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# B.M.S.College of Engineering, Bengaluru-560019

Autonomous Institute Affiliated to VTU

## January / February 2025 Semester End Main Examinations

**Programme: B.E.**

**Semester: VII**

**Branch: Biotechnology**

**Duration: 3 hrs.**

**Course Code: 22BT7PEGIN**

**Max Marks: 100**

**Course: Genome Informatics**

**Instructions:** 1. Answer any FIVE full questions, choosing one full question from each unit.  
2. Missing data, if any, may be suitably assumed.

			<b>UNIT - I</b>	<b>CO</b>	<b>PO</b>	<b>Marks</b>
<b>Important Note:</b> Completing your answers, compulsorily draw diagonal cross lines on the remaining blank pages. Revealing of identification, appeal to evaluator will be treated as malpractice.	1	a)	How does the chemical process in pyrosequencing contribute to nucleotide detection?	CO1	PO 1	<b>7</b>
		b)	What factors might contribute to the differences in read accuracy between Illumina and Ion Torrent sequencing?	CO1	PO 1	<b>7</b>
		c)	How would you assess the trade-offs between sequencing cost, speed, and accuracy across different NGS technologies?	CO1	PO 1	<b>6</b>
<b>OR</b>						
	2	a)	If a sequencing lab is constrained by budget but requires high accuracy, which sequencing method would be most suitable? Justify your choice.	CO1	PO5	<b>6</b>
		b)	Why do short-read sequencing technologies face challenges in genome assembly?	CO1	PO5	<b>6</b>
		c)	Design a flowchart comparing the workflow of three NGS techniques based on their core principles.	CO1	PO5	<b>8</b>
<b>UNIT - II</b>						
	3	a)	How would you adjust the assembly pipeline if your dataset contains a high number of sequencing errors?	CO1	PO5	<b>7</b>
		b)	What would be the consequences of ignoring sequence read correction methods in genome assembly?	CO1	PO5	<b>6</b>
		c)	What role do genome browsers play in analyzing large-scale sequencing data?	CO1	PO5	<b>7</b>
<b>OR</b>						
	4	a)	Why is De Novo Genome Assembly considered more challenging than reference-based assembly?	CO1	PO5	<b>7</b>
		b)	What are the advantages and limitations of various NGS data preprocessing methods?	CO1	PO5	<b>7</b>
		c)	Propose a strategy to improve genome assembly quality using current sequencing technologies.	CO1	PO5	<b>6</b>

<b>UNIT - III</b>					
5	a)	Why is genome assembly often compared to solving a complex puzzle?	CO2	PO2	<b>6</b>
	b)	How would you decide whether to use the Overlap Graph Approach or the De Bruijn Graph Approach for a given dataset?	CO2	PO2	<b>6</b>
	c)	If you were to design a new genome assembly algorithm, what key features would you include?	CO2	PO2	<b>8</b>
<b>OR</b>					
6	a)	What are the strengths and weaknesses of Greedy Algorithms in genome assembly?	CO2	PO2	<b>7</b>
	b)	What preprocessing steps would you recommend before applying the Overlap Layout Consensus (OLC) method?	CO2	PO2	<b>7</b>
	c)	Given an NGS dataset, what criteria would you use to determine the most suitable assembly algorithm?	CO2	PO2	<b>6</b>
<b>UNIT - IV</b>					
7	a)	How does RNA-seq contribute to biomarker discovery in cancer research?	CO3	PO2	<b>7</b>
	b)	If given an RNA-seq dataset from prostate cancer patients, how would you identify key differentially expressed genes?	CO3	PO2	<b>7</b>
	c)	Propose a study using NGS to investigate microRNAs in a specific type of cancer.	CO3	PO2	<b>6</b>
<b>OR</b>					
8	a)	How effective is targeted sequencing compared to whole-genome sequencing in identifying cancer mutations?	CO3	PO2	<b>7</b>
	b)	What are the implications of high-throughput RNA interference screens for personalized cancer therapy?	CO3	PO2	<b>8</b>
	c)	How does the integration of NGS with clinical oncology impact early cancer diagnosis and treatment?	CO3	PO2	<b>5</b>
<b>UNIT - V</b>					
9	a)	How does NGS facilitate the identification of polymorphisms in neuropsychiatric disorders?	CO4	PO1	<b>5</b>
	b)	What are the major challenges in using NGS for early disease diagnosis?	CO4	PO1	<b>6</b>
	c)	Develop a workflow for using NGS in personalized medicine for neuroinflammatory diseases.	CO4	PO1	<b>9</b>
<b>OR</b>					
10	a)	How would you use NGS to detect genetic variations linked to Alzheimer's disease?	CO4	PO1	<b>5</b>
	b)	How do different sequencing strategies compare in their ability to detect genetic markers for Parkinson's disease?	CO4	PO1	<b>6</b>
	c)	How can NGS contribute to precision medicine approaches in neurodegenerative disorders?	CO4	PO1	<b>9</b>

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