JAYPEE UNIVERSITY OF INFORMATION TECHNOLOGY, WAKNAGHAT

TEST -3 EXAMINATIONS- 2025

B.Tech VIII Semester (BT/BI)

COURSE CODE (CREDITS): 18B1WBT831 (3.0.0) MAX. MARKS: 35

COURSE NAME: Genetic Counselling

COURSE INSTRUCTORS: Dr. Tyson MAX. TIME: 2 Hours

Note: (a) All questions are compulsory.

(b) The candidate is allowed to make Suitable numeric assumptions wherever required for solving problems

Q.No	Question	CO	Marks
Q1	Discuss the difference between direct and non-directive genetic counseling in the context of Alzheimer's disease. Create a hypothetical scenario to explain your reasoning.	1	4
Q2	a) Huntington's disease is a neurodegenerative disorder with autosomal dominant inheritance." Justify this statement with a focus on genetic mechanisms and inheritance patterns. How does anticipation influence the disease's progression? b) As a genetic counselor, a woman (30 years) with a family history of Huntington's disease seeks advice before planning a pregnancy. Outline your counseling strategy and discuss the psychosocial support you would provide.	2+4	4+3
Q3	Compare the genetic predisposition to colorectal cancer between familial adenomatous polyposis (FAP) and Lynch syndrome (HNPCC). Describe the role of gene mutations in their development.	2	4
Q4	a) Retinoblastoma follows a two-hit hypothesis. Explain how RB1 gene mutations cause the condition and differentiate between hereditary and sporadic forms. b) As a genetic counselor, you are approached by a family with a history of retinoblastoma. The father had bilateral retinoblastoma, while the mother has no family history. How would you assess the inheritance risk for their future children?	2+4	4+4
Q5	Explain the pathophysiological mechanism of Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency. How does a deficiency in the G6PD	2	4

	enzyme lead to hemolytic anemia? Also, discuss the genetic basis of G6PD deficiency.		
Q6	a) In the quest to cure genetic disorders, gene therapy emerges as a beacon of hope. What distinguishes somatic gene therapy from germline modification, and why is the latter more controversial? b) CRISPR has been hailed as the ultimate tool for genomic correction. Describe how the CRISPER/Cas9 complex functions in editing DNA, and discuss a real-world application that showcases its potential.	3+3	4+4