825



Figure 111: SnackVar report for the p.Gly1125Ter position in CF5181003



Figure 112: SnackVar report for the p.Gly1125Ter position in CF2349244

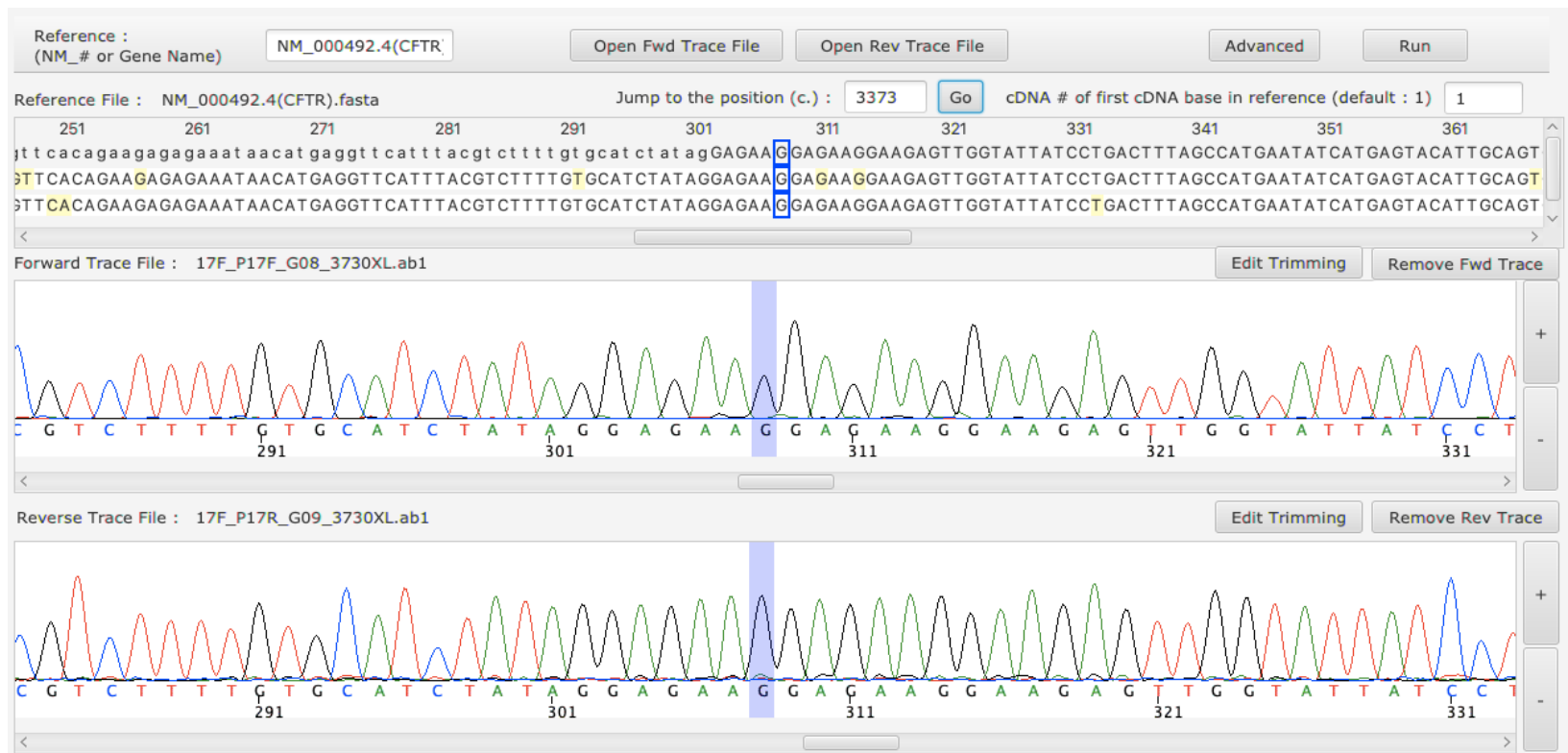


Figure 113: SnackVar report for the p.Gly1125Ter position in CF5830853



Figure 114: SnackVar report for the p.Gly1125Ter position in CF5158167

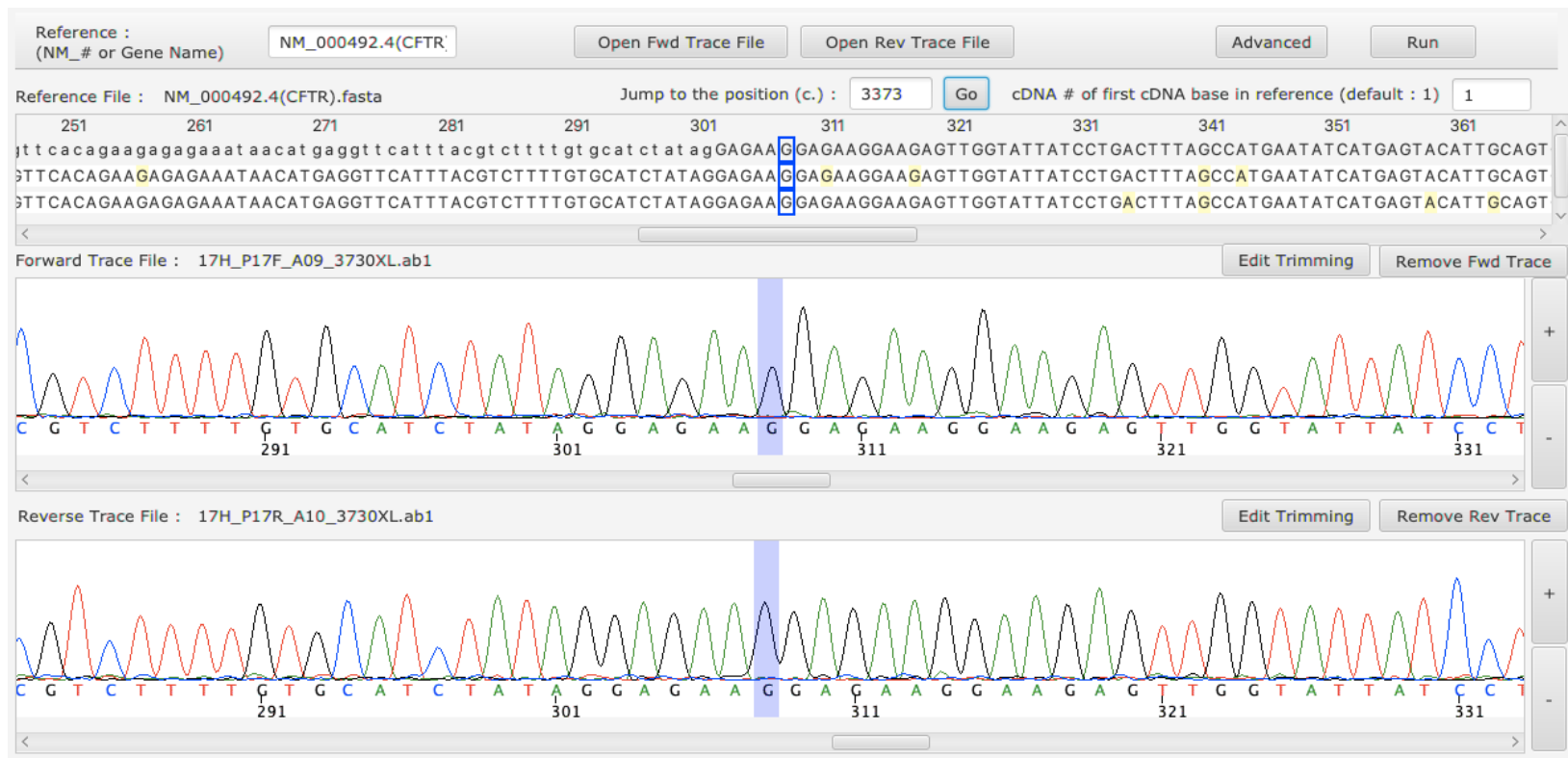


Figure 115: SnackVar report for the p.Gly1125Ter position in CF6803591

SnackVar Report

ID : CF2173052; p.Arg709*

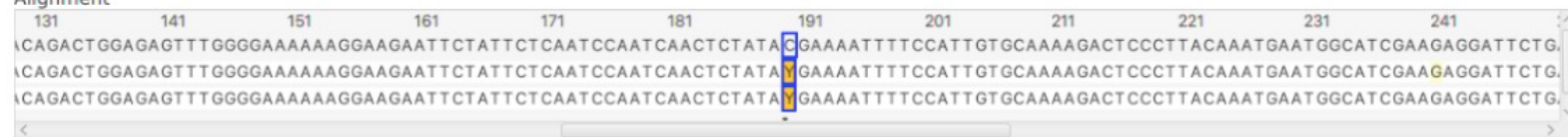
Description : Confirmation of the p.Arg709X variant in CF2173052

Reference sequence : NM_000492.4(CFTR).fasta

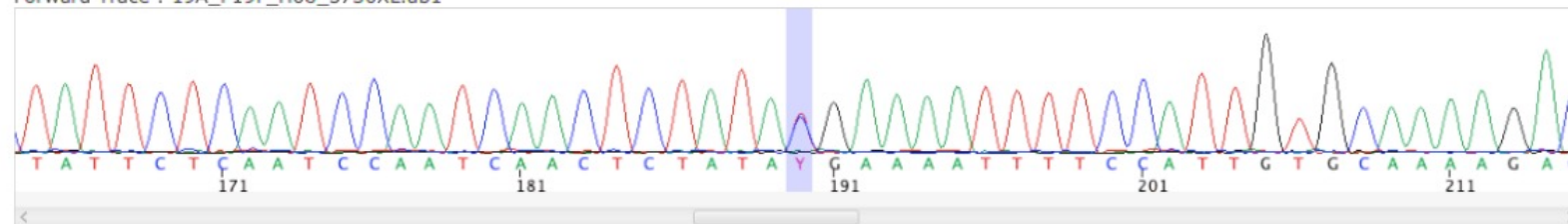
Variant 1

Description : c.2125C>T, p.(Arg709*), hetero

Alignment



Forward Trace : 19A_P19F_H08_3730XL.ab1



Reverse Trace : 19A_P19R_A09_3730XL.ab1

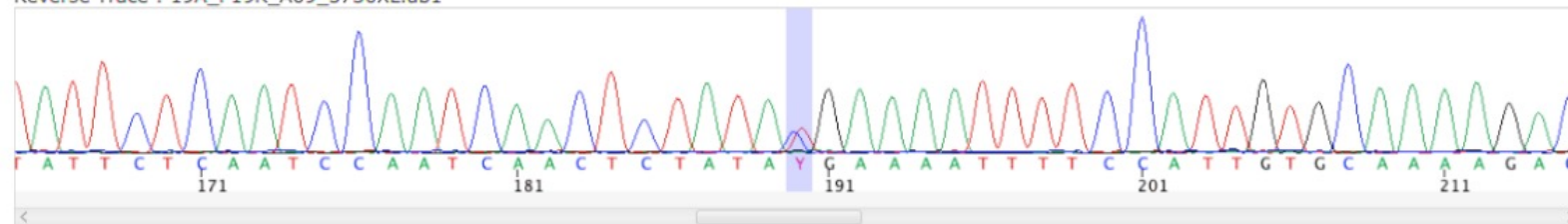


Figure 116: SnackVar report for the p.Arg709X variant in CF2173052

SnackVar Report

ID : CF2843425; p.Arg1158*

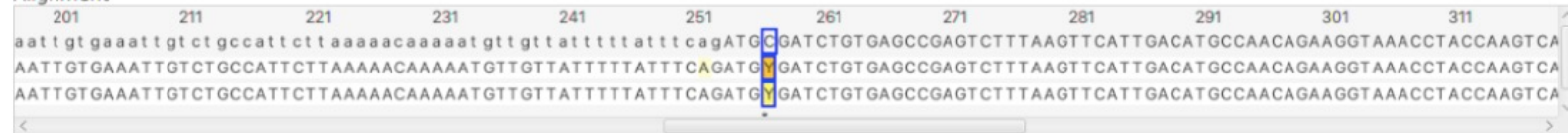
Description : Confirmation of p.Arg1158* in CF2843425

Reference sequence : NM_000492.4(CFTR).fasta

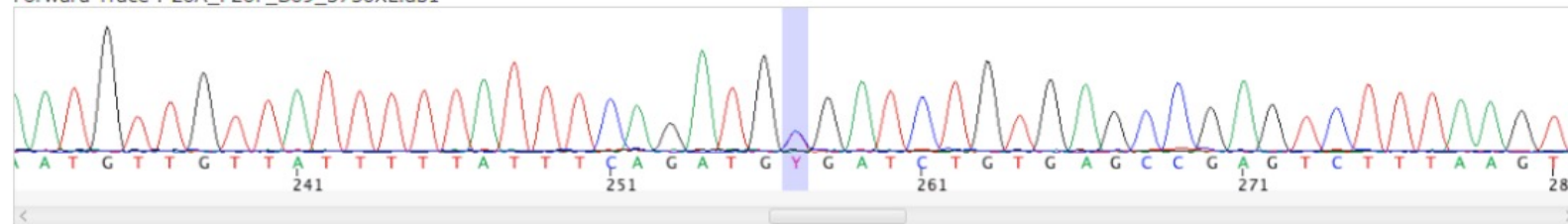
Variant 1

Description : c.3472C>T, p.(Arg1158*), hetero

Alignment



Forward Trace : 20A_P20F_B09_3730XL.ab1



Reverse Trace : 20A_P20R_C09_3730XL.ab1

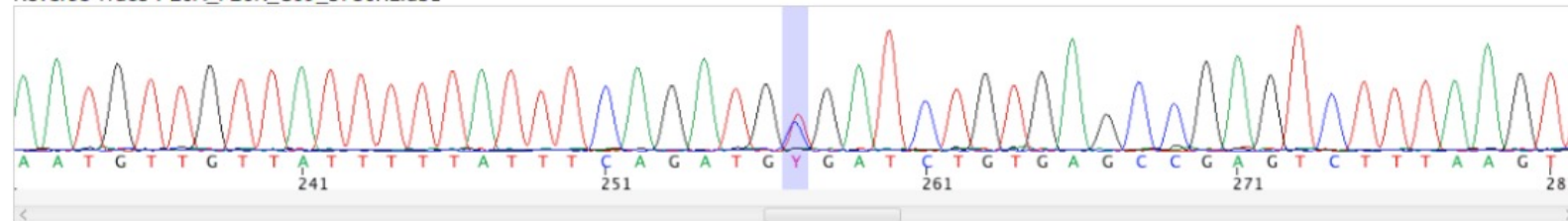


Figure 117: SnackVar report for the p.Arg1158X variant in CF2843425

SnackVar Report

ID : CF4495056; p.Ser877Phefs*29

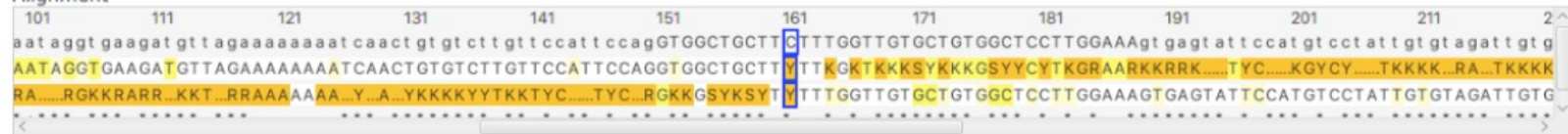
Description : Confirmation of p.Ser877Phefs*29 in CF4495056

Reference sequence : NM_000492.4(CFTR).fasta

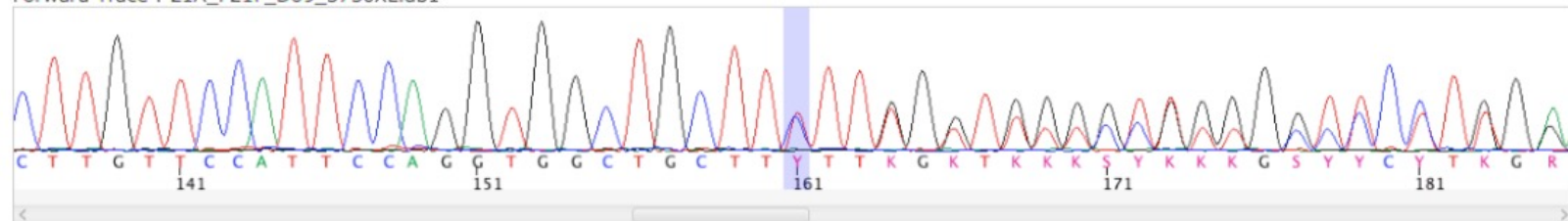
Variant 1

Description : c.2630del, p.(Ser877Phefs*29), hetero

Alignment



Forward Trace : 21A_P21F_D09_3730XL.ab1



Reverse Trace : 21A_P21R_E09_3730XL.ab1

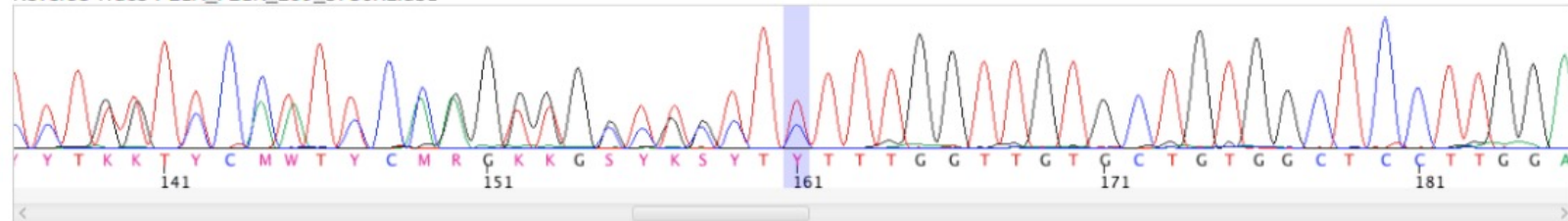
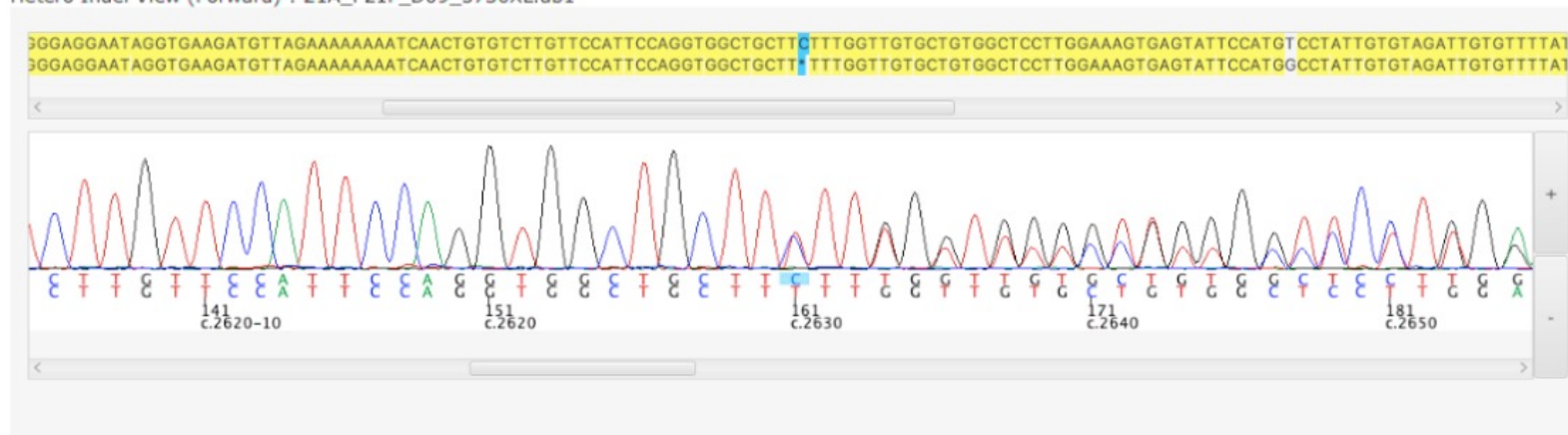


Figure 118: SnackVar report for the p.Ser877PhefsTer29 variant in CF4495056

Hetero Indel View (Forward) : 21A_P21F_D09_3730XL.ab1



Hetero Indel View (Reverse) : 21A_P21R_E09_3730XL.ab1



Figure 119: SnackVar report for the p.Ser877PhefsTer29 variant in CF4495056



Figure 120: SnackVar report for the c.4389 position in CF2433640



Figure 121: SnackVar report for the c.4389 position in CF2349244



Figure 122: SnackVar report for the c.4389 position in CF0018616



Figure 123: SnackVar report for the c.4389 position in CF0014912



Figure 124: SnackVar report for the c.4389 position in CF1782680



Figure 125: SnackVar report for the c.4389 position in CA8443975

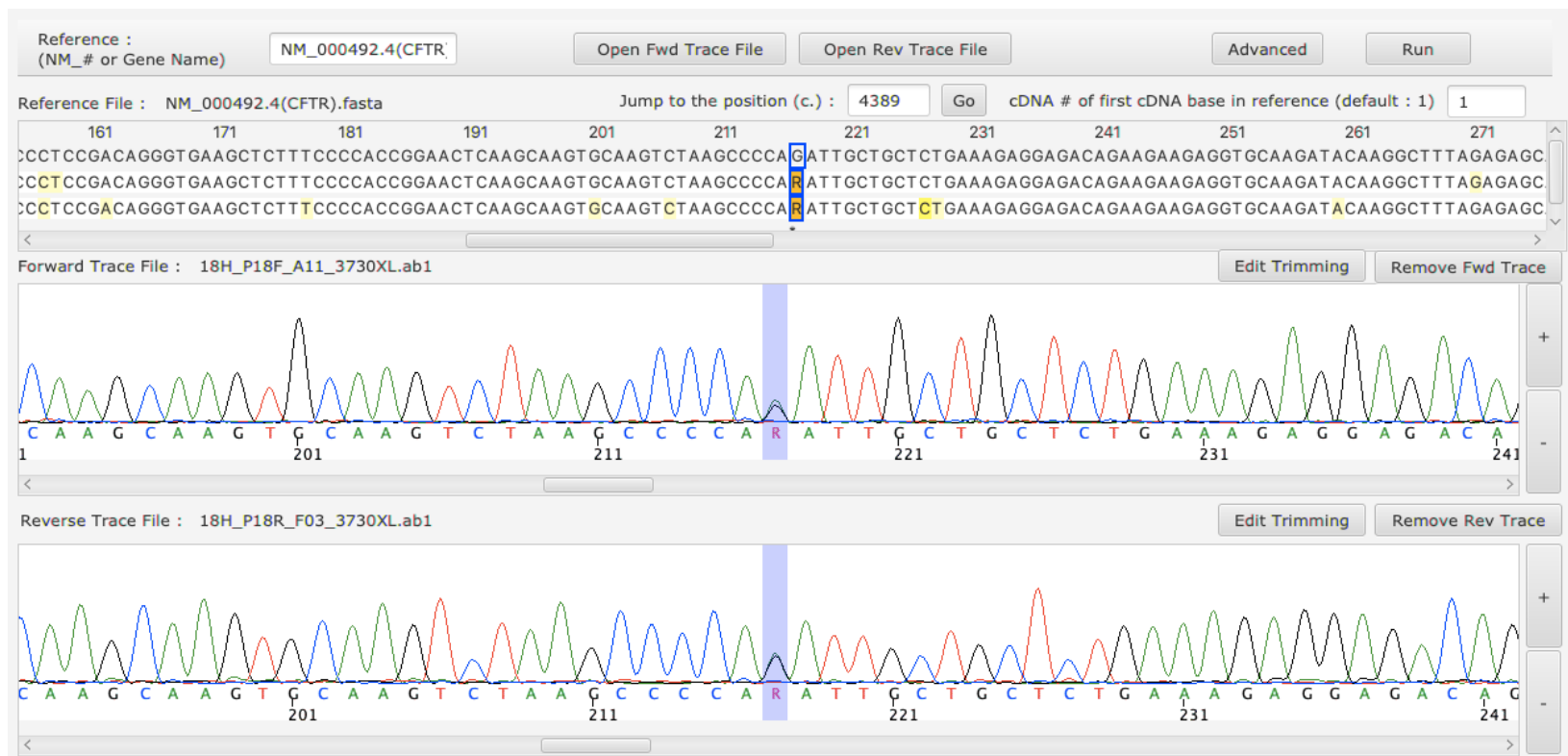


Figure 127: SnackVar report for the c.4389 position in CA1615190



Figure 128: SnackVar report for the c.4389 position in CF3719491



Figure 129: SnackVar report for the c.4389 position in CF3512286



Figure 130: SnackVar report for the c.4389 position in CF8754900



Figure 131: SnackVar report for the c.4389 position in CF5181003

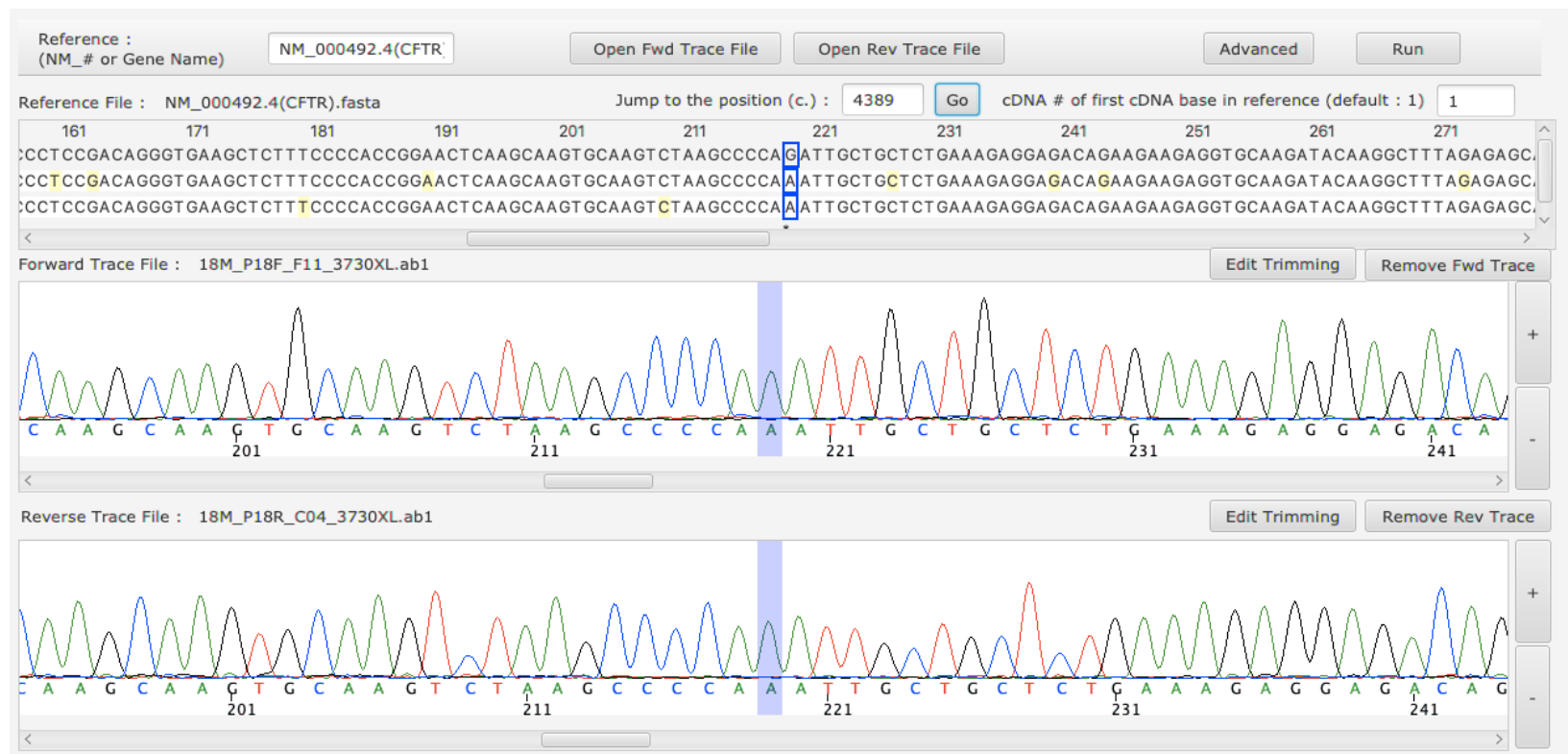


Figure 132: SnackVar report for the c.4389 position in CF4062212

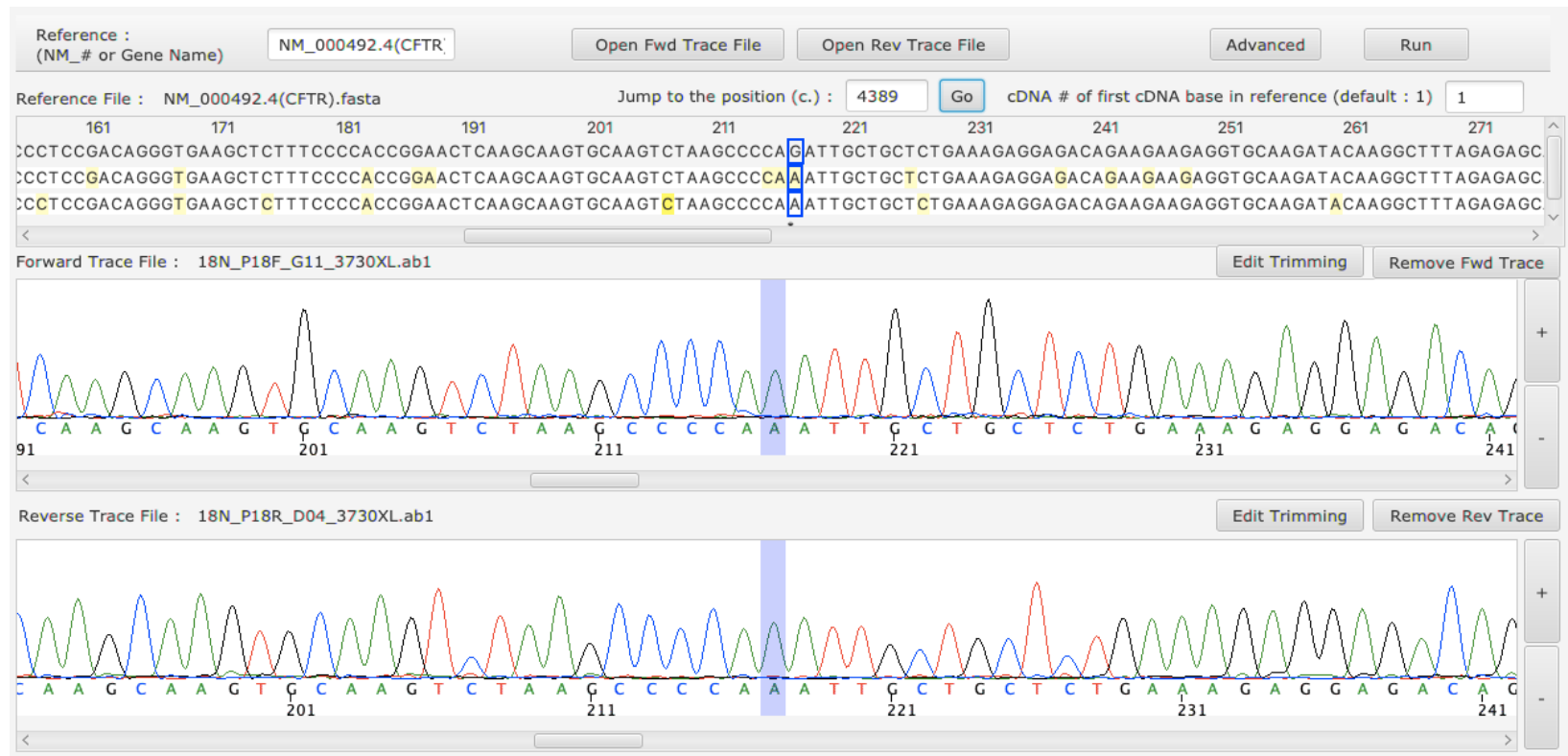


Figure 133: SnackVar report for the c.4389 position in CF6803591



Figure 134: SnackVar report for the c.4389 position in CF7778750

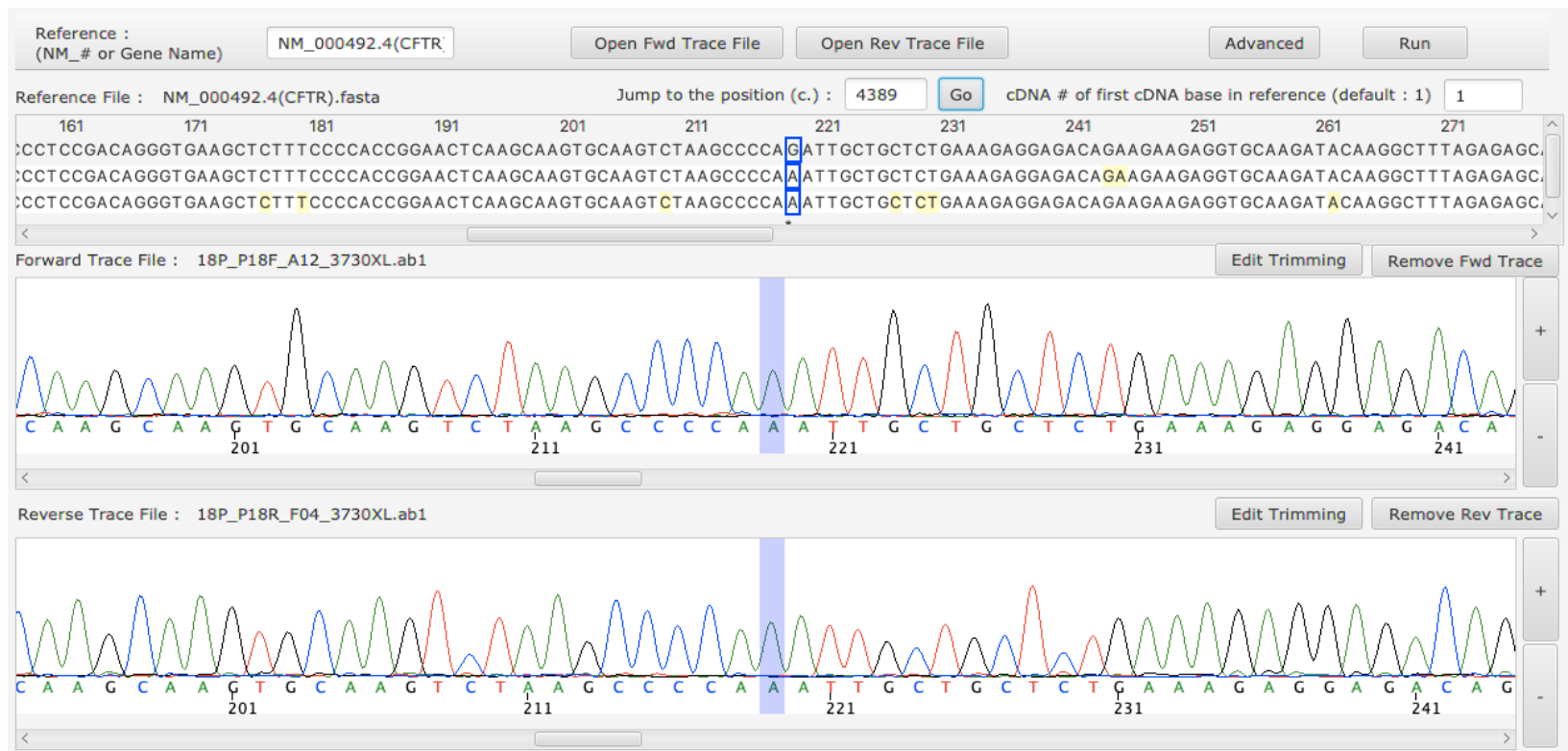


Figure 135: SnackVar report for the c.4389 position in CF5158167



Figure 136: SnackVar report for the c.4389 position in CF7600423

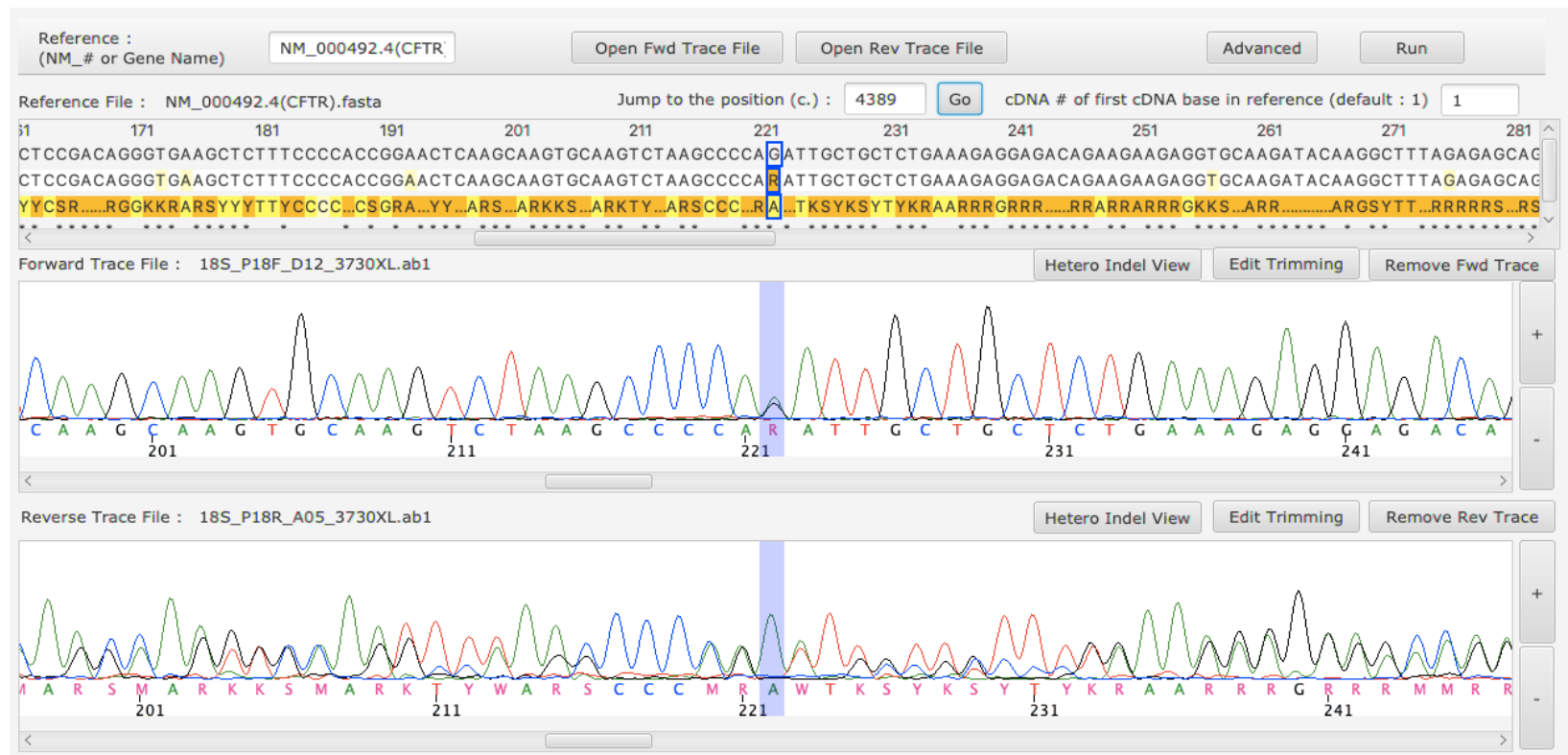


Figure 138: SnackVar report for the c.4389 position in CF3803349



Figure 139: SnackVar report for the c.4389 position in CF5107567

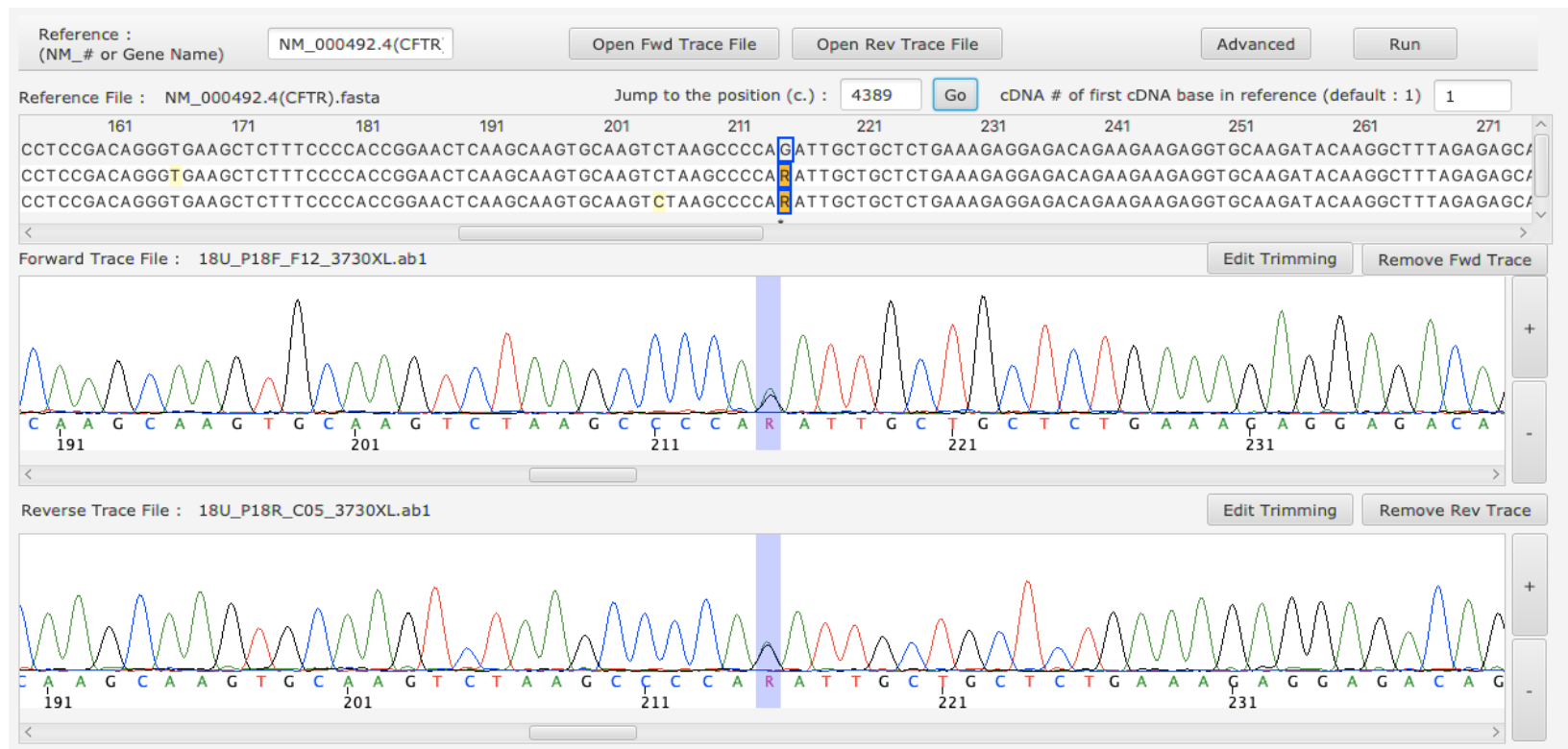


Figure 140: SnackVar report for the c.4389 position in CF5830853



Figure 141: SnackVar report for the c.4389 position in CF6268769



Figure 142: SnackVar report for the c.4389 position in CF6757915



Figure 143: SnackVar report for the c.4389 position in CF9442098

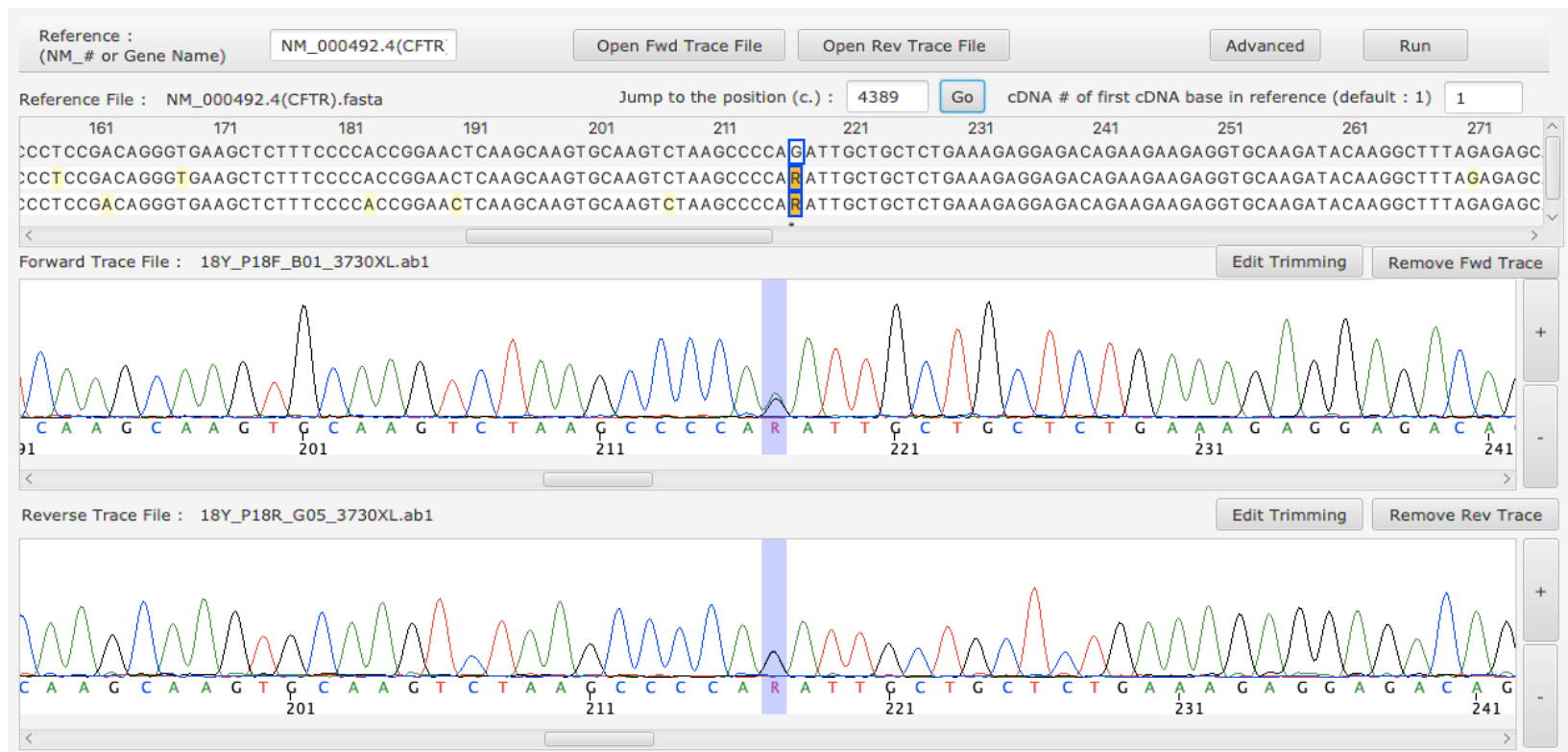


Figure 144: SnackVar report for the c.4389 position in CF5865254



Figure 145: SnackVar report for the c.4389 position in CF4223536



Figure 146: SnackVar report for the c.4389 position in CF9830825



Figure 147: SnackVar report for the c.4389 position in CF4869626



Figure 148: SnackVar report for the c.4389 position in CF4833948



Figure 149: SnackVar report for the c.4389 position in CF3796568



Figure 150: SnackVar report for the c.4389 position in CF5384911



Figure 151: SnackVar report for the c.4389 position in CF3239825



Figure 152: SnackVar report for the c.4389 position in CF1323468



Figure 153: SnackVar report for the c.4389 position in CF6188367



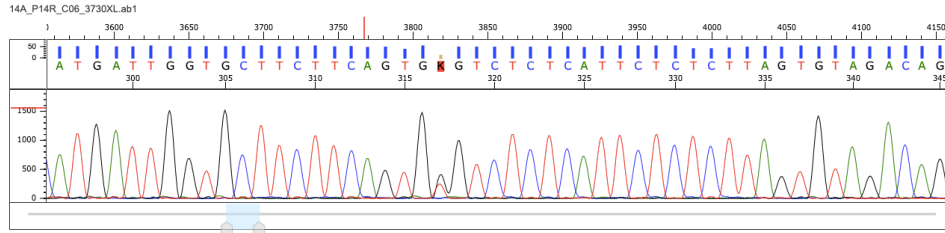
Figure 154: SnackVar report for the c.4389 position in CF6746590



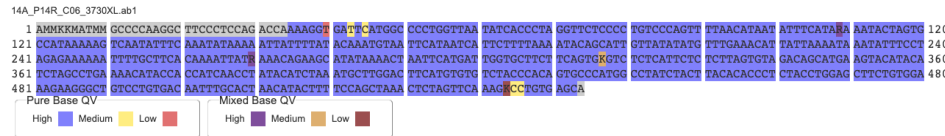
Figure 155: SnackVar report for the c.4389 position in CF4495056



Figure 156: SnackVar report for the c.4389 position in CF4283433



a) The reverse trace file at the location of the *1043A>C variant with “K” sequence base call visible in ThermoFisher Sanger Analysis software for CF4495056.



b) The sequence for the trace file with the *1043A>C variant with “K” base call visible in ThermoFisher Sanger Analysis software for CF4495056.

Figure 158: Sanger analysis of c.*1043A>C in CF4495056

Homo sapiens CF transmembrane conductance regulator (CFTR), mRNA

Sequence ID: [NM_000492.4](#) Length: 6070 Number of Matches: 1

Range 1: 5361 to 5904 [GenBank](#) [Graphics](#) [Next Match](#) [Previous Match](#)

Score	Expect	Identities	Gaps	Strand
983 bits(532)	0.0	540/545(99%)	2/545(0%)	Plus/Plus
Query 10	TTAGTGC-AATTGTCACAGGACAGCCCTTCTTTCCACAGAAGCTCCAGGTAGAGGGTGTG	68		
Sbjct 5361	TTAGTGCAAATTGTCACAGGACAGCCCTTCTTTCCACAGAAGCTCCAGGTAGAGGGTGTG	5420		
Query 69	TAAGTAGATAGGCCATGGGCACTGTGGGTAGACACACATGAAGTCCAAGCATTTAGATGT	128		
Sbjct 5421	TAAGTAGATAGGCCATGGGCACTGTGGGTAGACACACATGAAGTCCAAGCATTTAGATGT	5480		
Query 129	ATAGGTTGATGGTGGTATGTTTTCAGGCTAGATGTATGTACTTCATGCTGTCTACACTAA	188		
Sbjct 5481	ATAGGTTGATGGTGGTATGTTTTCAGGCTAGATGTATGTACTTCATGCTGTCTACACTAA	5540		
Query 189	GAGAGAATGAGAGACMCCTGGAAGAAGCACCAATCATGAATTAGTTTATATGCTTCTGT	248		
Sbjct 5541	GAGAGAATGAGAGACACTGGAAGAAGCACCAATCATGAATTAGTTTATATGCTTCTGT	5600		
Query 249	TTYATAATTTTGTAAGCAAAATTTTCTCTAGGAAATATTTATTTAATAATGTTTCA	308		
Sbjct 5601	TTTATAATTTTGTAAGCAAAATTTTCTCTAGGAAATATTTATTTAATAATGTTTCA	5660		
Query 309	AACATATATAACAATGCTGTATTTTAAAGAATGATTATGAATTACATTTGTATAAAATA	368		
Sbjct 5661	AACATATATAACAATGCTGTATTTTAAAGAATGATTATGAATTACATTTGTATAAAATA	5720		
Query 369	ATTTTTATATTTGAAATATTGACTTTTTATGGCACTAGTATTTTATGAAATATTATGTT	428		
Sbjct 5721	ATTTTTATATTTGAAATATTGACTTTTTATGGCACTAGTATTTTATGAAATATTATGTT	5780		
Query 429	AAAACCTGGGACAGGGGAGAACCTAGGGTGATATTAACAGGGGCCATGAATCACCTTTTG	488		
Sbjct 5781	AAAACCTGGGACAGGGGAGAACCTAGGGTGATATTAACAGGGGCCATGAATCACCTTTTG	5840		
Query 489	GTCTGGAGGGAAGCCTTGGGGCTGATGCAGTTGTTGCCACAGCTGTATGATTCCCAAGC	548		
Sbjct 5841	GTCTGGAGGGAAGCCTTGGGGCTGATGCAGTTGTTGCCACAGCTGTATGATTCCCA-GC	5899		
Query 549	CAGCA 553			
Sbjct 5900	CAGCA 5904			

Homo sapiens CF transmembrane conductance regulator (CFTR), mRNA

Sequence ID: [NM_000492.4](#) Length: 6070 Number of Matches: 1

Range 1: 5320 to 5862 [GenBank](#) [Graphics](#) [Next Match](#) [Previous Match](#)

Score	Expect	Identities	Gaps	Strand
983 bits(532)	0.0	538/543(99%)	0/543(0%)	Plus/Minus
Query 11	GCCCCAAGGCTTCCCTCCAGACCAAAGGTGATTCATGGCCCCCTGGTTAATATCACCTTA	70		
Sbjct 5862	GCCCCAAGGCTTCCCTCCAGACCAAAGGTGATTCATGGCCCCCTGGTTAATATCACCTTA	5803		
Query 71	GGTTCTCCCTGTCCAGTTTTTAACATAATATTTTCATARAATACTAGTGCCATAAAAAAG	130		
Sbjct 5802	GGTTCTCCCTGTCCAGTTTTTAACATAATATTTTCATAGAAAATACTAGTGCCATAAAAAAG	5743		
Query 131	TCAATATTTTCAAATATAAAAAATATTTTATACAAATGTAATTCATAATCATCTTTTTAAA	190		
Sbjct 5742	TCAATATTTTCAAATATAAAAAATATTTTATACAAATGTAATTCATAATCATCTTTTTAAA	5683		
Query 191	ATACAGCATTTGTATATATGTTGAAACATTATTAATAATAATATTCCTAGAGAAAAAA	250		
Sbjct 5682	ATACAGCATTTGTATATATGTTGAAACATTATTAATAATAATATTCCTAGAGAAAAAA	5623		
Query 251	TTTTGCTTCACAAAAATTATRAACAGAGCATATAAACTAATTCATGATTGGTGCTTCT	310		
Sbjct 5622	TTTTGCTTCACAAAAATTATRAACAGAGCATATAAACTAATTCATGATTGGTGCTTCT	5563		
Query 311	TCAGTGGGCTCTCTCATCTCTCTTAGTGTAGACAGCATGAAGTACATACATCTAGCCTGA	370		
Sbjct 5562	TCAGTGGGCTCTCTCATCTCTCTTAGTGTAGACAGCATGAAGTACATACATCTAGCCTGA	5503		
Query 371	AAACATACCACCATCAACCTATACATCTAAATGCTTGGACTTCATGTGTGTCTACCCACA	430		
Sbjct 5502	AAACATACCACCATCAACCTATACATCTAAATGCTTGGACTTCATGTGTGTCTACCCACA	5443		
Query 431	GTGCCCATGGCCTATCTACTTACACACCCCTCTACCTGGAGCTTCTGTGGAAGAAGGGCT	490		
Sbjct 5442	GTGCCCATGGCCTATCTACTTACACACCCCTCTACCTGGAGCTTCTGTGGAAGAAGGGCT	5383		
Query 491	GTCCTGTGACAATTTGCACTAACATACATTTTCCAGCTAAACTCTAGTTCAAAGKCCCTGTG	550		
Sbjct 5382	GTCTGTGACAATTTGCACTAACATACATTTTCCAGCTAAACTCTAGTTCAAAGGCTCTGTG	5323		
Query 551	AGC 553			
Sbjct 5322	AGC 5320			

Figure 159: Sanger analysis of c.*1043A>C in CF4495056

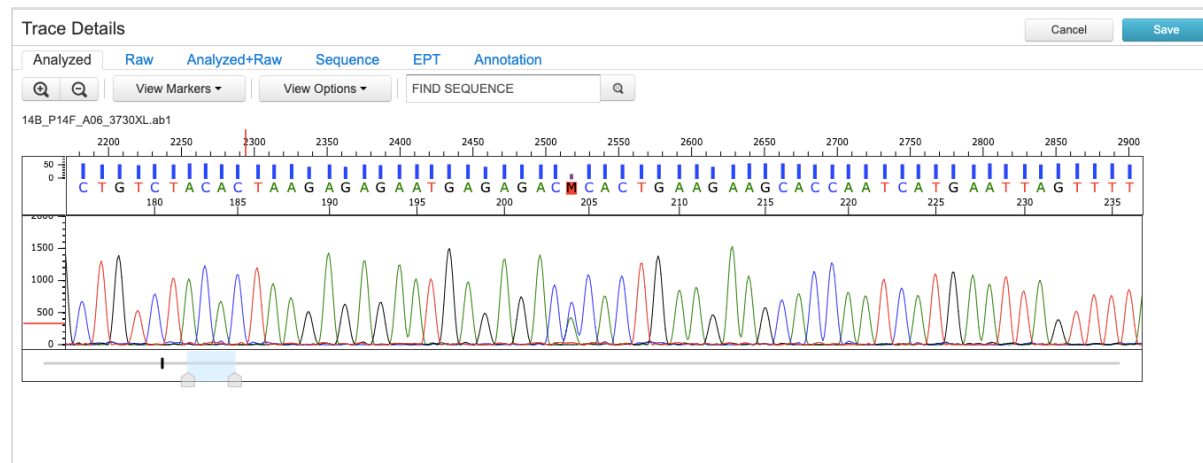


Figure 160: Sanger analysis of c.*1043A>C in CF6268769

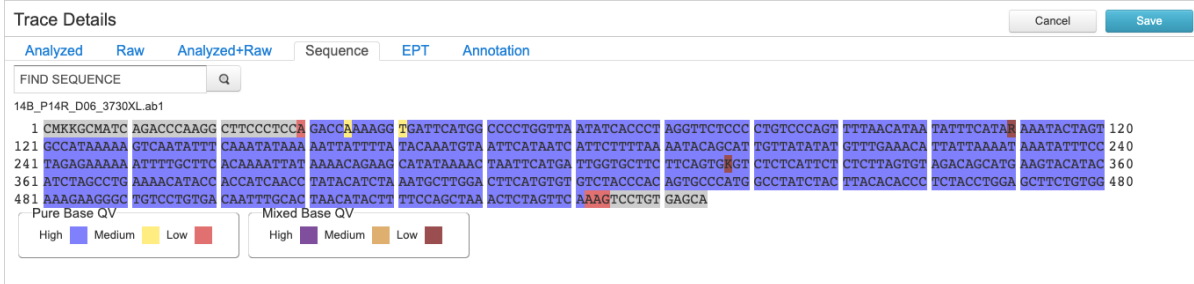
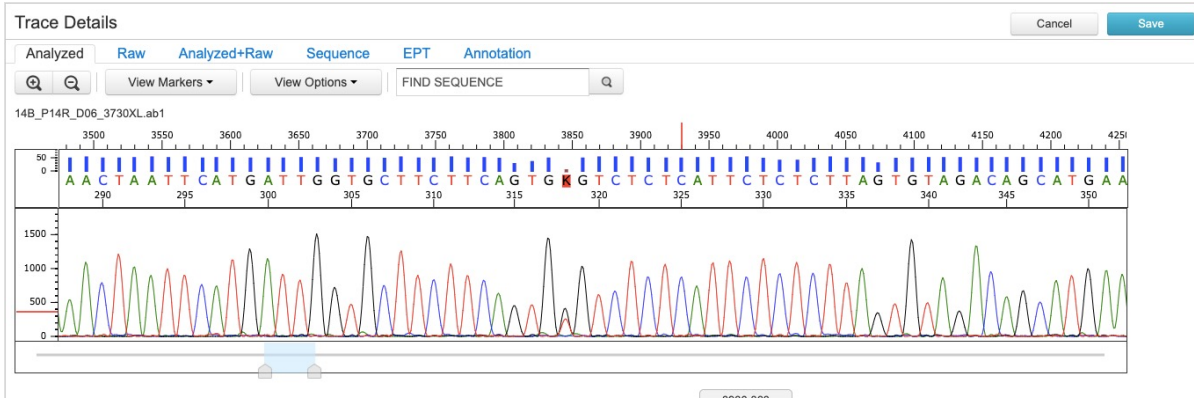


Figure 161: Sanger analysis of c.*1043A>C in CF6268769

Homo sapiens CF transmembrane conductance regulator (CFTR), mRNA

Sequence ID: [NM_000492.4](#) Length: 6070 Number of Matches: 1

Range 1: 5360 to 5902 [GenBank](#) [Graphics](#)

[Next Match](#) [Previous Match](#)

Score	Expect	Identities	Gaps	Strand
979 bits(530)	0.0	540/545(99%)	3/545(0%)	Plus/Plus
Query 8	GTTWAGTGC-AATTGTACAGGACAGCCCTTCTTTCCACAGAAGCTCCAGGTAGAGGGTG	66		
Sbjct 5360	GTT-AGTGCAAATTGTACAGGACAGCCCTTCTTTCCACAGAAGCTCCAGGTAGAGGGTG	5418		
Query 67	TGTAAGTAGATAGGCCATGGGCACTGTGGGTAGACACACATGAAGTCCAAGCATTAGAT	126		
Sbjct 5419	TGTAAGTAGATAGGCCATGGGCACTGTGGGTAGACACACATGAAGTCCAAGCATTAGAT	5478		
Query 127	GTATAGGTTGATGGTGGTATGTTTTTCAGGCTAGATGTATGTACTTCATGCTGTCTACACT	186		
Sbjct 5479	GTATAGGTTGATGGTGGTATGTTTTTCAGGCTAGATGTATGTACTTCATGCTGTCTACACT	5538		
Query 187	AAGAGAGAATGAGAGACACTGAAGAAGCACCAATCATGAATTAGTTTATATGCTTCT	246		
Sbjct 5539	AAGAGAGAATGAGAGACACTGAAGAAGCACCAATCATGAATTAGTTTATATGCTTCT	5598		
Query 247	GTTTTATAATTTTGTGAAGCAAAATTTTCTCTAGGAAATATTATTTTAAATATGTTT	306		
Sbjct 5599	GTTTTATAATTTTGTGAAGCAAAATTTTCTCTAGGAAATATTATTTTAAATATGTTT	5658		
Query 307	CAAACATATATAACAATGCTGTATTTTAAAAGAATGATTATGAATTACATTTGTATAAAA	366		
Sbjct 5659	CAAACATATATAACAATGCTGTATTTTAAAAGAATGATTATGAATTACATTTGTATAAAA	5718		
Query 367	TAATTTTATATTTGAAATATTGACTTTTTATGGCACTAGTATTTTATGAAATATTATG	426		
Sbjct 5719	TAATTTTATATTTGAAATATTGACTTTTTATGGCACTAGTATTTCTATGAAATATTATG	5778		
Query 427	TTAAACTGGGACAGGGGAGAACCTAGGGTGATATTAACGAGGGGCCATGAATCACCTTT	486		
Sbjct 5779	TTAAACTGGGACAGGGGAGAACCTAGGGTGATATTAACGAGGGGCCATGAATCACCTTT	5838		
Query 487	TGGTCTGGAGGGAAGCCTTGGGGCTGATGCAGTTGTGGCCACAGCTGTATGATTTCCAA	546		
Sbjct 5839	TGGTCTGGAGGGAAGCCTTGGGGCTGATGCAGTTGTGGCCACAGCTGTATGATTTCCAA-	5897		
Query 547	GCCAG 551			
Sbjct 5898	GCCAG 5902			

Homo sapiens CF transmembrane conductance regulator (CFTR), mRNA

Sequence ID: [NM_000492.4](#) Length: 6070 Number of Matches: 1

Range 1: 5320 to 5866 [GenBank](#) [Graphics](#)

[Next Match](#) [Previous Match](#)

Score	Expect	Identities	Gaps	Strand
987 bits(534)	0.0	542/547(99%)	0/547(0%)	Plus/Minus
Query 8	ATCAGACCCAAGGCTTCCCTCCAGACCAAAAAGGTGATTCATGGCCCCCTGGTTAATATCAC	67		
Sbjct 5866	ATCAGCCCCAAGGCTTCCCTCCAGACCAAAAAGGTGATTCATGGCCCCCTGGTTAATATCAC	5807		
Query 68	CCTAGGTTCTCCCTGTCCAGTTTAAACATAATATTTATARAATACTAGTGCCATAA	127		
Sbjct 5806	CCTAGGTTCTCCCTGTCCAGTTTAAACATAATATTTATAGAATACTAGTGCCATAA	5747		
Query 128	AAAGTCAATATTTCAAATATAAAAATTATTTTATACAATGTAATTCATAATCATTCTTT	187		
Sbjct 5746	AAAGTCAATATTTCAAATATAAAAATTATTTTATACAATGTAATTCATAATCATTCTTT	5687		
Query 188	TAAAAACAGCATTGTTATATATGTTTGAACATTTATAAAAATAAATTTTCCAGAGAA	247		
Sbjct 5686	TAAAAACAGCATTGTTATATATGTTTGAACATTTATAAAAATAAATTTTCCAGAGAA	5627		
Query 248	AAAAATTTGCTTCACAAAATTATAAAACAGAACATATAAACTAATTCATGATTGGTGC	307		
Sbjct 5626	AAAAATTTGCTTCACAAAATTATAAAACAGAACATATAAACTAATTCATGATTGGTGC	5567		
Query 308	TTCTTCAGTGTGCTCTCTCATTCTCTCTAGTGATAGACAGCATGAAGTACATACATCTAGC	367		
Sbjct 5566	TTCTTCAGTGTGCTCTCTCATTCTCTCTAGTGATAGACAGCATGAAGTACATACATCTAGC	5507		
Query 368	CTGAAAACATACCACCATCAACCTATACATCTAAATGCTTGGACTTCATGTGTCTACC	427		
Sbjct 5506	CTGAAAACATACCACCATCAACCTATACATCTAAATGCTTGGACTTCATGTGTCTACC	5447		
Query 428	CACAGTGCCCATGGCCCTATCTACTTACACACCCCTCTACCTGGAGCTTCTGTGGAAGAAG	487		
Sbjct 5446	CACAGTGCCCATGGCCCTATCTACTTACACACCCCTCTACCTGGAGCTTCTGTGGAAGAAG	5387		
Query 488	GGCTGTCTGTGACAATTTGCACTAACATACTTTTCCAGCTAAACTCTAGTTCAAAGTCC	547		
Sbjct 5386	GGCTGTCTGTGACAATTTGCACTAACATACTTTTCCAGCTAAACTCTAGTTCAAAGGTC	5327		
Query 548	TGTGAGC 554			
Sbjct 5326	TGTGAGC 5320			

Figure 162: Sanger analysis of c.*1043A>C in CF6268769

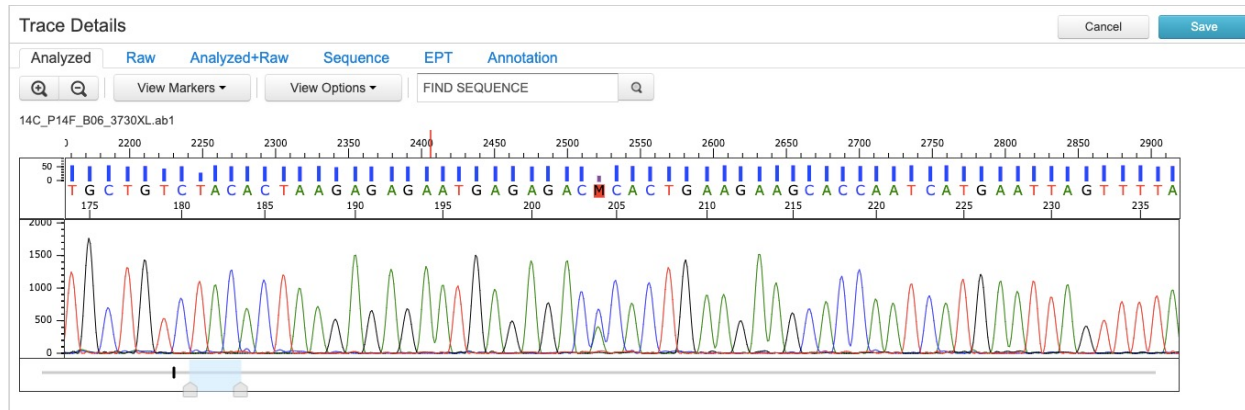


Figure 163: Sanger analysis of c.*1043A>C in CF2954129



Figure 164: Sanger analysis of c.*1043A>C in CF2954129

Homo sapiens CF transmembrane conductance regulator (CFTR), mRNA

Sequence ID: [NM_000492.4](#) Length: 6070 Number of Matches: 1

Range 1: 5320 to 5863 [GenBank](#) [Graphics](#)

[Next Match](#) [Previous Match](#)

Score	Expect	Identities	Gaps	Strand
981 bits(531)	0.0	539/544(99%)	1/544(0%)	Plus/Minus
Query 11	AGCCCC-AGGCTTCCCTCCAGACCAAAAGGTGATTTCATGGCCCTGGTTAATATCACCCCT	69		
Sbjct 5863	AGCCCCAAGGCTTCCCTCCAGACCAAAAGGTGATTTCATGGCCCTGGTTAATATCACCCCT	5804		
Query 70	AGGTTCTCCCTGTCCAGTTTTAACATAATATTTTCATARAATACTAGTGCCATAAAAA	129		
Sbjct 5803	AGGTTCTCCCTGTCCAGTTTTAACATAATATTTTCATAGAAATACTAGTGCCATAAAAA	5744		
Query 130	GTCAATATTTCAAAATATAAAATTTATTTTATACAAATGTAATTCATAATCATTTCTTTAA	189		
Sbjct 5743	GTCAATATTTCAAAATATAAAATTTATTTTATACAAATGTAATTCATAATCATTTCTTTAA	5684		
Query 190	AATACAGCATTGTTATATATGTTTGAACATTATTAAAAATAAATATTTCCTAGAGAAAAA	249		
Sbjct 5683	AATACAGCATTGTTATATATGTTTGAACATTATTAAAAATAAATATTTCCTAGAGAAAAA	5624		
Query 250	ATTTTGCTTCACAAAATTATAAAACAGAAGCATATAAAACAAATTCATGATTGGTGCTTC	309		
Sbjct 5623	ATTTTGCTTCACAAAATTATAAAACAGAAGCATATAAAACAAATTCATGATTGGTGCTTC	5564		
Query 310	TTCAAGTGGTCTCTCTATTCTCTCTTAGTGAGACAGCATGAAGTACATACATCTAGCCTG	369		
Sbjct 5563	TTCAAGTGGTCTCTCTATTCTCTCTTAGTGAGACAGCATGAAGTACATACATCTAGCCTG	5504		
Query 370	AAAAATACACCATCAACCTATACATCTAAATGCTTGGACTTCATGTGTCTACCCAC	429		
Sbjct 5503	AAAAATACACCATCAACCTATACATCTAAATGCTTGGACTTCATGTGTCTACCCAC	5444		
Query 430	AGTGCCCATGGCCTATCTACTTACACACCCCTCTACCTGGAGCTTCTGTGAAAGAAGGGC	489		
Sbjct 5443	AGTGCCCATGGCCTATCTACTTACACACCCCTCTACCTGGAGCTTCTGTGAAAGAAGGGC	5384		
Query 490	TGTCCTGTGACAAATTTGCACTAACATACTTTTCCAGCTAAACTCTAGTTCAAAGKCTGT	549		
Sbjct 5383	TGTCCTGTGACAAATTTGCACTAACATACTTTTCCAGCTAAACTCTAGTTCAAAGGCTGT	5324		
Query 550	GAGC 553			
Sbjct 5323	GAGC 5320			

Homo sapiens CF transmembrane conductance regulator (CFTR), mRNA

Sequence ID: [NM_000492.4](#) Length: 6070 Number of Matches: 1

Range 1: 5360 to 5904 [GenBank](#) [Graphics](#)

[Next Match](#) [Previous Match](#)

Score	Expect	Identities	Gaps	Strand
1000 bits(541)	0.0	543/545(99%)	0/545(0%)	Plus/Plus
Query 8	GTTAGTGCAAAATTGTCACAGGACAGCCCTTCTTCCACAGAAGCTCCAGGTAGAGGGTGT	67		
Sbjct 5360	GTTAGTGCAAAATTGTCACAGGACAGCCCTTCTTCCACAGAAGCTCCAGGTAGAGGGTGT	5419		
Query 68	GTAAGTAGATAGGCCATGGGCACTGTGGGTAGACACACATGAAGTCCAAGCATTTAGATG	127		
Sbjct 5420	GTAAGTAGATAGGCCATGGGCACTGTGGGTAGACACACATGAAGTCCAAGCATTTAGATG	5479		
Query 128	TATAGGTTGATGGTGGTATGTTTTCAGGCTAGATGTATGTACTTCATGCTGTCTACACTA	187		
Sbjct 5480	TATAGGTTGATGGTGGTATGTTTTCAGGCTAGATGTATGTACTTCATGCTGTCTACACTA	5539		
Query 188	AGAGAGAATGAGAGACACTGAAGAAGCACCAATCATGAATTAGTTTATATGCTTCTGT	247		
Sbjct 5540	AGAGAGAATGAGAGACACTGAAGAAGCACCAATCATGAATTAGTTTATATGCTTCTGT	5599		
Query 248	TTTTATAATTTTGTGAAGCAAAATTTTTCTCTAGGAAATATTTATTTTAATAATGTTTC	307		
Sbjct 5600	TTTTATAATTTTGTGAAGCAAAATTTTTCTCTAGGAAATATTTATTTTAATAATGTTTC	5659		
Query 308	AAACATATATAACAATGCTGTATTTTAAAGAATGATTATGAATTACATTTGTATAAAAA	367		
Sbjct 5660	AAACATATATAACAATGCTGTATTTTAAAGAATGATTATGAATTACATTTGTATAAAAA	5719		
Query 368	AATTTTATATTTGAAATATTGACTTTTTATGGCACTAGTATTTTATGAAATATTATGT	427		
Sbjct 5720	AATTTTATATTTGAAATATTGACTTTTTATGGCACTAGTATTTTATGAAATATTATGT	5779		
Query 428	TAAACATGGGACAGGGGAGAACCTAGGGTGATATTAAACAGGGGCCATGAATCACCTTTT	487		
Sbjct 5780	TAAACATGGGACAGGGGAGAACCTAGGGTGATATTAAACAGGGGCCATGAATCACCTTTT	5839		
Query 488	GGTCTGGAGGGAAGCCTTGGGGCTGATGCAGTTGTTGCCACAGCTGTATGATTTCCAGC	547		
Sbjct 5840	GGTCTGGAGGGAAGCCTTGGGGCTGATGCAGTTGTTGCCACAGCTGTATGATTTCCAGC	5899		
Query 548	CAGCA 552			
Sbjct 5900	CAGCA 5904			

Figure 165: Sanger analysis of c.*1043A>C in CF2954129

SnackVar Report

ID : CF1133987; p.Ser549Asn

Description : Confirmation of p.Ser549Asn in CF1133987

Reference sequence : NM_000492.4(CFTR).fasta

Variant 1

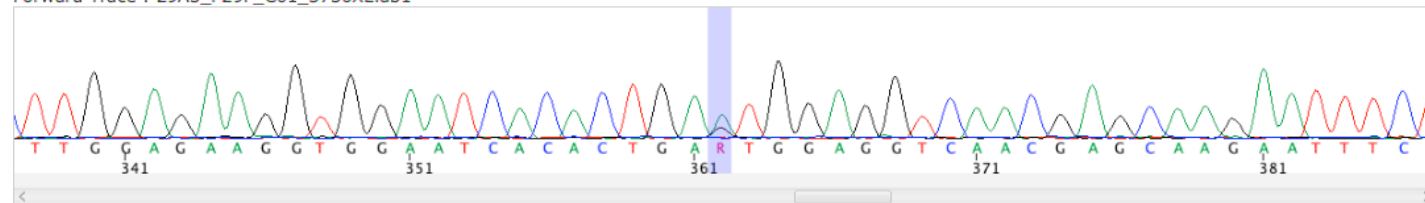
Description : c.1646G>A, p.(Ser549Asn), hetero

Alignment

Alignment view showing the variant c.1646G>A, p.(Ser549Asn) in the CFTR reference sequence (NM_000492.4). The alignment shows the reference sequence (top) and the variant sequence (bottom). The variant is highlighted in yellow. The alignment is shown for the region 311 to 421.

Position	Reference	Variant
311	ACATCTCCAAGTTTGCAGAGAAAGACAATATAGTTCTTGGAGAAGGTGGAATCACACTGAGTGGAGGTCAACGAGCAAGAATTTCTTTAGCAAGgtgaataactaatatttggctctagcaag	ACATCTCCAAGTTTGCAGAGAAAGACAATATAGTTCTTGGAGAAGGTGGAATCACACTGAGTGGAGGTCAACGAGCAAGAATTTCTTTAGCAAGGTGAATAACTAATTATTGGTCTAGCAAG
321		
331		
341		
351		
361		
371		
381		
391		
401		
411		
421		

Forward Trace : 29A3_P29F_C01_3730XL.ab1



Reverse Trace : 29A3_P29R_D01_3730XL.ab1

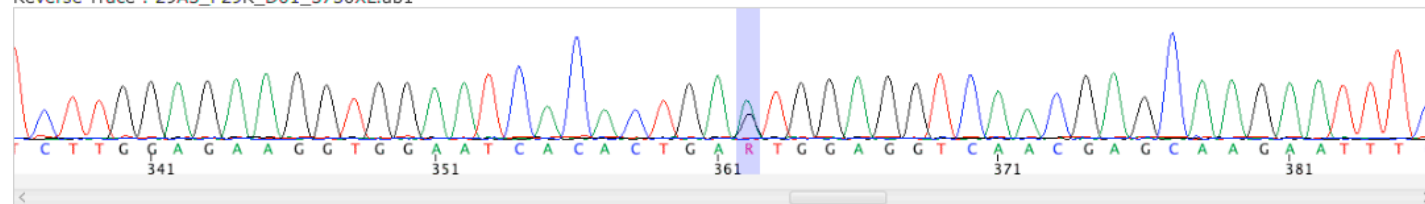


Figure 166: SnackVar report for the p.Ser549Asn variant in CF1133987