

Long Answer Practical questions

15. What do N50 and L50 mean? Write a code in any language of your choice to calculate for a given contigs file Q8_contigs.fa. (**DO NOT** use any inbuilt library function like biopython etc). (10)

OR

Using the reads given below build a single contig based on appropriate overlaps between the reads (10)

```
TGGGATTGGGGTTTT
      GGTTTTCCCCTCCCA
            CTCCCATGTGCTCAA
                  GCTCAAGACTGGCGC
```

16. From the UCSC browser;

- a. Within the hg19 genome, navigate to the following location
chr8:128,748,701-128,748,900 and fill out the tabulated details of the COSMIC mutation
COSM1161715 (5)

Gene	Nucleotide substitution	Type of mutation	Associated Disease	Length of Associated CpG island

- b. From the mm10 mouse genome, Report the following for the **Brca1** and **Brca2** gene (5)

Gene	Length of gene	Number of exons	Approx length of 10th exon	# of isoforms