

Additional file 1: 21 PEM patterns

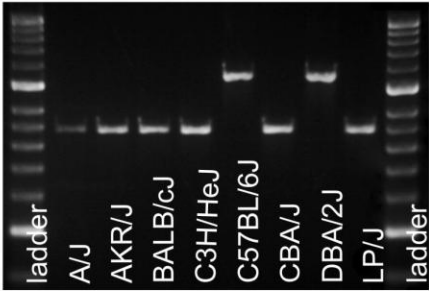
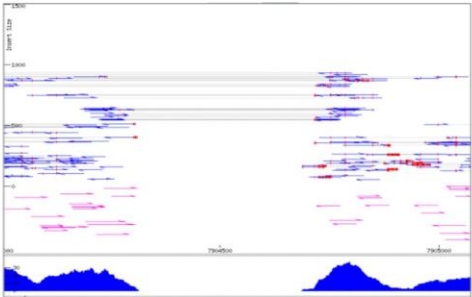
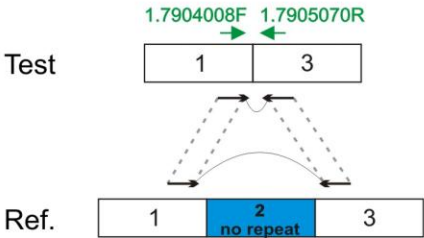
High Confidence PEM patterns

Example in Look Seq

PCR validation

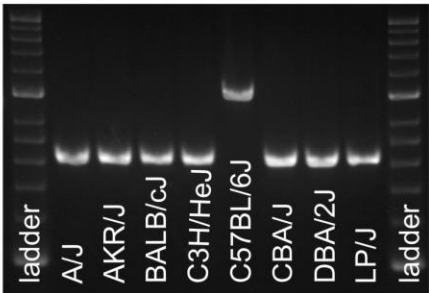
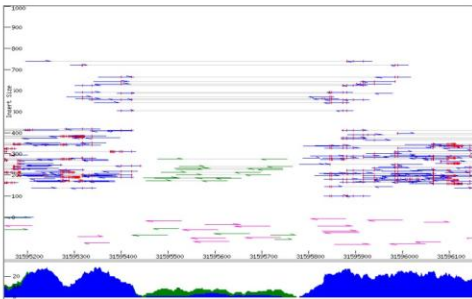
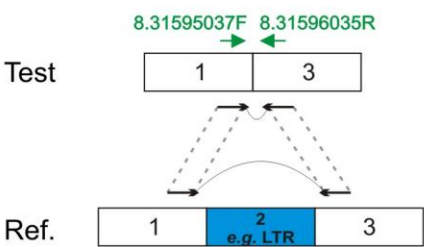
H1

Deletion of 390 bp [111110101]
chr1:7,904,302-7,904,691 bp



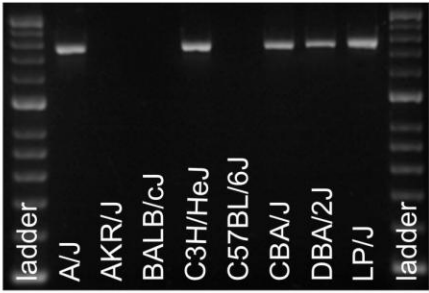
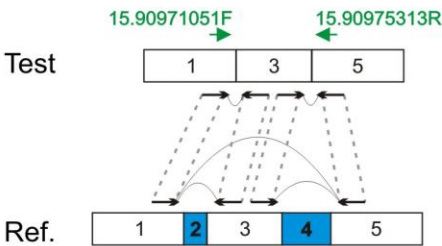
H2

Deletion of 656 bp [111110111]
chr8:31,595,128-31,595,783 bp



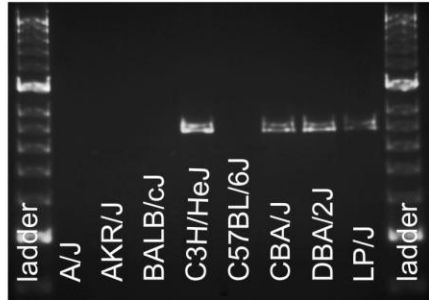
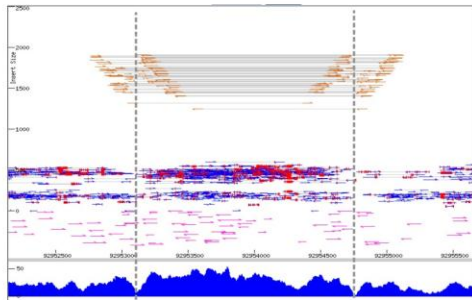
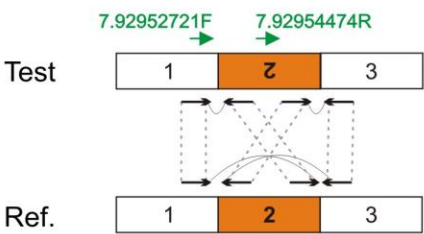
H3

del-384bp_nml-273_del-2414 [100101111]
1st del - chr15:90,971,314-90,971,697 bp
2nd del - chr15:90,971,971-90,974,384 bp



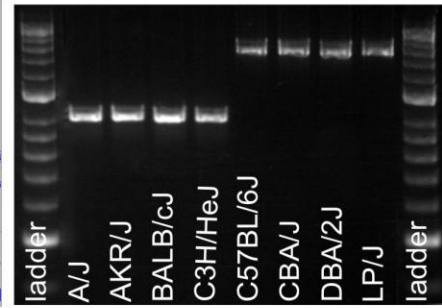
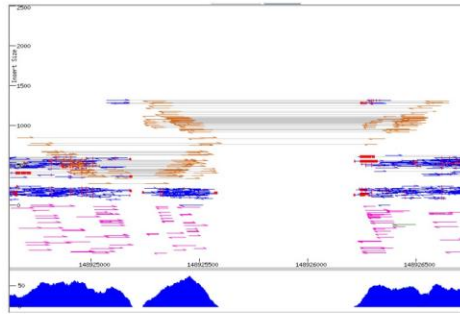
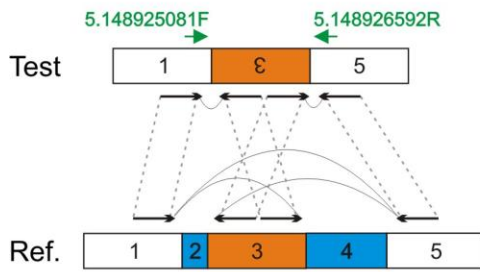
H4

Inversion of 1643 bp [000101111]
chr7:92,953,110-92,954,752 bp



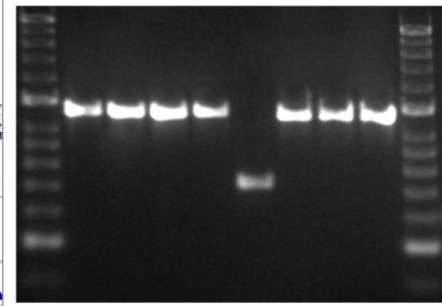
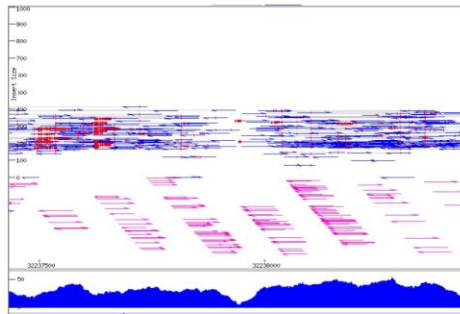
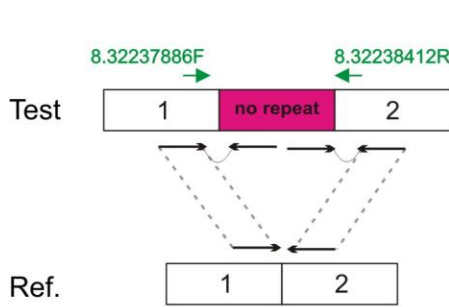
H5

del-71bp_inv-325_del-645 [11110000]
1st del - chr5:148,925,178-148,925,248 bp
inv - chr5:148,925,249-148,925,573 bp
2nd del - chr5:148,925,574-148,926,218 bp



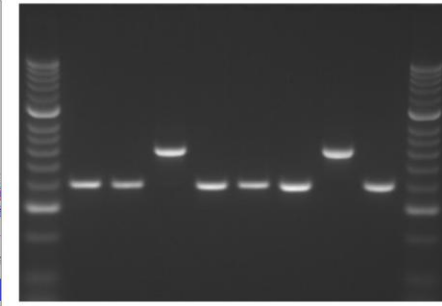
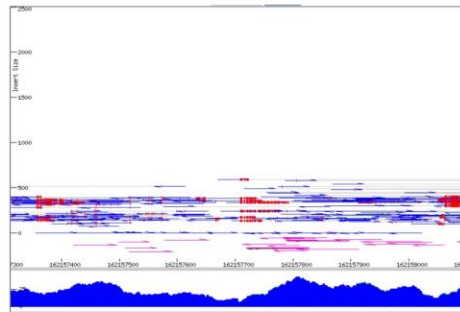
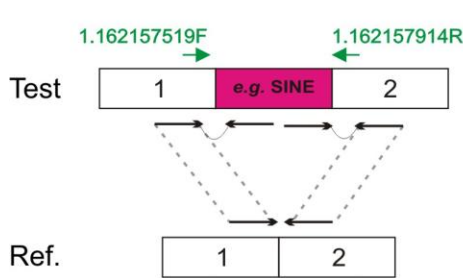
H6

Insertion of 391 bp [11110111]
chr8:32,237,946-32,237,948



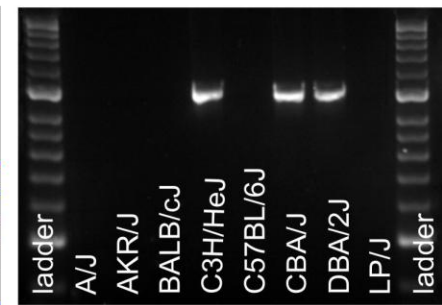
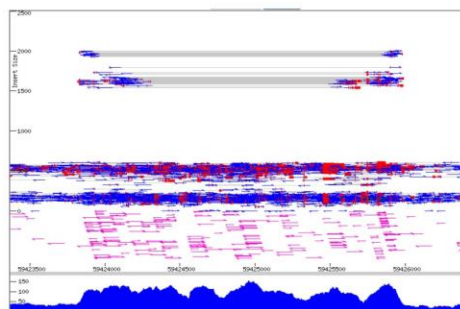
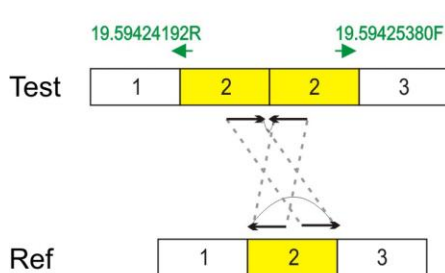
H7

Insertion of ~200 bp [00100010]
chr1:162,157,709-162,157,710



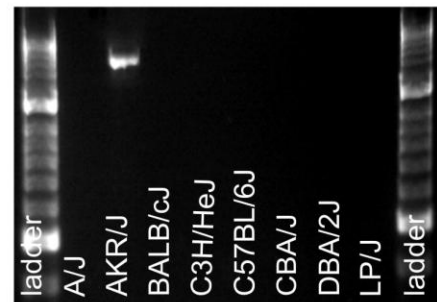
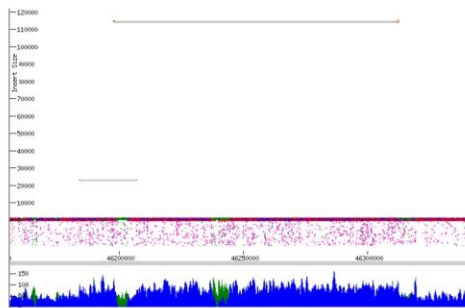
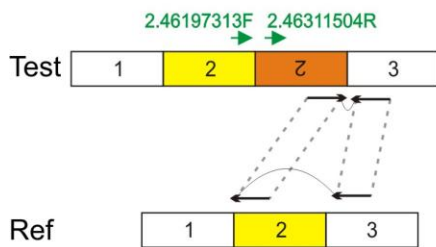
H8

Tandem duplication of 2181 bp [00010110]
chr19:59,423,833-59,425,976 bp



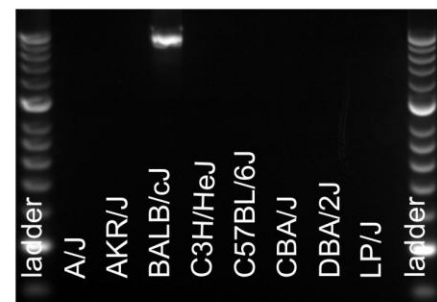
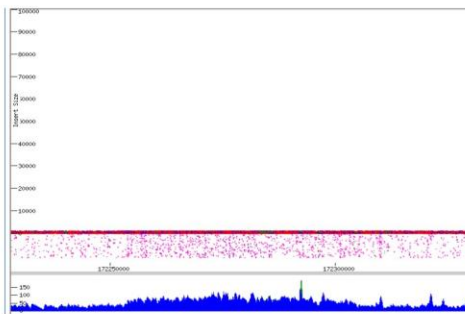
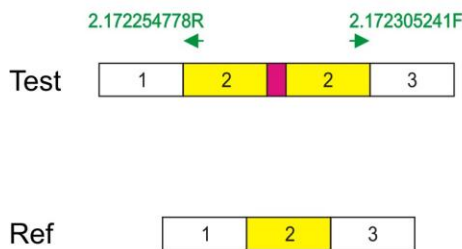
H9

Inverted tandem gain of ~128,6731 bp [01000000]
chr2:46,183,800-46,312,472 bp



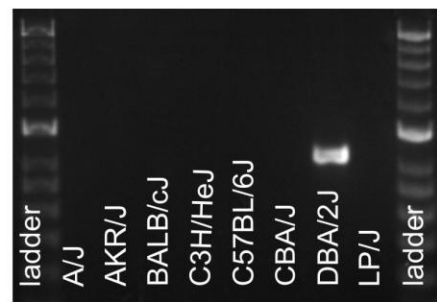
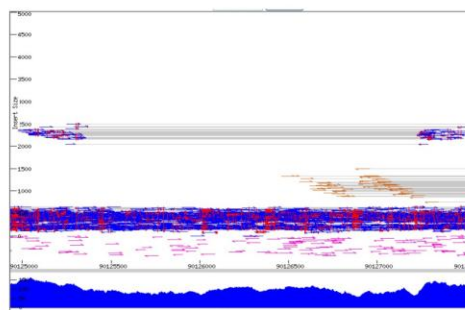
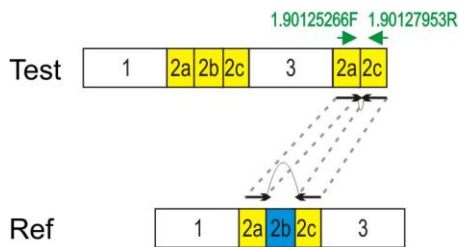
H10

Tandem gain of ~51681 bp with ins ~700bp [00100000]
chr2:172,253,836-172,305,516 bp



H11

Deletion in gain of ~1854 bp [00000010]
chr1:90,125,368-90,127,221 bp



Questionable PEM patterns Example in LookSeq

PCR validation

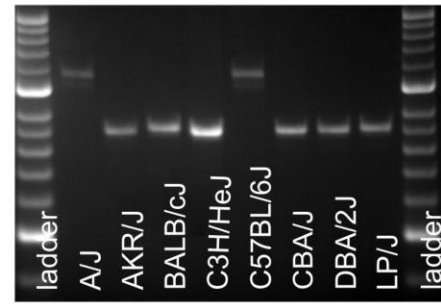
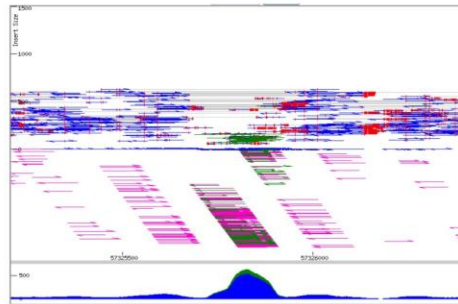
Q1

Deletion of 185 bp [01110111]
chr19:57,325,727-57,325,911 bp

Test



Ref.



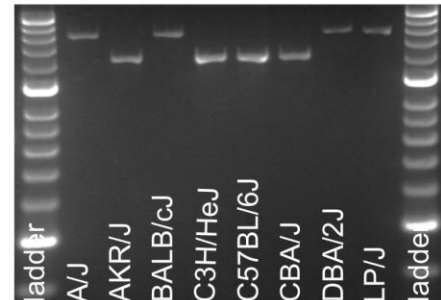
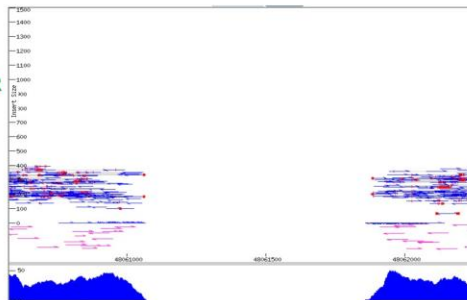
Q2

Deletion of ~ 836 bp with ins ~ 1200 bp [10100011]
chr19:48,061,057-48,061,892 bp

Test



Ref.



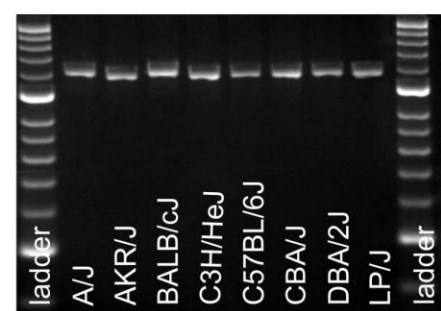
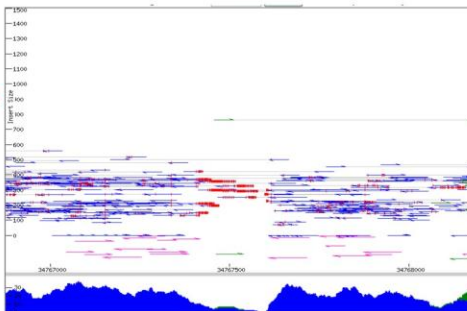
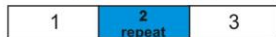
Q3

Deletion of ~ 163 bp with ins ~ 250 bp [10100000]
chr19:34,767,448-34,767,610 bp

Test



Ref.



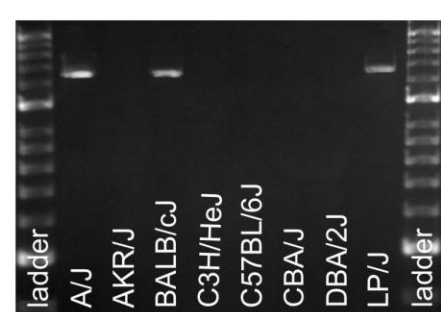
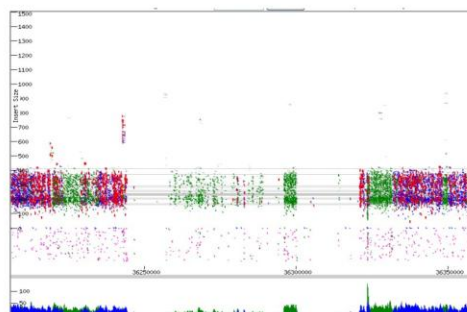
Q4

Deletion of 76444 bp [10100001]
chr16:36,244,199-36,320,642 bp

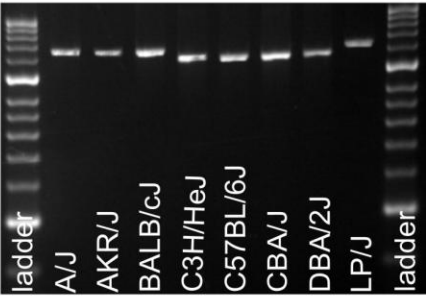
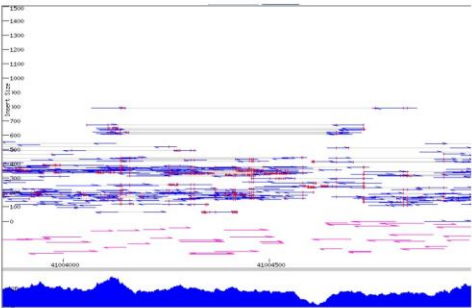
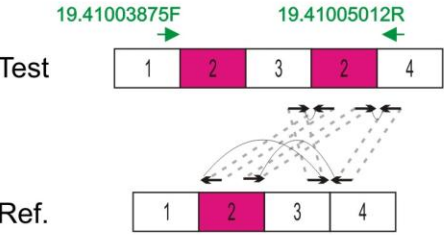
Test



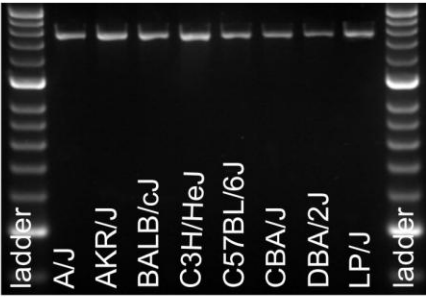
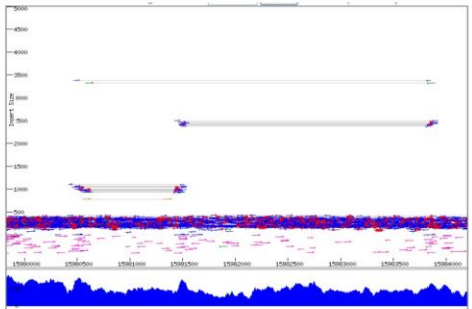
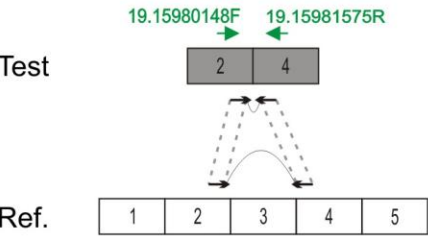
Ref.



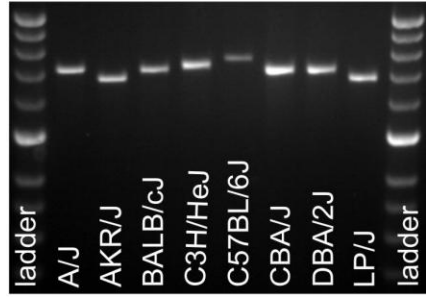
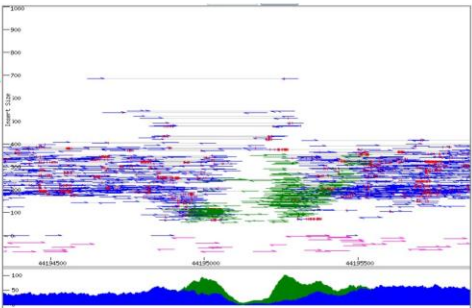
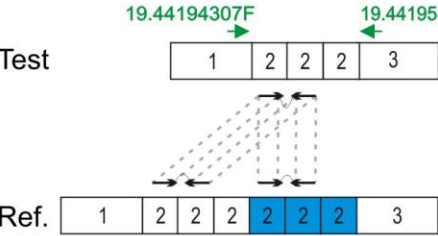
Q5
False deletion, instead insertion of 97 bp [11100001]
chr19:41,004,153-41,004,751 bp



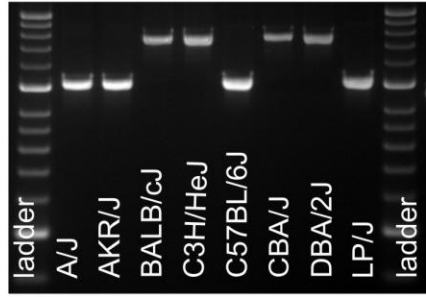
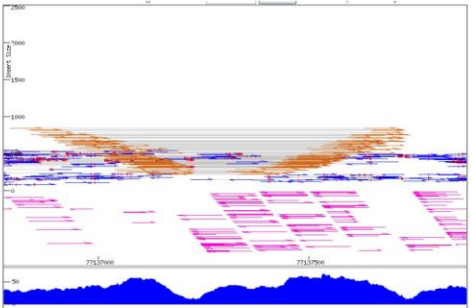
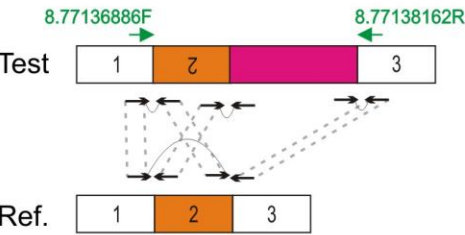
Q6
False deletion
chr19:15,980,612-15,981,451 bp



Q7
Deletion of variable size
chr19:44,194,931-44,195,258 bp

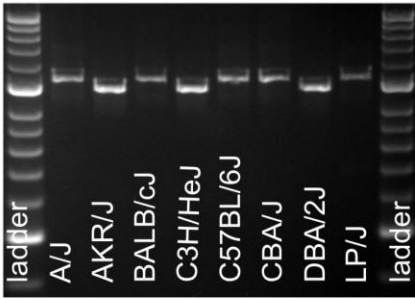
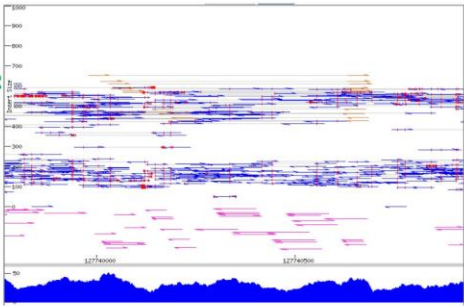
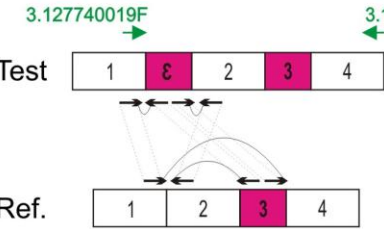


Q8
Inversion of ~ 548 bp with ins of ~400 bp [00110110]
chr8:77,137,213-77,137,760 bp



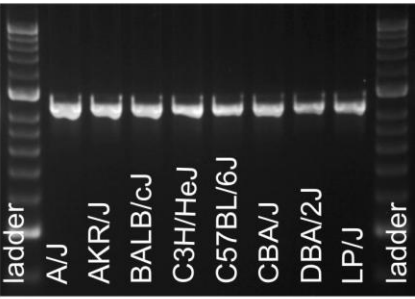
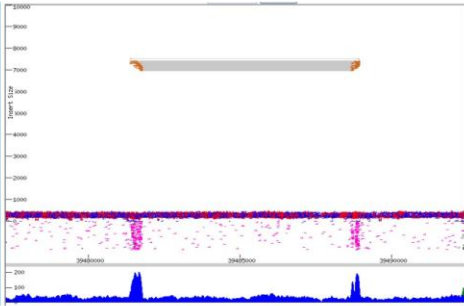
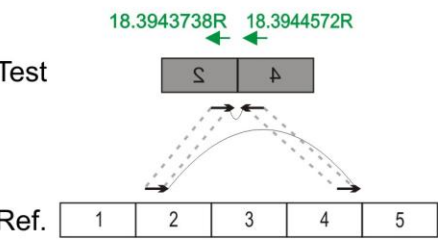
Q9

Inverted linked insertion of 68 bp [10110101]
chr3:127,740,121-127,740,698 bp



Q10

False inversion of about 7598 bp [11110111]
chr18:39,481,367-39,488,964 bp



Pattern relative to the reference: ■ Del ■ Ins ■ Inv ■ Dup ■ pseudogene