Name: **Enrolment No:**	

UPES

End Semester Examination, May 2025

Course: Precision Medicine & Wellness **Semester : VIII**
Program: B.Tech Biotechnology **Duration : 3 Hours**
Course Code: HSBT4014 **Max. Marks: 100**

Instructions: Read all questions carefully

S. No.	Section A Short answer questions/ MCQ/T&F (20Qx1.5M= 30 Marks)	Marks	COs
Q 1	Which 'omics' approach is primarily used to study variations in DNA sequences? A) Transcriptomics B) Proteomics C) Genomics D) Metabolomics	1.5	CO1
Q 2	Transcriptomics refers to the study of: A) DNA methylation B) Protein-protein interactions C) RNA transcripts D) Metabolic pathways	1.5	CO1
Q 3	Proteomics provides insights into: A) Mutation rates in DNA B) Expression levels of mRNA C) Structure and function of proteins D) Metabolite interactions	1.5	CO1
Q 4	Which 'omics' technique is most suited to measure metabolic changes in disease states? A) Genomics B) Metabolomics C) Transcriptomics D) Epigenomics	1.5	CO1

Q 5	Which of the following best defines a biomarker in the context of disease identification? A) A therapeutic drug B) A biological molecule indicating a disease state C) A DNA editing tool D) A vaccine component	1.5	CO2
Q 6	What is the primary goal of biomarker validation? A) To find new genes B) To confirm the reliability and accuracy of a biomarker in predicting disease C) To modify drug metabolism D) To replace genetic therapy	1.5	CO2
Q 7	Which of the following is NOT a typical use of disease biomarkers? A) Early diagnosis B) Patient stratification C) Determining planetary alignment D) Monitoring treatment response	1.5	CO2
Q 8	The Minimal Genome Project aims to: A) Sequence all genes of cancer cells B) Identify the largest known genome C) Determine the smallest set of genes essential for life D) Replace all human genes with synthetic versions	1.5	CO2
Q 9	Which of the following is a genetic variation? A) Nutritional deficiency B) Single nucleotide polymorphism (SNP) C) Exposure to radiation D) Smoking behavior	1.5	CO3
Q 10	Non-genetic variation can arise from which of the following? A) Frameshift mutations B) Inherited gene deletions C) Environmental exposure D) Chromosomal inversion	1.5	CO3
Q 11	A point mutation refers to: A) Insertion of multiple genes B) Substitution of a single base pair C) Duplication of an entire chromosome D) Mitochondrial gene recombination	1.5	CO3

Q 12	Which test is commonly used in prenatal genetic screening? A) Skin biopsy B) Amniocentesis C) EEG D) Colonoscopy	1.5	CO3
Q 13	Pharmacogenomic testing is primarily used to: A) Identify pathogens in infections B) Determine blood type C) Predict individual responses to medications D) Analyze vitamin deficiencies	1.5	CO4
Q 14	Tumor profiling typically includes the analysis of: A) Skin type and blood pressure B) Mutations in oncogenes and tumor suppressor genes C) Dietary intake D) Antibody levels	1.5	CO4
Q 15	What is a key ethical concern in using patient genetic data for clinical decisions? A) High costs of drugs B) Data privacy and informed consent C) Lack of insurance coverage D) Medical device calibration	1.5	CO4
Q 16	Which of the following criteria is *least* critical in validating a biomarker for clinical utility? A) Sensitivity and specificity B) Cost of mass spectrometry instrumentation C) Reproducibility across populations D) Biological relevance to disease mechanism	1.5	CO4
Q 17	What distinguishes a prognostic biomarker from a predictive biomarker? A) Prognostic biomarkers are only used in cancer; predictive are used elsewhere B) Prognostic biomarkers predict treatment response; predictive biomarkers predict disease outcome C) Prognostic biomarkers predict disease outcome regardless of treatment; predictive biomarkers indicate likely response to a therapy D) Both are interchangeable terms	1.5	CO5
Q 18	Which technique is considered gold standard for validating protein biomarkers at the translational level? A) ELISA B) Northern Blot C) Sanger Sequencing D) SNP microarray	1.5	CO5

Q 19	Which of the following was a direct implication of the Minimal Genome Project in synthetic biology? A) Discovery of BRCA1 gene B) Creation of the first eukaryotic artificial chromosome C) Synthesis of Mycoplasma mycoides with only essential genes D) Completion of the 1000 Genomes Project	1.5	CO5
Q 20	Which of these was *not* a key goal of the Human Genome Project (HGP)? A) Identify all the genes in human DNA B) Sequence the entire human genome C) Engineer synthetic chromosomes D) Improve tools for data analysis	1.5	CO5
Section B **(4Qx5M=20 Marks)**			
Q 1	Discuss the role of transcriptomics in identifying disease-specific gene expression patterns. How can transcriptomic data be used for diagnosis or treatment decisions?	5	CO1
Q 2	Evaluate how metabolomics can be used to detect metabolic changes in disease states. Write an example where metabolomics led to a novel disease insight or therapeutic strategy.	5	CO2
Q 3	Describe the objectives and significance of the Minimal Genome Project. How does identifying the smallest set of genes essential for life contribute to the field of synthetic biology and precision medicine?	5	CO3
Q 4	Outline the goals and findings of the Cancer Genome Project. How does the knowledge gained from this project aid in developing targeted therapies and personalized treatment plans for cancer patients?	5	CO3
Section C **(2Qx15M=30 Marks)**			
Q 1	**Case Scenario:** You are part of a research team working to develop a biomarker panel for early detection of triple-negative breast cancer (TNBC). Using data from the Human Genome Project and Cancer Genome Project, your team identifies a candidate biomarker. However, it requires further validation through proteomics and patient cohort studies.	15 (5+5+5)	CO2

	Question: a) Describe the step-by-step process for identifying and validating a biomarker in this context. b) Explain how the Human Genome Project and Cancer Genome Project have accelerated biomarker discovery in oncology. c) What potential role can the Minimal Genome Project play in refining biomarker-based diagnostics?		
Q 2	**Case Scenario:** A couple planning to start a family undergoes carrier screening and finds that both partners are carriers of a mutation in the CFTR gene, which causes cystic fibrosis. They opt for prenatal testing during pregnancy. After birth, the newborn is further screened using next-generation sequencing (NGS) for any additional undetected Mendelian conditions. **Question:** a) Describe the role of carrier testing, prenatal screening, and newborn screening in managing Mendelian diseases. b) Discuss ethical considerations that arise from such comprehensive genomic screening. c) How do advancements in sequencing technologies improve diagnostic accuracy and early interventions?	15 (5+5+5)	CO5
Section D **(2Qx10M=20 Marks)**			
Q 1	Discuss the role of genetic screening and diagnosis in personalized medicine. Include the following in your answer: a) Types of genetic screening (e.g., population-based, diagnostic, predictive) b) Benefits and limitations of genetic screening c) Ethical considerations involved in genetic diagnosis	10	CO2
Q 2	Discuss how genomic approaches contribute to understanding the genetic basis of complex diseases. Highlight the role of genome-wide association studies (GWAS), single nucleotide polymorphisms (SNPs), and next-generation sequencing (NGS) in disease diagnostics and risk prediction.	10	CO4