Khan Inan
Professor Truong
BI-GY 7683

Tumor 1: first clinical visit. Sequence 1:
AGATTTTGGGCGGGCCAAACTGCTGGGTGCGGAAGAGAAAGAATACCAT

This first clinical visit and sequence shows us a sequence that is a mutation of chr7. The mutation is shown below where there is a single substitution mutation and the T base has changed to a G base. A single point mutation like this has most likely resulted in the tumor in question because chromosome 7 is responsible for controlling the growth and division of cells.

```
00000001 agattttgggcgggccaaactgctgggtgcggaagagaaagaataccat 00000049
>>>>>>>> |||||||||||| |||||||||||||||||||||||||||||||||||| >>>>>>>>
55191811 agattttgggctggccaaactgctgggtgcggaagagaaagaataccat 55191859
```

Tumor 1: first clinical visit: Sequence 2:
CACCTCCACCGTGCAGCTCATCACGCAGCTCATGCCCTTCGGCTGCCTC

This first clinical visit for sequence 2 seems to point towards what seems like a better case. Although this sequence was taken from a tumor sample, the sequence in question seems to be unaffected and “healthy.” Sequence is also from chromosome 7 and is a perfect match to a section of chr7

```
00000001 cacctccaccgtgcagctcatcacgcagctcatgcccttcggctgcctc 00000049
>>>>>>>> ||||||||||||||||||||||||||||||||||||||||||||||||| >>>>>>>>
55181355 cacctccaccgtgcagctcatcacgcagctcatgcccttcggctgcctc 55181403
```

QUERY	SCORE	START	END	QSIZE	IDENTITY	CHROM	STRAND	START	END	SPAN
YourSeq	49	1	49	49	100.0%	chr7	+	55181355	55181403	49

Tumor 1: later clinical visit: Sequence 1:
AGATTTTGGGCTGGCCAAACTGCTGGGTGCGGAAGAGAAAGAATACCAT

This later visit for sequence 1 shows that the previous substitution mutation no longer exists. Where previously the T base was changed to a G, it is now back to a T. This suggests that the mutation has either reverted, or this specific section of chromosome 7 is no longer affected.

```
00000001 agattttgggctggccaaactgctgggtgcggaagagaaagaataccat 00000049
>>>>>>>> ||||||||||||||||||||||||||||||||||||||||||||||||| >>>>>>>>
55191811 agattttgggctggccaaactgctgggtgcggaagagaaagaataccat 55191859
```

QUERY	SCORE	START	END	QSIZE	IDENTITY	CHROM	STRAND	START	END	SPAN
YourSeq	49	1	49	49	100.0%	chr7	+	55191811	55191859	49

Tumor 1: later clinical visit: Sequence 2:
CACCTCCACCGTGCAGCTCATCATGCAGCTCATGCCCTTCGGCTGCCTC

This sequence is the opposite case of sequence 1, because it previously seemed healthy, but now it has a mutation. The mutation in question is a substitution where a C base has changed to a T base. This is still chromosome 7

```
00000001 cacctccaccgtgcagctcatcatgcagctcatgcccttcggctgcctc 00000049
>>>>>>>> ||||||||||||||||||||||||| ||||||||||||||||||||||||| >>>>>>>>
55181355 cacctccaccgtgcagctcatcacgcagctcatgcccttcggctgcctc 55181403
```

Tumor 2:
GTCAAGGCACTCTTGCCTACGCCACTAGCTCCAACTACCACAAGTTTAT

This sequence is a bit different compared to the previous sequences in that the mutation now affects chromosome 12. It is still a substitution mutation and the C base in chromosome 12 has been replaced by a T base. Chromosome 12 is responsible for providing instructions for protein synthesis so it makes sense why a mutation in this gene would cause a tumor.

```
00000001 gtcaaggcactcttgcctacgccactagctccaactaccacaagtttat 00000049
>>>>>>>> ||||||||||||||||||||||||| ||||||||||||||||||||||||| >>>>>>>>
25245326 gtcaaggcactcttgcctacgccaccagctccaactaccacaagtttat 25245374
```

Tumor 3:
CGACAGTCCCAGGAGGCTTCGGGGGGGTCCTGACACTGCACCCTGGCC

This last tumor is a deletion mutation and it affects chromosome 11. The deletion in this case is one where a C base has been removed. Chromosome 11 is believed to also be responsible for providing instructions for making proteins.

```
00000001 cgacagtcccaggaggcttc.gggggggtcctgacactgcaccctggcc 00000048
>>>>>>>> |||||||||||||||||||| |||||||||||||||||||||||||||| >>>>>>>>
64804600 cgacagtcccaggaggcttccgggggggtcctgacactgcaccctggcc 64804648
```

Bonus interpretation question:

What I believe has occurred in the case of tumor 1 is that the specific point of the sequence where the cancerous mutation has occurred has changed location. What led me to believe this is that from the first visit of sequence 1 to the later visit it seems that the mutation has reverted or “healed.” If someone were to just look at sequence 1 for this tumor across the two visits they

would think that the mutation that created the tumor no longer exists. However if you look at sequence 2, you would see that a sequence that started off normal, now has the substitution mutation in the later visit. This suggests to me that the mutation that caused the tumor has changed locations along the sequence.