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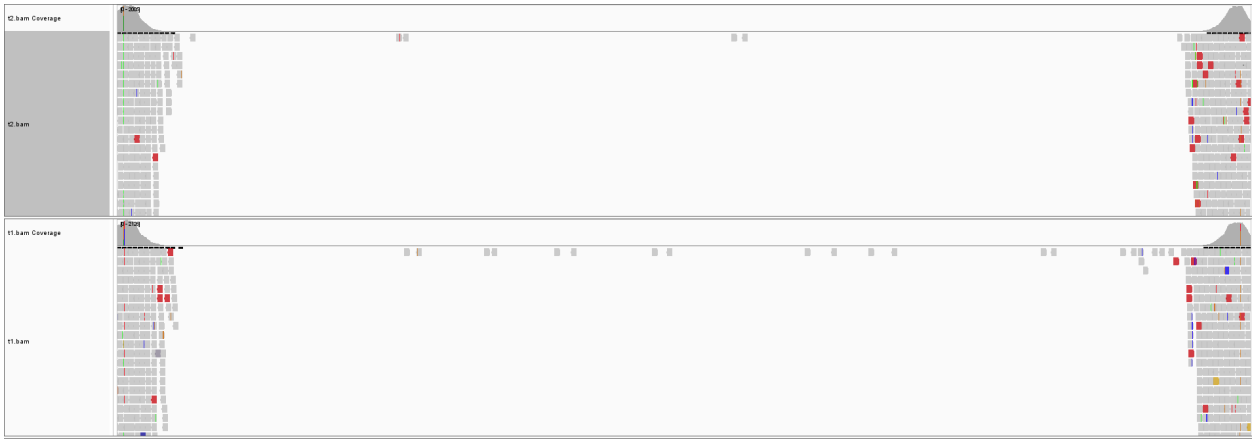
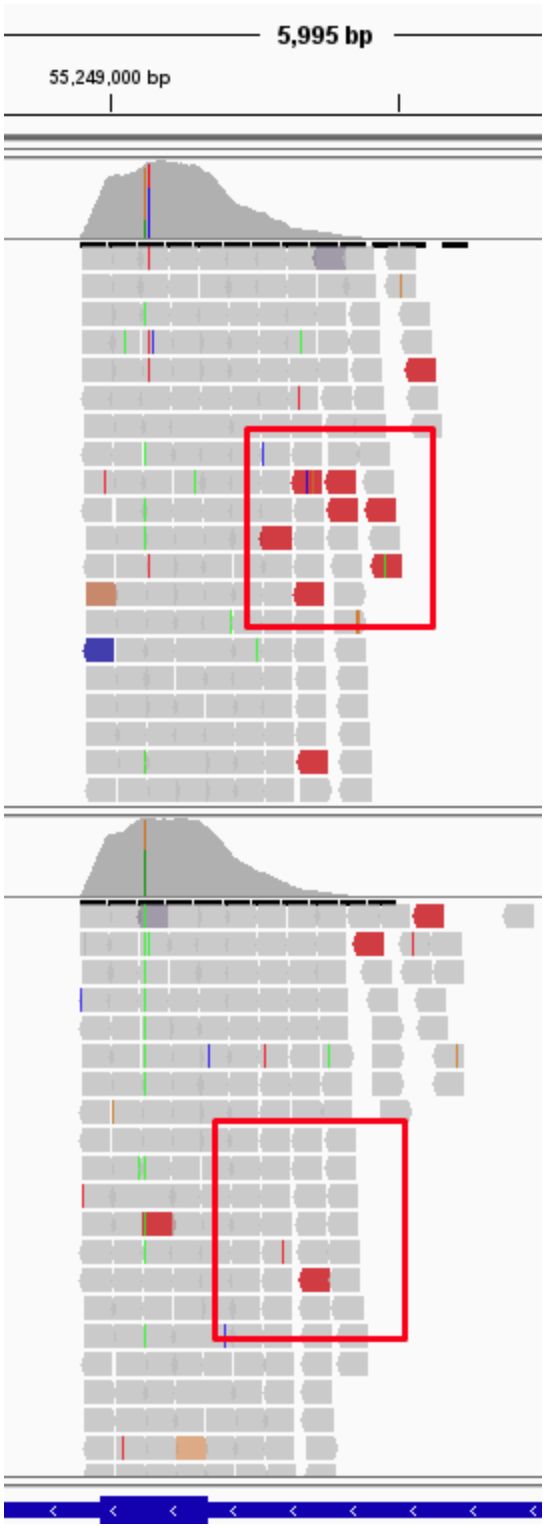


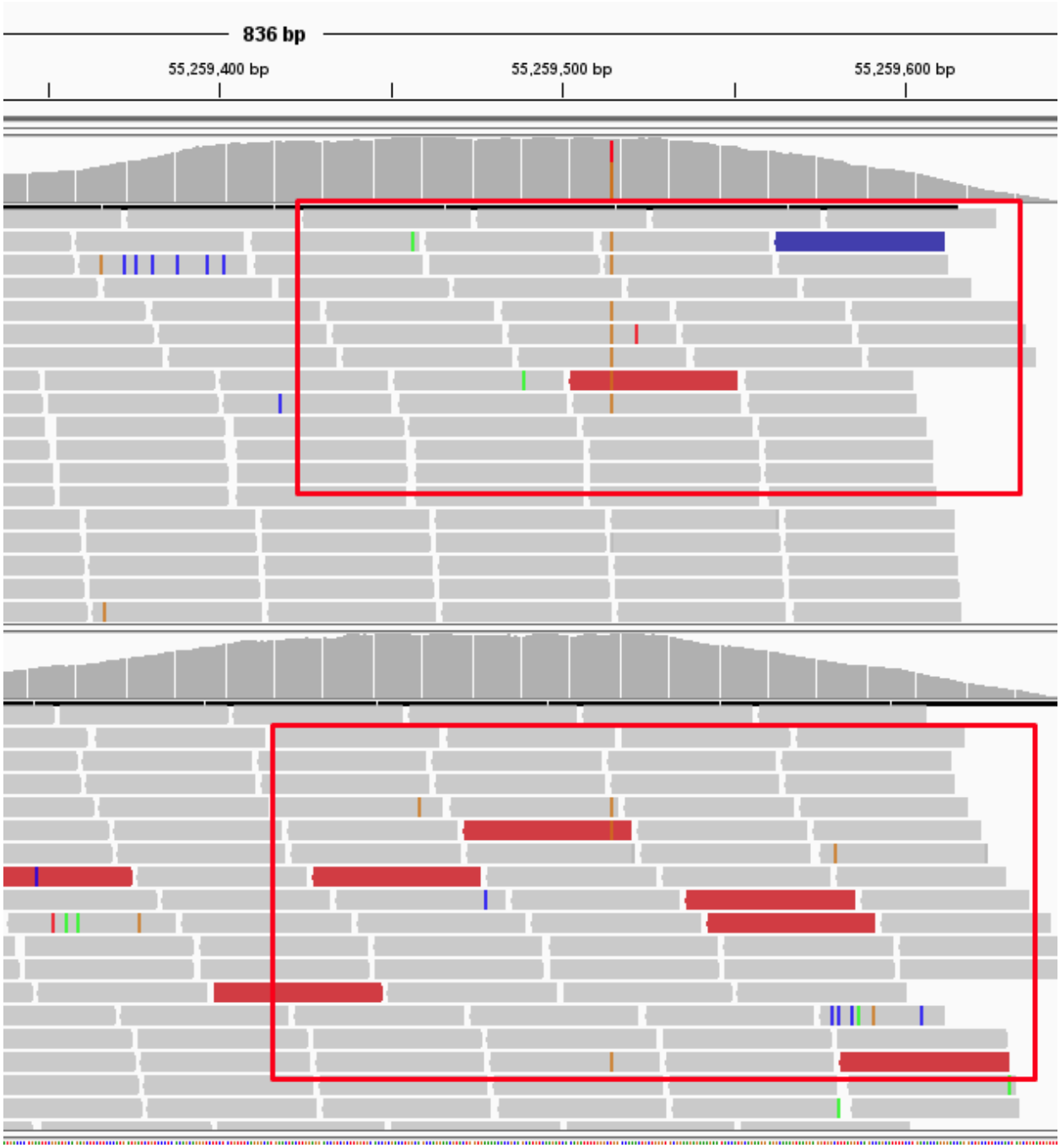
Shown above here are the two bam files

1. As we can see there are many different red sections and upon closer inspection there are also yellow and blue sections as well. Looking into the user guides for IGV online, I found that red indicates sections of deletion while blue indicates sections of insertion. What is interesting to me is that from the t1.bam file to the t2.bam file there is a decreased number of deletions and they have also moved locations. A section is highlighted below where there is a change in the number of deletions from t1 to t2 in the 55,249,100 bp section of the alignment



additionally there are also a different area near the 55,259,100 section as shown below

And lastly this specific section at 55,259,500



I located these three sections with the biggest amount of changes from t1 to t2 and I believe that these various changes on chromosome 7 would affect the synthesis of 97-amino-acid preproNPY

2. The main differences that I found between the time points was that there are changes to the number of deletions. According to IGV, red sections of the alignments refer to insert sizes that are larger than average and thus have evidence of possible deletions. From comparing the two files t1 and t2 I can definitely see that they have a different amount of

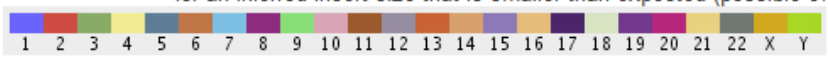
red inserts and in different locations. I mainly interpret this result as the patient's tumor changing between the two different time points. The main reason for the difference in these two mutation profiles is that the cancer cells may have mutated and changed the number of deletions into ones that cause the tumor to grow uncontrollably.

3. By taking into account that the median error rate for sequences on IGV is around 5% I would definitely say that these two tumor deep sequences have an above average error rate. Besides the red inserts, there were also blue and other colored inserts which suggests that certain portions have been rearranged. As shown below, IGV labels inserts different colors when there is an inter-chromosomal rearrangement, and I don't believe all these rearrangements are due to the tumor itself, but some can be a cause of errors during deep sequencing

The inferred insert size can be used to detect structural variants, such as:

- deletions
- insertions
- inter-chromosomal rearrangements

IGV uses color coding to flag anomalous insert sizes. When you select *Color alignments>by insert size* in the popup menu, the default coloring scheme is:

-  for an inferred insert size that is larger than expected (possible evidence of a deletion)
-  for an inferred insert size that is smaller than expected (possible evidence of an insertion)
- 

for paired end reads that are coded by the chromosome on which their mates can be found