JAYPEE UNIVERSITY OF INFORMATION TECHNOLOGY, WAKNAGHAT

TEST 3 EXAMINATION - 2024

B.Tech.-VIII Semester (BT)

COURSE CODE (CREDITS): 18B1WBT831 (3-0-0) MAX. MARKS: 35

COURSE NAME: Genetic Counselling

COURSE INSTRUCTORS: Prof. Sudhir Kumar MAX. TIME: 2 Hours

Note: (a) All questions are compulsory.

(b) Marks are indicated against each question in square brackets.

(c) The candidate is allowed to make suitable numeric assumptions wherever required for solving problems.

1. a). Which type of gene therapy is right for treating specific disease?
   b). Is gene therapy safe over long term? [CO-3] [2.5+2.5]

2. Prepare a case study of any genetic disease and discuss what is Directive and Non-Directive Genetic Counseling in relation to it. [CO-4] [5]

3. a). What are the goals of Pre natal diagnosis?
   b). If 50% of the children of a couple (boys as well as girls) are suffering from color blindness, predict genotypes of the parents giving reasons. [CO-1] [2.5+2.5]

4. How do genetic mutations in diseases such as Duchenne Muscular Dystrophy (DMD), Emery-Dreifuss Muscular Dystrophy (EDMD), Myotonica Dystrophy and Limb-Girdle Muscular Dystrophy (LGMD) influence the approach to genetic counseling, and what specific considerations must be taken into account for effective diagnosis and management in patients with potential combined disease genetic mutations? [CO-4] [4]

5. Describe the two-hit hypothesis as it relates to the development of retinoblastoma. How do different mutations of gene contribute to the onset of this cancer? Discuss the molecular mechanisms by which the loss of tumor suppressor gene in retinoblastoma function leads to uncontrolled cell proliferation. [CO-2] [4]

6. Discuss the pathophysiological mechanisms by which different genetic mutations in the G6PD gene lead to varying degrees of enzyme deficiency and clinical severity in G6PD deficiency. Additionally, describe the evolutionary advantage of G6PD deficiency individuals. [CO-2] [4]

7. Colorectal cancer (CRC) is known to progress through a series of well-defined genetic alterations. Describe the adenoma-carcinoma sequence in the context of colorectal cancer, detailing the key genetic mutations involved at each stage. [CO-2] [4]

8. Explain the role of mutations in the SNCA, LRRK2, and PARK2 genes in the development of Parkinson's disease (PD). How do these mutations contribute to the pathophysiology of PD? Describe the genetic counseling process for families with a history of Parkinson's disease, including risk assessment, the implications of genetic testing, and the management of psychological and familial concerns. [CO-4] [4]