JAYPEE UNIVERSITY OF INFORMATION TECHNOLOGY, WAKNAGHAT

TEST -2 EXAMINATIONS- 2025

B.Tech- VIII Semester (BT/BI)

COURSE CODE (CREDITS): 18B1WBT831 (3.0.0) MAX. MARKS: 25

COURSE NAME: Genetic Counselling

COURSE INSTRUCTORS: Dr. Tyson MAX. TIME: 1 Hour 30 Min

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	Genetic counseling helps individuals understand their genetic risks. a) What are the key steps involved in genetic counseling for a family with a history of Parkinson's disease? b) Discuss the ethical considerations associated with predictive genetic testing for neurodegenerative disorders.	1+4	2+3
Q2	Muscular dystrophies disorders often have a genetic basis. a) Explain the molecular mechanisms underlying Duchenne Muscular Dystrophy (DMD) and how mutations in the respective gene lead to disease progression. b) Myotonica Dystrophy (DM) is an example of a Trinucleotide repeat expansion disorder. Explain how repeat expansions in the desired gene contribute to disease pathology and how the number of repeats influences disease severity.	2	3+2
Q3	Prenatal screening and diagnostic tests help in detecting genetic disorders before birth. a) Differentiate between Chorionic Villus Sampling (CVS) and Amniocentesis in terms of procedure, timing, and risks. b) What is quad screening, and how does it help in the detection of chromosomal abnormalities such as Down syndrome?	4	3+2

Q4	A couple planning for a child consults a genetic counselor after the woman is found to be a carrier of Cystic Fibrosis (CF) during routine screening. The counselor recommends genetic testing for her partner, despite no known family history of CF. (A) Why is it necessary to test the partner, and how does the inheritance pattern of CF influence the risk of the child being affected? (B) CF is caused by specific genetic mutations that lead to defects in a key protein. Explain the most common genetic mutation associated with CF, the pathophysiological defects it causes, and the diagnostic and treatment approaches used for managing the disease.	1+2	2+3
Q5	A 12-year-old boy presents with progressive muscle weakness, joint contractures (especially in the elbows and Achilles tendons), and difficulty bending his arms and legs. His family reports that his father also had similar muscle weakness and developed cardiac issues in adulthood. Based on the symptoms and family history, which muscular dystrophy is the most likely diagnosis, and what is the underlying genetic cause of this condition?	2	5