STELLA MARIS COLLEGE (AUTONOMOUS) CHENNAI -600 086 (For candidates admitted from the academic year 2019 - 2020 & thereafter)

M. Sc. DEGREE EXAMINATION, APRIL 2024 **BIOINFORMATICS** FOURTH SEMESTER

COURSE	:	CORE
PAPER	:	ADVANCES IN BIOINFORMATICS
SUBJECT CODE	:	19BI/PC/AB44
TIME	:	3 HOURS

SECTION – A

ANSWER ALL QUESTIONS

(20 x 1=20)

MAX. MARKS: 100

1. Which of these projects would be best suited for Next Generation Sequencing?

- a) To determine if a tumour sample contains a common missense mutation
- b) To find the transcriptome of a tumour sample
- c) To genotype ten genomic DNA samples for a known single nucleotide polymorphism
- d) All of the above.
- 2. Once the sequences are obtained from your Next Generation Sequencing experiment what is the first thing you should do?
 - a) Perform a bioinformatics analysis of your data
 - b) Check your data using a different method
 - c) Publish your results
 - d) Further investigate the sequences of interest.
- 3. Personalised medicine has the potential to yield plenty of health and economic benefits. Which of the following would not be a benefit of personalised medicine?
 - a) Increased number of medical jobs
 - b) Improved medical decision making
 - c) Delivery of most effective therapies d) Optimise disease prevention strategies
- 4. The most commonly occurring variant in the human genome is
 - a). tandem-repeat polymorphism. b) premature stop codon.
 - c). nucleotide base insertion. d) single-nucleotide polymorphism
- 5. Which of the following statements are not true Microarrays Are used for analysis of
 - ii. Contain RNA sequences i. transcriptomes
 - iii. Contain DNA sequences iv. Are smaller than DNA chips
 - a) i and ii correct b) ii and iii correc c) i and iii correct d) ii and iv correct
- 6. In terms of understanding the pathways between genes and behaviour, it is fairly safe to say that
 - a) we know more about the environment than the genes
 - b) we know more about the genes than the environment
 - c) the new field of molecular genetics is the best way to gain a full understanding of the gene/environment interactions in the pathways
 - d) OTL analysis has actually hindered the understanding of these pathways by suggesting so many separate gene contributions to basic behaviour processes
- 7. When you scan the microarray in the scanner, the data show some black spots. What do these represent?
 - a)The DNA that has been replicated in healthy cells.
 - b) The mRNA that was washed away in the washing solution.
 - c) The DNA that was not transcribed and expressed in healthy cells.
 - d) The mRNA that was not bound by Oligo-d-tails in the beads.

- 8. The two most common processes that lead to production of multiple functional proteins from the same DNA sequence are:
 - a) RNA editing and alternative splicing.
 - b) Protein folding and posttranslational covalent modifications.
 - c) Alternative splicing and posttranslational covalent modifications.
 - d) Posttranslational covalent modification and transcriptional regulation.
 - 9. In an analysis of eukaryotic gene, you identify several non-overlapping open reading frames, but they are not all in the same frame. Which explanation makes the most sense?
 - a) By random chance, a second reading frame within the gene also has an open reading frame.
 - b) This gene includes introns which are not multiples of three.
 - c) This is a mutant allele that has had several small insertions.
 - d) All of these.
 - 10. In which situation would you expect the mutation frequency to increase over time?
 - a) The deleterious effect of the mutation is balanced with the mutation rate.
 - b) A lethal allele is created at a high mutation rate.
 - c) A neutral allele is created at low mutation rate.
 - d) All of the above.
 - 11. Explain about the UCSC genome.
 - 12. Explain the term "Gene regulatory dynamics".
 - 13. Write importance of Crispr.
 - 14. Write a short notes on FastQC.
 - 15. Explain the applications of targeted mutagenesis.
 - 16. What is regulatory sequence motifs?
 - 17. Define NCBI refseq.
 - 18. Write a short notes on FASTQ file formats.
 - 19. Explain the methods adopted for quantifying RNA.
 - 20. How to create the expression table?

SECTION – B

ANSWER ANY FOUR QUESTIONS. EACH ANSWER SHOULD NOT EXCEED 500 WORDS. ALL QUESTIONS CARRY EQUAL MARKS. $(4 \times 10 = 40)$

- 21. Explain the principles of Next generation sequencing and write its importance in medicine.
- 22. Describe in details about the alpha and beta diversity of metagenomic studies.
- 23. List out the Linux commands and its functions.
- 24. Discuss about the repair and data analysis of the edited genome.
- 25. Explain the epigenetic modifications.
- 26. Summarize the file formats to analyze NGS data.
- 27. Write a case study to perform RNA seq analysis.

SECTION – C

ANSWER ANY TWO QUESTIONS. EACH ANSWER SHOULD NOT EXCEED 1200WORDS. ALL QUESTIONS CARRY EQUAL MARKS.(2 x 20 = 40)

- 28. Write an essay on analysis of metagenome data and logical steps for metagenome analysis.
- 29. Describe the RNA dynamics at the level of transcription and post-transcriptional processing.
- 30. Explain in details about the salient features of transcriptomics and write the logical steps for analysing RNA seq data.
- 31. Briefly explain Crispr-Cas9 mechanism and its applications.